
Preimplantation Genetic Testing (PGT)

Process, Risk, and Consent

What is pre-implantation genetic testing (PGT)

Your embryos can be tested for abnormal chromosomes before they are transferred to the uterus. This is called preimplantation genetic testing, or PGT. It is done in a lab, using in vitro fertilization (IVF). Five-ten cells are taken from each embryo and sent for genetic testing. As soon as the cells are taken, the embryos are frozen while we wait for the results of the testing. Once the genetics lab tells us which embryos are healthy, plans are made to transfer a normal embryo into the uterus, where the embryo may attach to the lining of the uterus and produce a pregnancy.

What genetic tests can be performed?

Cells from the embryo can be tested for a gene that may produce a specific disease. This will show if an embryo is normal (no disease) or affected (has the disease). It will also show if the embryo is carrying the disease but unaffected by it. An unaffected carrier is someone who can pass the disease to his or her children, but does not personally have any signs of the disease.

Cells from the embryo can also be tested to see if the embryo has more or fewer chromosomes than the usual 46. Having too many or too few chromosomes can cause disorders such as Down syndrome (Trisomy 21) or Trisomy 13. This test can also look for out-of-order chromosomes, called translocations, which can cause problems with growth or function.

The number of tests for genetic problems keeps growing every year.

What is PGT-M?

PGT-M stands for “preimplantation genetic testing for monogenic disorder”. PGT-M involves testing an embryo before it implants for a specific, known genetic disorder. PGT-M is used so that embryos unaffected by the disorder can be returned to the uterus.

What is PGT-A?

PGT-A stands for “preimplantation genetic testing for aneuploidy”. PGT-A involves testing an embryo to see if it has the usual 46 chromosomes. After an egg retrieval and fertilization by sperm, women will have some embryos that have 46 chromosomes, and some embryos with too many or too few chromosomes. Many embryos with chromosomal abnormalities will not lead to a pregnancy, or will result in a miscarriage. The percentage of embryos that are abnormal can be affected by many factors, including the age and health history of the parents.

How is genetic testing performed?

Several cells are taken from an embryo and sent for genetic testing. After the cells are taken, the embryos are frozen. Once the genetic testing results come back, plans are made to transfer a genetically normal embryo into the uterus.

Who should have PGT-M or PGT-A?

PGT-M is for someone who has or is a carrier of a known genetic disorder. PGT-A may be recommended for someone with recurrent miscarriage, or someone with multiple failed fertility treatments. Some couples also can choose to do PGT-A for personal reasons.

What happens once the PGT results come back?

After PGT, a normal embryo is chosen for embryo transfer. On the morning of the embryo transfer the chosen embryo will be warmed, and the remaining embryos identified as normal by genetic testing will remain frozen. Embryos that are not genetically, chromosomally, or morphologically suitable for embryo transfer will be disposed.

Quality control in the lab is extremely important. Sometimes, USF IVF may use abnormal embryos for quality control prior to discarding them in accordance with normal laboratory procedures and applicable laws. None of this material will be used to establish a pregnancy or a cell line.

Risks and limitations of PGT include, but are not necessarily limited to, the following:

- Embryos may not develop far enough to have cells removed for PGT
- An embryo may be damaged during cell removal for PGT
- Cells may be damaged or lost during or after the procedure
- The cell removal procedure may not be successful in obtaining enough cells for genetic testing to be carried out
- The genetics lab may not be able to analyze the cells, and/or may not be able to provide a diagnosis or screening result
- There is a small risk of misdiagnosis on the part of the genetics laboratory
- All technologies used for PGT have limitations, and may not be able to identify all genetic abnormalities.
- After genetic evaluation, it is possible that no suitable embryos will be available for embryo transfer
- Transfer of a genetically normal embryo does not guarantee a successful attachment or pregnancy
- Miscarriages can still occur after embryo transfer and implantation of a genetically normal embryo
- Transfer of an embryo identified as normal by PGT does not eliminate the risk of having a genetically abnormal baby, of having a baby with birth defects or congenital malformations, of having a baby with other developmental delays, or of having a baby with any/other health issues

I/we agree that USF IVF is not responsible for the occurrence of any of the above or similar events, and release USF IVF from associated liability if any of these events were to occur.

USF IVF will take reasonable steps to reduce the possibility of technical failure in the transport of cells from our facility to the genetics laboratory. However, it is possible that loss or specimen damage in the transport process, theft, an act of nature, or other events could occur. I agree that USF IVF is not responsible for any loss or damage to the biopsied specimen during the transport process. I release USF IVF from associated liability if any of these events were to occur.

We (I) acknowledge that we have read and understood the information provided above regarding the PGT process and its risks, and agree to go forward with this treatment as our signatures below testify.

Patient name printed

Partner name printed (if applicable)

Embryo Biopsy for Preimplantation Genetic Testing

Patient Signature

Partner Signature (if applicable)

Date

Disposal of embryos that are not genetically, chromosomally, or morphologically suitable for embryo transfer

Patient Signature

Partner Signature (if applicable)

Date

USF IVF Staff:

Print name

Signature

Date

Consent is valid for 1 year from date of signature

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