

**INFORMATION****Preimplantation Genetic Diagnosis for Single Gene Diseases and/or HLA Typing (PGD)**

Single gene disorders; are hereditary diseases resulting from a mutation or alteration formed on a specific gene of the patient's DNA. Examples of single gene disorders include diseases such as Beta Thalassemia, Alfa Thalassemia, Cystic Fibrosis, Sickle Cell Anemia, Spinal Muscular Atrophy (SMA) and Wiscott Aldrich Syndrome. For People with a family history of single-gene disease, this disease is more likely to be inherited to children.

With Preimplantation Genetic Diagnosis, genetically healthy or affected embryos can be distinguished. Nowadays, PGD is the most commonly used method when determining whether an embryo before pregnancy is genetically healthy. Before PGD is performed, officials must first determine if a DNA test to determine your genetic condition is possible. Only if such a test can be developed, embryos can be obtained which there is no risk of a specific genetic abnormality.

Before performing the PGD procedure, a study called "SETUP process" should be performed with the DNA obtained from you and your relatives. In this process, a personal test method for the disease and mutation you carry will be developed and this method will be used during the PGD procedure. This process may last about 6-8 weeks. In some rare cases this period can be prolonged. This work will be organized by Acibadem Labgen using genetic testing methods to identify possible single gene disorders and HLA typing in your eggs and/or embryo. It is limited to genetic testing with the aim of determining whether it is possible to design a reliable molecular analysis technique.

In the PGD study, researchers will also decide whether this method is right for you. At the conclusion of the study you will be asked to confirm whether or not you have approved PGD studies by signing the acceptance form which explained the risks, benefits, PGD and Embryo Transfer (ET). We will only be able to participate in PGD work if you give it your approval.

**Polar Body Biopsy**

The maturing egg produces a small cell called the "First Polar Body". The egg then produces the "Second Polar Body" following the fertilization. Since polar bodies carry the genetic information found in the egg, tests on these cells can be used to obtain information about the genetic structure of the egg. Polar bodies which have a role in development of the embryo are pulled out of a hole formed in the outer layer of the egg. Then the egg is taken to fertilization process. Polar bodies are used for genetic diseases or risk status of the mother, and they do not give information about disease or carrier status from the father.

**Blastomer Cell Analysis**

After approximately 68-72 hours following fertilization and at least 6-8 embryo cells stage, this method is performed by taking one of the cells called blastomer on the 3rd day of embryonic development. Blastomer biopsy is used for the detection of single gene disorders that can result from both the mother and the father.

**Trophododerm Tissue Analysis**

Trophectoderm tissue analysis is performed in the embryo on the blastocyst stage. It is usually done by taking 4-5 cells on the 5th day after fertilization. With this method, single gene disorders can be detected in the embryo, which can inherit both from mother and father.

**Problems that can be encountered on Preimplantation Genetic Diagnosis of Single Gene Diseases**

All patients can encounter problems during ART Practice. In short, in some cases, the patient does not respond to the drug treatment and the treatment cycle can be canceled before the eggs are collected. Scarcely, there is no egg development. In addition, there may be cases where no egg is fertilized and embryos to be subjected to genetic testing can not be obtained.

If the disease being examined is a dominant disease, 50% of the embryos, 25% of the embryos if it is a recessive disease, and 18% of the embryos if it is a recessively inherited disease are expected to be suitable for transfer. Therefore, when post biopsy genetic identification is performed, it can be determined that the all of the embryos are affected or HLA incompatible with the patient. In these cases the transfer will not take place because there will not be a suitable embryo for the transfer.

As a result of genetic tests, it can be determined that some embryos are carriers in terms of disease similar to mother and father. The patient should know before the start of treatment that when HLA match is detected only in carrier embryos and transfer of these embryos is decided, if the child to be born is married to a carrier for the same disease again in the future, there is a risk of transmitting the disease to the child.

Research shows that embryos obtained from routine ICSI patients may sometimes contain abnormal chromosomes. We also know that as the woman's age progresses, her risk of having a baby with a chromosomally abnormal chromosomal abnormality, such as Down Syndrome, increases. For this patients, the risk of miscarriage due to chromosomal abnormalities is also increasing. Because preimplantation genetic diagnosis is a new technique and there is a risk of misdiagnosis about 1%, we recommend Prenatal Diagnosis if the patient is pregnant.

LG-MG-FR-005

**INFORMATION AND CONSENT FORM FOR PREIMPLANTATION GENETIC  
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If technically possible, screening may be done for some chromosomal abnormalities in the embryo, other than the target disease (e.g. Thalassemia). If this is not possible, however, it should be noted that only the target disease will be screened and therefore the fetus will not be examined for another disease (e.g. Down Syndrome).

**Freezing Untransferred Embryos**

After the genetic tests, non-HLA-matched healthy embryos can be frozen by taking approvals of the couple. However, in cases where only mutation analysis is performed, healthy embryos that are not transferred can also be frozen. The frozen embryos can be thawed on request for a new transfer and transfer it to the mother.

If you have read and understood this form written to inform you, if you understand and accept that the Preimplantation Genetic Diagnosis (PGD) application is necessary for the health of your probable baby, please write your name and surname in the following spaces with your own handwriting.

Participation in this process may allow us to obtain new and available information on other couples and/or medical sciences. Therefore, under the guarantee that our identity will not be disclosed, we allow making photographic and/or audiovisual recording and publishing of laboratory procedures related to our participation in this study for advanced medical education and research, additionally, observing the operations performed or laboratory studies by Acibadem Labgen staff, the use of embryos identified as diseased and/or not transferred in scientific research.

**CONFIDENTIALITY**

Information obtained about us in this study will be kept confidential and our identity will not be revealed without our consent.

**COST**

We have been informed about HLA Tissue typing in the embryo, pre-implantation genetic diagnosis (PGD) and IVF/ET costs. We are committed to paying.

We have read the objective of the PGD applications and the potential risks and accept it of our own accord.

\* As per the Patients' Rights Regulation; 1 form copy will be given to you. Notify us when the form is not given.

**CONSENT**

We applied to ..... IVF and Genetic Diagnosis Center for mutation screening and/or HLA typing procedures by Preimplantation Genetic Diagnosis (PGD) methods after embryo biopsy obtained by microinjection (ICSI) application.

Relevant to the subject, we got detailed information by talking with ..... We also read and understood the "Patient Information Form" which contains general information about PGD.

For PGD and/or HLA processes to be performed we have understood;

- 1) On the third or fifth day following ICSI post-fertilization, 1 or more cells will be removed from embryos by biopsy (1 cell on 3rd day, approx. 4-5 cells on 5th day),
- 2) Cells obtained by biopsy;
  - a) Will be examined for the genetic disease we carry,
  - b) If necessary, it will be examined for HLA typing,
- 3) As a result of the tests performed, all embryos can be found as affected,
- 4) As a result of the tests performed, all embryos can be found as HLA incompatible,
- 5) In cases where there is no healthy embryo or only one embryo and carrier embryos are detected, these embryos can be transferred if HLA compatibility is also present,
- 6) It is possible that more than one ICSI application may be required as the chance of finding an HLA compatible and healthy embryo is low (18, 7%),
- 7) There is a risk of misdiagnosis of about 1% in such procedures,
- 8) Prenatal diagnosis (CVS, amniocentesis or cordocentesis) should be performed in order to confirm the diagnosis if pregnancy has occurred but 0.2-2% chance of pregnancy loss is possible.

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We had the opportunity to ask questions and discuss the procedures to be followed, and we received satisfactory answers. We have been informed about the cost of these procedures.

The details of the applications, the duration, the possible outcomes and the complications, the risks, the consequences that would arise if I don't accept the treatment were explained in detail. We have been informed about the costs of these operations. We authorize ..... Hospital which I know to be a private hospital and Acibadem Labgen in which the necessary examinations are done with physicians, nurses and other health workers, for making this application freely without any pressure or directing, with our own will. We GIVE PERMISSION to make this application with our own consent.

We have read, understood and fully accepted all the steps mentioned above.

	Name-Surname	Date	Signature
Mr.			
Mrs./Ms.			
Translator			

**CERTIFICATION**

I certify that I have given consultancy service the above named spouses and that I disclose the relevant procedures, benefits, risks, alternatives and costs by answering their questions in my knowledge.

I believe that they fully understand my explanations and the answers to their questions.

Every step from the beginning to the end of the PGD process is carried out in accordance with the International PGD Guideline, which is specially prepared for the PGD. In accordance with the content and context of the "Convention on the Protection of Human Rights and Dignity in the Presence of Biology and Medical Practice", it has been published in the 10 June 1998 dated 23368 numbered Official Gazette by Ministry of Health that gender determination can not be made in human embryos other than medical reasons.

All transactions made and information obtained will be protected in our center by adhering to the confidentiality conditions between the patient and the physician. Your personal identity will not be identified in any future medical studies or publications.

	Name-Surname	Date	Signature
Doctor			