

What is BabySEQ?

BabySEQ is a Non-Invasive Prenatal Testing (NIPT) method, a new-generation screening that detects not only chromosomal disorders such as Down syndrome, Trisomy 13, and Trisomy 18, but also microdeletion syndromes in the fetus, without the need for any invasive procedure that may involve risk. Currently, NIPT-based tests like BabySEQ are known to be the most accurate tests for screening these conditions. BabySEQ, which is performed in our center, has become an increasingly preferred screening method in recent years due to its non-invasive nature and ease of application. BabySEQ includes four different panels, depending on your preference.

	BabySEQ BASIC PANEL	BabySEQ EXTENDED PANEL	BabySEQ RARE*	BabySEQ XL*
Common Aneuploidies (T21, T18, T13, X/Y)	✓	✓	✓	✓
All Other Aneuploidies	✓	✓	✓	✓
Deletion and Duplication Syndromes		✓	✓	✓
Cystic Fibrosis + SMA + Fragile X Carrier Screening			✓	✓
Carrier Screening for 700 Diseases				✓

For these tests, it is recommended to collect one tube of blood from the father (purple-top EDTA tube) and one tube of blood from the mother using the BabySEQ blood collection tube provided by Intergen.

What is BabySEQ RARE?

In addition to the extended BabySEQ panel, the BabySEQ RARE package also screens for SMA, Cystic Fibrosis, and Fragile X Syndrome. Scan the QR code to view the BabySEQ RARE Information Brochure.



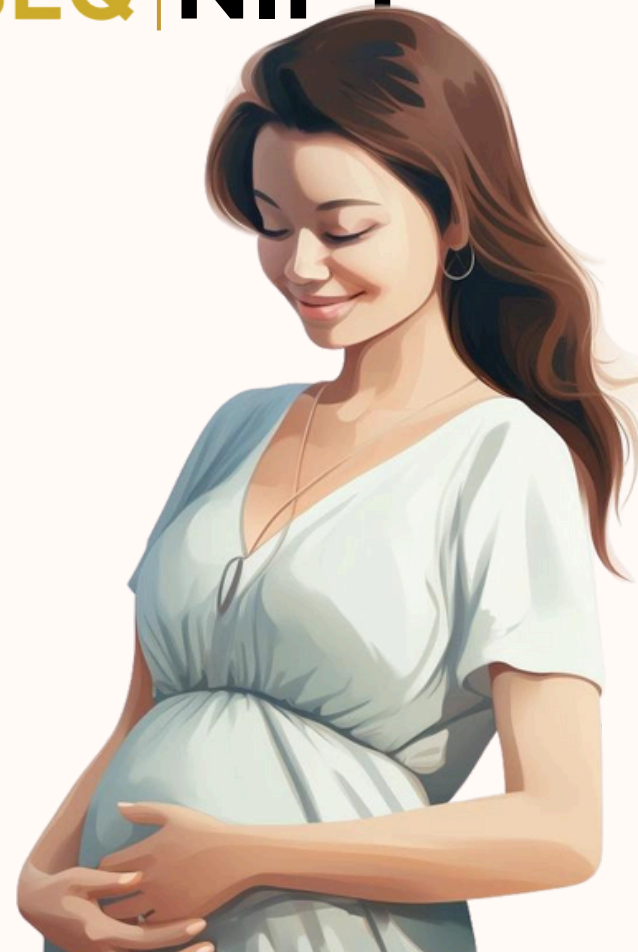
In the BabySEQ Rare package, carrier testing is also performed for the father.



In the BabySEQ XL package, carrier testing is also performed for the father.

For detailed information about the BabySEQ Rare and XL packages, please consult your physician.

babySEQ | NIPT



What is Being Tested?

The most common autosomal and gonosomal aneuploidies are shown below. **The Basic Panel** screens for all chromosomal aneuploidies.

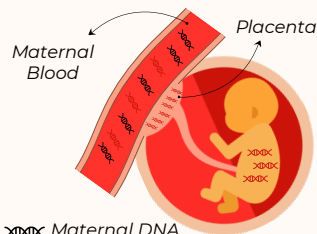
Aneuploidy		
Trisomy 21 (Down Syndrome)	Trisomy 7	Trisomy 15
Trisomy 18 (Edwards Syndrome)	Trisomy 8	Trisomy 16
Trisomy 13 (Patau Syndrome)	Trisomy 9	Trisomy 17
45, X (Turner Syndrome)	Trisomy 10	Trisomy 19
47, XXY (Klinefelter Syndrome)	Trisomy 11	Trisomy 20
47, XXX	Trisomy 12	Trisomy 22
	Trisomy 14	Other
		Aneuploidies

In the Extended Panel, in addition to all chromosomal aneuploidies, deletion/duplication syndromes are also screened.

For screening purposes, deletion/duplication syndromes ≥ 7 Mb in size are evaluated.

The list of deletion/duplication syndromes screened is shown on the right.

Note: Apart from the changes defined in the table, if any alteration greater than 7 Mb is detected, it will be indicated in the report.



X X X Maternal DNA
X X X Cell-Free Fetal DNA

During pregnancy, fragments of fetal DNA, predominantly originating from the placenta, pass into the mother's circulation. After the 10th week of pregnancy, with the BabySEQ NIPT method, we can analyze this fetal DNA present in the maternal bloodstream.

Deletion/Duplication Syndromes

<p>1p32-p31 Deletion Syndrome 1q41-q42 Deletion Syndrome 1q43-q44 Deletion Syndrome 2p12-p11.2 Deletion Syndrome 2p15-p16.1 Deletion Syndrome 2q13 Deletion Syndrome 2q13 Duplication Syndrome 2q31.1 Microdeletion Syndrome 2q31.1 Duplication Syndrome 2q35 Duplication Syndrome 3p25.3 Deletion Syndrome 3pter-p25 Deletion Syndrome 3q13.31 Deletion Syndrome Dandy-Walker Syndrome (3q Deletion/Duplication) 3q26 Microduplication Syndrome 3q29 Deletion Syndrome 4q21 Deletion Syndrome Axenfeld-Rieger Syndrome, Type L (4q25) 5p13 Duplication Syndrome 5q12 Deletion Syndrome 5q14.3 Deletion (Proximal) Syndrome Sotos Syndrome (5q35 Deletion) 6p22 Microdeletion Syndrome 6q11-q14 Deletion Syndrome 6q24-q25 Deletion Syndrome Coffin-Siris Syndrome 1 (6q25) Greig Cephalopolysyndactyly Syndrome (7p14) 7p22.1 Microduplication Syndrome 7q11.23 Deletion (Distal) Syndrome Williams-Beuren Syndrome (WBS) (7q11.23) Currarino Syndrome (7q36) 7q36.3 Duplication Syndrome 8p11.2 Deletion Syndrome 8p23.1 Deletion Syndrome 8q12 Microduplication Syndrome Nablus Mask-Like Facial Syndrome (NMLFS) (8q22.1)</p>	<p>Trichorhinophalangeal Syndrome, Type 2 (TRPS2) (8q23.3-q24.11) 9p Deletion Syndrome 9p13 Microdeletion Syndrome 9p24.3 Deletion Syndrome 9q33.33-q34.11 Microdeletion Syndrome Kleefstra Syndrome 1 (KLEFS1) (9q34.3) 10p11.21-p12.31 Microdeletion Syndrome DiGeorge Syndrome – Velocardiofacial Syndrome Complex 2 (DSG2) (10p14) 10q22.3-q23.2 Deletion Syndrome Split Hand/Foot Malformation 3 (SHFM3) (10q24) 10q26 Deletion Syndrome Potocki-Shaffer Syndrome (11p12-p11.2) WAGR Syndrome (11p13) WAGRO Syndrome (11p13-p14) 11q13.2-q13.4 Deletion Syndrome 11q22.2-q22.3 Microdeletion Syndrome 11q23 Deletion Syndrome Distal Trisomy 11q Syndrome 12p12.1 Microdeletion Syndrome 12q14 Microdeletion Syndrome 12q15-q21.1 Microdeletion Syndrome 13q14 Deletion Syndrome 14q11-q22 Deletion Syndrome Frias Syndrome (14q22-q23) 14q24.1-q24.3 Microdeletion Syndrome Prader-Willi Syndrome (15q11-q13) 15q13.3 Deletion Syndrome (BP4/BP5) 15q14 Microdeletion Syndrome 15q25.2 Deletion (Proximal) Syndrome 15q26-qter Deletion Syndrome 16p11.2-p12.2 Microdeletion Syndrome 16p12.2 Deletion (Proximal) Syndrome 16p13.11 Duplication Syndrome 16p13.11 Deletion Syndrome Rubinstein-Taybi Syndrome (16p13.3) Alpha Thalassemia-Intellectual Disability Syndrome linked to Chromosome 16 (ATR-16 Syndrome) (16p13.3)</p>	<p>16q22 Deletion Syndrome Smith-Magenis Syndrome (17p11.2) Yuan-Harel-Lupski Syndrome (YUHAL) (17p12-p11.2) 17p12 Deletion Syndrome 17p12 Duplication Syndrome 17p13.1 Deletion Syndrome Miller-Dieker Lissencephaly Syndrome (MDLS) (17p13.3) (Deletion) Miller-Dieker Lissencephaly Syndrome (MDLS) (17p13.3) (Duplication) 17p13.3 Telomeric Duplication Syndrome 17q12 Deletion Syndrome 17q21.31 Deletion Syndrome 17q23.1-q23.2 Deletion Syndrome Tetrasomy 18p Syndrome 18p Deletion Syndrome 18q Deletion Syndrome 19p13 Duplication Syndrome 19q13.11 Microdeletion Syndrome 20p13 Microdeletion Syndrome 21q22.11-q22.12 Microdeletion Syndrome DiGeorge Syndrome – Velocardiofacial Syndrome (22q11.2) (Proximal) 22q11.2 Deletion Syndrome (Distal) 22q13 Deletion Syndrome 22q13 Duplication Syndrome Xp11.22 Duplication Syndrome Xp11.22-p11.23 Duplication Syndrome Xp11.23 Microdeletion Syndrome Xp11.3 Deletion Syndrome Xp21 Microdeletion Syndrome Xp21.2 Microduplication Syndrome Xp22.31 Microdeletion Syndrome Xq21 Microdeletion Syndrome Xq22.3 Telomeric Deletion Syndrome Xq27.3-q28 Duplication Syndrome Xq28 Deletion Syndrome</p>
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