PREIMPLANTATION GENETIC TESTING (PGT)
TEST INFORMATION FORM

The purpose of this informed consent text is to inform you about PGT tests and to obtain your consent. Providing this information before we deliver healthcare services is an obligation for us and a right for you. Please read the text carefully. If you have any questions, do not hesitate to contact us through the specified communication channels.

Patient Full Name:

ID No:

1- What is PGT, When is it Performed?

Preimplantation Genetic Testing (PGT) is a test performed during in vitro fertilization (IVF) treatment in embryos to detect "single-gene disorders known in the family" and/or "chromosomal abnormalities." PGT enables the selection of genetically healthy embryos before embryo transfer. Different PGT tests are specifically developed for each family according to the genetic risks in the family:

PGT Setup: The PGT Setup is the process of establishing the infrastructure required for genetic analysis in the laboratory and designing a patient-specific test. These are studies that enable us to distinguish on which of the chromosome pairs the genetic alteration present in the family is located. For this reason, sometimes it may be necessary to take samples from the mother and father of the spouses, and sometimes from other affected individuals in the family. Without these studies, it cannot be distinguished whether the genetic alteration is on the chromosome inherited from the mother or on the chromosome inherited from the father, and therefore the selection of a healthy embryo in the embryos cannot be performed. During this process:

- Preparations are made for the analysis of genetic diseases by taking the patient and family history into consideration.
- Relevant genetic analysis methods are determined.
- For monogenic diseases, the test is specifically designed.

The PGT Setup is a critical step to ensure that tests provide accurate results and that patient-specific risks are managed effectively.

PGT-M (For the Diagnosis of Monogenic Disease): PGT-M is performed to detect the presence of this disease in embryos in individuals who have a genetic disease in their family history. For this test, the family is first interviewed and the risks in the family are determined. Especially for families in which there is consanguinity between the spouses, but also for all families, information is provided about additional tests that can be performed to evaluate whether they have risks for another disease other than the disease that brought them to us. For families who do not want these additional tests, preliminary studies are carried out for the genetic disease reported to us. A study strategy specific to that family is created. While performing the PGT-M study, both the variant that is the cause of the disease present in the family is examined, and markers close to this gene are determined to increase the reliability of the test. In order to determine these markers, it is necessary to perform the "setup" study, the details of which are given above.

PGT-A (For Aneuploidy Screening): PGT-A is used to detect numerical chromosomal anomalies in embryos (for example, the presence of an extra chromosome such as Trisomy 21 – Down syndrome, or the absence of one/several chromosomes). Aneuploidy is a genetic alteration that may cause pregnancy losses, incompatibility of the embryo with life, or serious health problems at birth.

This method is also used when one of the spouses has a Robertsonian translocation. It is evaluated whether the number of chromosomes is numerically normal. However, in this case, additionally, we also recommend performing a "uniparental disomy" test for these two chromosomes in order to distinguish embryos that started abnormally and then were rescued, and to test, for the chromosomes involved in the translocation, whether one is inherited from the mother and the other from the father. In order to perform this test from embryos, a preparatory study (Setup study) is required. It is not known exactly at which stage of embryo development the rescue stage from trisomy or monosomy occurs. Therefore, even though the probability is low, performing this test is recommended. For families who do not want to have this test during IVF, if pregnancy occurs, prenatal diagnosis (from amniocentesis or chorionic villus sampling) is definitely recommended, and UPD study during this process is also definitely recommended.

□ I want uniparental disomy testing to be performed on the embryos as well.

□ I do not want uniparental disomy testing to be performed on the embryos. I only want the number of chromosomes to be evaluated.

PGT-SR (Structural Rearrangement Study): This is a test performed to evaluate small losses and gains that may occur in embryos in individuals who are balanced translocation carriers or inversion carriers. This test is primarily recommended if there is a structural chromosomal change in one of the parents.

Combined PGT (Monogenic Disease and Aneuploidy Screening): Both chromosomal anomalies and monogenic disease(s) creating risk in the family are screened. Chromosomal disorders can always occur in IVF studies. Some chromosomal changes that are spontaneously eliminated in naturally conceived pregnancies may be present in embryos in IVF pregnancies. If there is a single-gene disease risk in the family, since the risk of this is higher, embryos are first screened for this disease (PGT-M). Aneuploidy testing (PGT-A) is applied to embryos that do not carry the single-gene disease, that is, embryos with a normal PGT-M result.

PGT-HLA (For HLA Typing): It enables the selection of an embryo with a tissue type suitable for transplantation. It is used especially for cases where sibling-to-sibling transplantation is considered, that is, for the selection of an embryo that is tissue-compatible with the affected child already present at home.

PGT-M-HLA: This is a special test that enables both monogenic disease(s) creating risk in the family and HLA typing to be performed at the same time. In this test, PGT-M study is performed for the disease present in the family, and then, among the embryos that do not carry this disease, selection of the embryo with compatible tissue is performed.

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2- What Do the Test Results Mean, Is There a Margin of Error, Can It Be Considered Completely Reliable?

PGT tests are used "to reduce the family's risk of having an affected child." The analysis is performed using DNA obtained from a very small number of cells taken from the embryo. There is a margin of error. In particular, in Day 3 (cleavage stage) biopsies, 1 (rarely 2) cell(s) are taken; in Day 4 biopsies (it is difficult to take; there are compact cells—morula stage), 5–10 cells are taken; and in Day 5 (blastocyst stage) biopsies, 5–10 trophectoderm cells are taken. Because reliability is low, our center does not perform analysis from Day 3 and Day 4 biopsies. As a genetic result, the most reliable method is Day 5 biopsy. Nevertheless, the biopsy obtained may not represent the entire embryo, especially in terms of chromosomal disorders. The interpretation of the results depends on the individual situation and the methods used in genetic analysis. The meanings of the results are as follows:

- **Suitable:** Indicates that the embryo is suitable for transfer in terms of the genetic problems examined. This result does not mean that the embryo is completely genetically healthy. However, it greatly reduces the risk of disease. The result obtained may not represent the entire embryo. Since the test is performed from a very small amount of DNA, it may be misleading. For this reason, prenatal diagnosis is recommended if pregnancy occurs. This result does not show other genetic diseases or newly occurring mutations other than the diseases studied. For autosomal recessive diseases, the result of embryos that are "carriers" is stated as "heterozygous" in the report. In embryos carrying a "heterozygous" variant for recessive diseases, no clinical finding is expected. However, although rarely, in some cases and in some variants, mild clinical findings may occur. For these changes, the spouses are already carriers. For autosomal dominant diseases, it is evaluated whether the genetic variant present in one of the spouses (sometimes it may be in both spouses) is present in the embryo. Whether or not the spouse/spouses carrying the variant have clinical findings, the embryo may be affected in this group of diseases. Therefore, the probability that embryos carrying this variant will be affected varies depending on the level at which the variant affects the protein and which gene it is in. For this reason, detailed information is written in the report according to the penetrance of the gene and the characteristics of the variant. Some genes may be associated with diseases inherited both recessively and dominantly. In this case as well, the genetic results of the spouses, and the gene and variant are evaluated in detail and the risks are stated in the report. In X-linked diseases, the mother is a carrier. In 50% of male embryos, there is a risk of disease associated with this variant. An evaluation is made by considering both this variant and the sex. Clinical findings may sometimes be observed in women carrying X-linked diseases as well. A detailed note is also written in the report on this matter. Detailed information for mitochondrial diseases is given in section 4.2.
- **Not suitable:** Indicates that the embryo is not suitable for transfer in terms of the genetic problems examined. It indicates the presence of the genetic variant studied in the embryo in relation to the genetic problems tested. In this case, your physician will evaluate the situation with you in detail and will present alternative options.
- **Amplification could not be obtained:** Means that no result could be obtained due to insufficient genetic material or technical reasons. In this case, the test may need to be repeated. If "re-biopsy" can be performed, i.e., if a biopsy can be taken again from the embryo, the test is repeated free of charge. Sometimes this may not be possible. (The embryologist and clinician decide whether re-biopsy can be performed.) In such a case, transfer of this embryo is not recommended.
- **Please read the explanation:** It is used in situations where "additional information regarding embryo transfer" is required as a result of the test performed for you. This special situation will be explained to you and the clinician, and how to proceed with the procedure will be determined with your and your clinician's opinion.

3- What Other Information Do I Need to Know About the Test?

3.1. The time given for the tests is approximate; results may be obtained in a shorter or longer time.

3.2. Your remaining DNA samples after the test may be stored indefinitely to be used for test validation or training. After completion of the test requested above, you may apply to the laboratory and request destruction of your blood and DNA sample. You may also specify this in the approval section of this form. Not allowing your sample to be used for test verification or training does not affect your test result.

3.3. The changes detected as a result of the test may sometimes need to be supported with additional "diagnostic tests." In this case, you will be contacted and informed about these tests. If you do not have the recommended additional tests performed, diagnostic success may decrease. Additional payments may be required for those with high costs.

3.4. Some or all of the tests may be sent to another center if deemed medically necessary.

3.5. Rarely, you may need to send or provide a second material/sample. If results cannot be obtained, the test may be repeated.

3.6. If you do not provide your sample at our center and it arrives by cargo or courier, you will be informed by message that the sample has reached our center. Payment information is also provided in the message. After you make the payment to the account information provided to you and share the receipt, your procedures are initiated. During this time, sample security is ensured at our center.

3.7. If you withdraw from the test after making the payment, depending on the stage of the test, we may refund the remaining part other than the expenses incurred up to that point; however, if a significant part of the test has been completed, it is not possible for us to issue a refund.

3.8. Some data obtained from the tests may not be reported for scientific or ethical reasons.

3.9. Your test results will be stored in Intergen records.

3.10. If your material was not collected by Intergen, Intergen is not responsible for whether the material was collected correctly, the suitability of the material, or the information provided in other centers.

3.11. If our physicians evaluate that a test other than the test requested by your physician is more suitable for establishing a diagnosis, in order to protect your benefit as a patient and to prevent you from having to provide a sample again, you and/or your physician will be informed and the test considered appropriate will be recommended. Our center reserves the right to refuse tests that are not medically appropriate to perform.

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3.12. Upon your request, your test result may be sent to you via communication methods such as WhatsApp, Gmail, Hotmail, etc. However, we would like to inform you that such applications are of foreign origin, which means that the message sent reaches the servers of foreign-origin companies (their servers), that is, it is transferred abroad. Ensuring the confidentiality of the data by the companies owning the application is not under the control of our center, and you can access their privacy policies from their own websites.

3.13. As a licensed healthcare institution, in order to perform genetic testing, we are required to obtain your identity, contact, health, and genetic data. In accordance with our obligation of confidentiality, we protect these data and ensure their privacy. Unless there is your consent or a legal obligation, we do not share your data with third parties/institutions. If you give consent, provided that your personal data are anonymized, and without prejudice to your right to privacy and personal rights, we may use your data for scientific purposes in the studies we conduct. You have the right to learn whether your personal data are processed, to request information regarding your personal data if they have been processed, to learn the purpose of processing of your personal data and whether they are used in accordance with that purpose, to know the third parties to whom your personal data are transferred domestically or abroad, to request correction of your personal data if they are processed incompletely or incorrectly, to request deletion or destruction of your personal data and to request notification of such actions to third parties to whom personal data have been transferred, to object to the emergence of a result against you by analyzing the processed data exclusively through automated systems, and to request compensation for damages in case you suffer damage due to unlawful processing of your personal data. We will respond to your applications within 30 days.

4- Criteria for Inclusion of Genetic Variants in the Study (Genetic Variants Included in the PGT-M):

4.1. Single-gene diseases and changes reported as VUS:

The results obtained and the changes detected as a result of genetic tests performed in our center are evaluated, as in the whole world, according to ACMG standards and guidelines (American College of Medical Genetics and Genomics – ACMG Standards and Guidelines). According to this guideline, changes are classified as pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign. Changes classified as pathogenic and likely pathogenic are included in the study to be performed because their association with disease is very strong. If a variant that we consider not to include in the study is detected in you, you will be informed about this and the reasons.

Changes evaluated as VUS have now begun to be divided worldwide into strong and weak. Especially strong VUS are more likely to be clinically related. It is not possible to state that a change evaluated as a "strong VUS" has "no clinical significance." However, after very extensive functional studies, broad segregation studies, etc., these interpretations may change. Especially for autosomal dominant diseases (diseases that show clinical findings when a variant is present in one of the two existing genes), although one of the spouses carries this variant, that person may not show disease findings. In this case, interpretation may require evaluation of family information, whether there are patients in the family related to this gene, sometimes family screening to be performed, and clinical evaluation of the person carrying the variant. If these additional tests are recommended to you and you do not have them performed, the reliability of the test decreases. In light of this information, you will be informed about the changes detected as VUS, and with your approval, it will be decided whether they will be included in the PGT.

Results of the risk assessment studies performed for you:

Gene name/Associated disease/Inheritance	Classification according to ACMG criteria	I received genetic counseling (Write "I received" or put a "+" sign)	Please indicate whether you want the test to be performed for the gene or not.

4.2. The mother carrying a mosaic disease mutation in mitochondrial DNA:

Mitochondrial genes are transmitted only from the mother; therefore, all children in the family inherit the same mutation from the mother. When a mutation is detected at a certain level rather than in all cells in the mother, this change is referred to as "heteroplasmic." In mitochondrial diseases, in the presence of heteroplasmy, the clinical course may vary greatly among individuals. In each individual, normal and mutant mtDNA are present together as a mixture. The ratios measured in blood may not represent all tissues. For this reason, clinical variability (clinical variabilite) that is not consistent with the ratios detected in blood may be observed. The same applies to embryos. A low mutation ratio detected in the examined cells will reduce the risk; however, it will not be possible to make a definitive clinical interpretation. Sometimes, even when the variant ratio is high, clinical findings may not develop. In the PGT-M study we will recommend to you, the aim is to reduce the risk by selecting an embryo with a low mutant mtDNA ratio. It is not possible to eliminate the risk completely. Many literature data on this subject have also begun to emerge.

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İNTERGEN Genetics and Rare Diseases Diagnosis Center

CONSENT FORM

1- Genetic Test Consent Form

(To be completed by the patient.)

"I have read the information form about the genetic test. I have been verbally informed about the test, my questions have been answered, and I have been given a reasonable amount of time to read the written information. I understand the purpose of the test, its results, its reliability, the reporting period, and the processes of processing my personal data."

Dear Patient; in accordance with the statement above, if you consent to the genetic test being performed on your material under the conditions specified in the information form, please write **"I consent to the genetic test being conducted."** in the space below:

(Since any intervention in the field of healthcare can only be performed after obtaining informed consent, it will not be possible for us to perform the test if you do not give your consent.)

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2- Additional Test Consent Form If Medically Necessary

(To be completed by the patient.)

Dear Patient; as explained in the information form, in genetic studies, if it is deemed medically necessary in order to establish a diagnosis, additional tests that may assist in diagnosis may be performed, or during the process we may evaluate performing a test other than the one initially specified. In addition, secondary findings that may arise as a result of the tests, apart from the findings that led to your application, will be reported if deemed medically necessary. If you accept this, please mark the box below stating "I accept." If you do not accept the performance of additional tests, please mark the box below stating "I do not accept." We would like to state that if you do not have the recommended additional tests performed, diagnostic success may decrease. If the additional tests are subject to a fee, our team will inform you. □ ***I accept.*** □ ***I do not accept.***

3- Scientific Research Consent Form

(To be completed by the patient.)

Dear Patient; as Intergen Genetic Diseases Evaluation Center, in order to protect and improve public health, we may conduct scientific studies by anonymizing the health and genetic data under our responsibility. In these studies, we take all necessary administrative and technical measures to ensure privacy and the protection of personal data. Under these conditions, please indicate whether you consent to the use of the data you have shared with us in scientific studies by marking one of the boxes below: □ **I give my consent.** □ **I do not give my consent.**

4- Consent Form for Sending Test Results

(To be completed by the patient.)

Dear Patient; please mark the box for the method you want us to use when sending your test results to you. If there is any change in the information, please inform us as soon as possible. If you do not want your report to be shared with your doctor/the referring institution, please indicate this below. In communications established with us via foreign-origin applications such as WhatsApp, Gmail, and similar applications whose servers are located abroad, we provide the information below in order to inform you about the processing of your personal data.

When a message or email is sent to our center via foreign-origin applications, as a healthcare institution under an obligation of confidentiality, we protect all personal data of yours—of a general and special nature—such as identity, contact, health, and genetic data, that may be obtained through that message or email, and we do not share them with third parties unless there is a legal obligation. However, sending a message or email through these applications means sharing the general and special categories of personal data contained in the message or email not only with our Center, but also with the commercial company in the country that developed the application, i.e., transferring your data abroad. In this case, ensuring the confidentiality of the data obtained by the applications is not under the control of our Center.

□ **Send to my phone via a messaging application. Application to be used:** □ **WhatsApp** □ **Other:**

Phone number to which the test report will be sent:

(For foreign-origin messaging applications, transfer of data abroad may occur.)

□ **Send to my email address. Email address:**

(For foreign-origin emails, transfer of data abroad may occur.)

Do you approve that my test results be sent to the doctor or institution that referred you to us, as you indicated, and that your medical results be shared with your doctor?

□ **I give my consent** □ **I do not give my consent**

I approve that my test results be shared with the following persons:

Full Name: Relationship:

PATIENT	ON BEHALF OF INTERGEN
Full Name:	Full Name:
Date:	Date:
Signature:	Signature:

The above Information Form and Explicit Consent Form have been signed between Intergen and the Patient in order to determine the terms and conditions of the service to be provided by Intergen, to inform the Patient accordingly, and to obtain the Patient's consent.

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