SUPRATECH

CONSENT FORM FOR SINGLE-GENE PREIMPLANTATION DIAGNOSIS

Purpose:

Pre-implantation Genetic Diagnosis (PGD) is a technique used in conjunction with In- Vitro fertilization (IVF) to detect embryos with extra or missing chromosomes (aneuploidy) or for conditions caused by single gene defects. Embryos that are affected by certain chromosomal conditions can lead to failure of implantation, pregnancy loss, or result in the birth of a child with physical and/or mental problems. The purpose of PGD is to help prevent adverse outcomes by identifying affected embryos in the laboratory and preventing them from being transferred into the uterus. PGD can help in the selection of chromosomally normal embryos for transfer in order to increase the chance of pregnancy, reduce the chance of miscarriage, and reduce the chance of children born with medical conditions or genetic disorders.

Method:

NGS PGD method (preimplantation genetic diagnosis based on next-generation sequencing platform) uses the most up-to-date techniques of human genome sequencing (reading of genetic information) for testing embryos and opens up new diagnostic possibilities. It is used as part of in vitro fertilization and provides comprehensive information concerning embryo's DNA with regard to diseases or genetic mutations. It provides physicians with a unique opportunity to help couples who are exposed to an increased risk of genetic abnormalities in the fetus. **This is the first solution of this kind in the world.**

Preimplantation Diagnosis with next-generation sequencing NGS PGD

- **Possibility of testing all 24 chromosomes simultaneously with unprecedented accuracy!**
- **Possibility of testing for a variety of monogenic diseases!**
- **Credible result:** Within PGD NGS each sample is assigned an additional molecular code, eliminating the possibility of error since the moment of collecting material from the embryo. In addition, the test credibility is enhanced by a direct connection of DNA reading with the obtained information. Owing to chips within semiconductor technology in the world of DNA molecule is directly reflected in the electronic signal.
- **Embryo safety-reducing the number of biopsies for the diagnosis:** Usually just one embryo biopsy is sufficient to obtain a reliable result. In the case of existing methods, occasionally, the biopsy and the test had to be repeated.
- **The possibility of combining the tests of chromosomes and single-gene diseases in a single test!** Until now, it was not possible to combine chromosomal aneuploidy and monogenic diseases tests. Today, due to PGD - NGD it is possible, just one embryo biopsy will suffice.
- **NGS method is considered to be referential for all the other techniques:** DNA sequencing is described as the reference method (model for others), mainly due to the direct nature of the genetic material reading. Other methods (FISH and microarrays) use markers and light as change markers and indirectly test the genetic material. For this reason, these methods are currently being abandoned for the use of NGS.
- **Lower costs of test:** The special design of the Personal Genome Machine apparatus allows for a significant reduction in the cost of tests in comparison with existing methods. Due to which, we increase the availability of PGD preimplantation genetic diagnosis among patients.

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RISKS AND DISCOMFORTS:

- Preimplantation diagnosis only screens for some of the common chromosomal abnormalities or specific genes. Not all chromosomal abnormalities or gene defects can be detected and there is still a risk of delivering a baby with a chromosomal or gene disorders. Also, in 1-5% of cases, the results of Preimplantation Genetic Diagnosis (PGD) may be inconclusive. In other words, the test failed to pick up an abnormality that exists or no signal was visible.

- Because preimplantation genetic analysis is limited by the technology and the number of cells examined, it is recommended that any patient who conceives after this technique consider routine prenatal diagnosis through chorionic villus sampling (CVS) or amniocentesis to confirm that there are no detectable genetic or chromosomal abnormalities present within the fetus. CVS or amniocentesis would be offered to you based on your age or genetic risk alone. The refusal to undergo CVS or amniocentesis may leave you in the same position as if you had conceived a child naturally, with the same risks of producing a child who has genetic or chromosomal abnormalities. Congenital abnormalities, birth defects, genetic abnormalities, mental retardation and other possible deviations from normal can occur following natural conception, conventional In Vitro Fertilization (IVF), and may also occur following the transfer of embryos that have undergone PGD. Damage or destruction of the embryo is also a potential risk of PGD, although this risk is small. We have reviewed the costs of treatment and will be personally responsible for all expenses. The expenses include, but are not limited to, hospital charges, laboratory charges, and physician professional fees.

All of our questions have been answered, and we know that any future questions concerning our care will be answered by our physician. We have been assured that all information about us obtained during these procedures will be handled confidentially and that neither our identity nor specific medical details will be revealed by clinic personnel without our consent.

We received PGD informed consent counseling from (Name) _____ at (Institution or Center) _____on (Date) _____ The patient information and consent forms regarding In vitro Fertilization (IVF) and Intracytoplasmic Sperm Injection (ICSI) have previously been or will be reviewed and signed prior to PGD testing. We have read the general information for single-gene PGD and we understand that the methods include:

- a) Removal (biopsy) of 1 or 2 cells from suitable embryos three days after insemination by ICSI
- b) The biopsied cells will be tested for the genetic disease for which our children are at risk
- c) The diagnosis may show that all the embryos are affected
- d) In the unlikely event that single-gene PGD testing fails to yield any results, we have the choice of whether or not to transfer embryos that may or may not be affected with disease
- e) In circumstances of recessive disease or disorders which require inheritance of two alterations, embryos that are determined to have a single alteration will most likely be unaffected and may be transferred

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After the embryo transfer, we wish that those embryos that have been determined to be affected with disease, and therefore not frozen for future transfer, be sent to the **Supratech Micropath Laboratory & Research Institute Pvt. Ltd.** (also known as Supratech) to confirm affected status. These embryos will be discarded after conformational testing.

We are aware that single-gene PGD testing has an estimated 5% risk of misdiagnosis; therefore, no guarantee has been given to us regarding the outcome of this test. We have been strongly advised to have prenatal diagnosis testing to confirm the single-gene PGD test results, and we understand the risk associated with not having prenatal diagnosis testing. We also understand the risks involved with chorionic villus sample (CVS) and amniocentesis. If we elect to have prenatal testing performed, we agree to have the sample tested at the Supratech.

We have been informed that some studies report that congenital abnormalities, birth defects, genetic abnormalities, mental retardation, and/or other possible differences may occur in children born following IVF, cell biopsy, and PGD testing. We understand that these problems also occur in 3-5% of children resulting from natural conception without PGD testing.

We are aware that additional genetic alterations associated with our specific disease but not identified in us might exist in an embryo and will not be examined.

We have been informed of the possible risks and consequences associated with PGD testing.

We have had the opportunity to ask questions and discuss the procedure and we have received satisfactory answers.

We consent to these procedures.

Patient's Full Name: _____ Age: _____

Partner's Full Name: _____

Address: _____

_____

Patient Signature_____ Date _____

Partner Signature_____ Date _____

Witness Signature_____ Date _____