MASSACHUSETTS GENERAL HOSPITAL
FERTILITY CENTER

INFORMED CONSENT FOR EMBRYO BIOPSY

FOR USE IN PREIMPLANTATION GENETIC TESTING

This consent form describes pre-implantation genetic testing (PGT), which is used to diagnose a genetic abnormality in embryos created through in vitro fertilization. PGT involves taking cells from cleavage or blastocyst stage embryos. These cells are analyzed for chromosome or gene problems before they are transferred (replaced) into the uterus. PGT has been offered to you because a) you or your partner are at increased risk of having a child with a chromosomal abnormality or a specific gene mutation and have decided to undergo in-vitro fertilization, or b) because you are undergoing in-vitro fertilization for other reasons and have also decided to screen your embryos for aneuploidy (a condition related to extra or missing chromosomes). If you choose to undergo PGT, your embryo analysis will be directed to the genetic risk of concern. Our methods cannot predict all genetic disorders.

For couples with a balanced chromosomal translocation, the purpose of PGT is to evaluate your embryos for the chromosomes involved in the translocation. Embryos containing unbalanced rearrangements of these chromosomes will not be transferred as they would lead to an abnormal baby or a miscarriage. You will not know whether the embryos transferred into your uterus are normal embryos or balanced embryos (carrying the balanced translocation).

For couples at risk for a specific genetic disease, embryos will be tested for that gene, and embryos affected with the disease will not be transferred back into the uterus.

For women who are carriers of a sex-linked disorder, only female embryos will be transferred, as half of the male embryos would be affected.

For couples at risk for aneuploidy or simply screening for aneuploidy, the purpose of PGT is to evaluate the embryos for recognizable chromosomal abnormalities. Embryos thought to have abnormalities in the number of chromosomes (3 chromosomes instead of two, one chromosome instead of two), will not be transferred as they could lead to either a failed cycle, an abnormal baby or a miscarriage.

As part of the PGT screening process, you will be offered a consultation with a genetic counselor to discuss your particular risks of having a chromosomally or genetically abnormal pregnancy. He/she will take a detailed family history including specific questions about possible single gene disorders in your family. It is possible that the genetic counselor might recommend additional testing to first be performed on both of you (or other family members) to be sure that our methods can find your specific chromosome abnormality. This is most commonly recommended for couples proceeding with PGT over concerns related to gene mutations.

Embryo genetic testing may help us to transfer back into your uterus only those embryos found not to be abnormal. However, we also recommend an amniocentesis (taking a sample of fluid from around the baby) or chorionic villus sampling (biopsy of the placenta), through your obstetrician specialist when you are pregnant.

You understand that your IVF cycle may be discontinued if, upon controlled ovarian hyperstimulation, there are not enough mature follicles produced in the ovaries or if the eggs are released from the ovaries before the eggs can be retrieved surgically. In addition, if the female partner is thought to be at risk for the illness Ovarian Hyperstimulation Syndrome, the IVF cycle may have to be canceled.

The PGT procedure involves microscopic removal of cells from the embryo after fertilization. First, a small hole is created in the zona pellucida (outer coating) of the embryo, a procedure that also "assists" with the hatching of the embryo later in the development. Trophectoderm cells are removed from the developing blastocyst on Day 5 or 6 of development. Genetic analysis of these cells will be performed directed by the condition for which you are at increased risk. After the cells are removed for testing, your biopsied embryos will be cryopreserved.

The biopsied cells are prepared for genetic testing by placing them in special tubes and sent by courier to the PGT Reference Laboratory (Reference Lab) performing the genetic test. You will have already contracted the Reference Lab to perform the genetic testing on the cells that we send. When the Reference Lab has completed the testing on the cells, they will notify our office. At that time, we will all discuss which embryos should be transferred to your uterus for the initiation of a pregnancy.

Following embryo transfer, you will be monitored with routine blood tests for human chorionic gonadotropin (hCG). Once you are pregnant, your pregnancy will be followed in a routine fashion.
Please inform your obstetrician that prior to the establishment of the pregnancy, PGT was utilized. This will help the prenatal care team to accurately interpret the results of the prenatal tests. We request permission to contact your obstetrician for the results of your chorionic villus sampling (biopsy of a piece of placenta for chromosomal or gene analysis), or amniocentesis (testing of cells in the fluid around the baby for chromosomal or gene analysis), prenatal care and delivery, and to contact your child's pediatrician for growth and development information.

Do you agree to allow us to contact your obstetrician for information regarding the amniocentesis, CVS, prenatal care and delivery and your pediatrician for information about your child's growth and development?

Yes ____ No ____

RISKS AND DISCOMFORTS

- Embryos may be damaged during the biopsy procedure reducing their ability to initiate a normal pregnancy.
- Risks of the genetic testing of cells of the embryo are that the cells could test normal when, in fact, other cells in the embryo are abnormal, leading to transfer of an abnormal embryo. The scientific literature reports that this occurs in about 0.3% to 5.6% of the cells that are diagnosed.
- Alternatively, the cell biopsy may test abnormal when the remainder of the embryo is in fact normal, but not transferred due to this error.
- Additionally, at this time we are unable to test for all chromosomal abnormalities; therefore, even if embryos test normal for the chromosomes tested, other abnormalities could be missed, leading to an abnormal pregnancy.
- Birth defects and other types of problems may occur in pregnancies that are completely normal chromosomally and would not be found by our testing.
- There is a possibility that none of your embryos will test normal and that none will be recommended for transfer.

ALTERNATIVES

- Conceive a pregnancy on your own and have an abnormal pregnancy terminated once the diagnosis is made by amniocentesis or chorionic villus sampling,
- Conceive a pregnancy on your own and carry an abnormal pregnancy to term,
- Insemination of the female patient with sperm from a donor that has proven to not carry the gene for the disease,
- Use of eggs obtained from an egg donor who has been proven to not carry the gene for the disease, or
- Adoption

COSTS

PGT costs are separate from IVF. There will be charges for the embryo biopsy procedure. In addition, there will be the charges for the genetic diagnosis that is performed on the cells removed from the embryos. An outside diagnostic laboratory that you have contracted with to do this work is performing these tests. You understand that you will be responsible for all of the costs for this procedure, including potentially the cost of standard IVF (if not covered by your insurance). You understand that you are responsible for paying these costs prior to the cycle if not covered by your insurance. Such costs do not include the IVF medications, which you will receive from the pharmacy of your choice and pay for yourself, if not covered by insurance.

CONSENT

This consent will function as an addendum to the consent form entitled Informed Consent for In Vitro Fertilization, which we executed previously. We, the undersigned, have read this document, understand the purposes, risks, and benefits of this procedure and have been given the opportunity to ask questions about it, which have been answered to our satisfaction. We consent to having our embryos biopsied for use in pre-implantation genetic testing.

Patient	Partner (if applicable)
Print Name	Print Name
Date of Birth	Date of Birth
Date	Date