

Preimplantation Genetic Testing

Preimplantation genetic testing (PGT) is a way to test an embryo for genetic disease or an inherited condition. Testing is done before an embryo is placed into the uterus. In vitro fertilization (IVF) is required for PGT.

How is PGT done? Eggs are collected from the ovaries and fertilized at a clinic to create embryos. Five to six days after an egg is fertilized, a few cells are carefully removed from the embryo. The cells are sent to a genetic laboratory for testing. The embryo is frozen until the test results are ready. Results usually take about 1-2 weeks, but may take longer. Embryos with normal results are selected for transfer to the uterus.

Why would you have PGT?

- You have a genetic disease or chromosome condition that runs in the family.
- You and your partner are carriers of a recessive disease.
- You want to select a sibling match for bone marrow transplant.
- You want to check for normal chromosomes in your embryos.

What are the limits of PGT?

- You may not get a result for some embryos.
- You may get an incorrect result.
- Testing would still be recommended during pregnancy.
- Your baby could have a condition that is not detected by PGT.
- You may have embryos that are not transferred.
- Your cost for IVF and PGT may not be fully covered by medical insurance.

Types of Preimplantation Genetic Testing

There are three different types of PGT. These tests can be combined.

PGT-M

PGT-M (PGT for “Monogenic diseases”) This test looks for genetic diseases that are due to changes in a single gene, such as cystic fibrosis or sickle cell disease. PGT-M is used when a baby is at risk for a specific inherited disease.

PGT-SR

PGT-SR (PGT for “Structural Rearrangements”) This test looks for inherited chromosome conditions that are due to chromosome changes in one or both parents, such as a translocation or inversion.

PGT-A

PGT-A (PGT for “Aneuploidy”). This test screens the embryo for random changes in the chromosomes. The test looks for extra or missing chromosomes (aneuploidy). It helps lower the chance for chromosome conditions that do not run in families, such as Down syndrome.

FREQUENTLY ASKED QUESTIONS

Does taking cells for PGT harm the embryo?

The cells taken for testing come from the part of an embryo that develops into the placenta. The small number of cells removed does not appear to harm the placenta or fetus.

Is it safe to freeze embryos?

Freezing and thawing an embryo appears to be safe. Pregnancy outcomes with frozen embryos are as successful as using fresh (never frozen) embryos. Most embryos survive the thawing process.

Does PGT include testing for Down syndrome and other chromosome conditions?

It depends on which test is being done. Most chromosome conditions do not run in the family. Instead they happen by chance when an egg is fertilized. PGT-A is used to check for random chromosome differences in an embryo. PGT-SR checks for inherited chromosome conditions and may also test for random chromosome conditions, including Down syndrome. PGT-M does not test for chromosome conditions.

Can I have more than one PGT test?

The same embryo sample may be used for more than one PGT test. If you are having PGT for an inherited condition, you may also request PGT-A to check for random chromosome conditions. There may be an added cost to have more than one test.

How much does PGT cost?

The cost for PGT includes the cost for the IVF procedure as well as genetic testing on each embryo. The cost for one IVF cycle can be \$10,000 to \$15,000 (or more). Medications needed for IVF can cost \$5,000 to \$8,000. There is also a separate cost for the embryo biopsy that can be \$3,000 or more. Plus, there is a cost paid to an outside laboratory to test each embryo. Most insurance plans do not fully cover IVF and PGT costs.

GLOSSARY

Aneuploidy – Too many or too few chromosomes in a cell. Usually there are exactly 46 chromosomes in each cell.

Biopsy – A sample of cells taken for testing.

Blastocyst – An embryo at day 5 or 6 after fertilization. This is when a few cells can be taken for PGT.

Embryo – The earliest stage of a developing baby. This term is used from the time an egg is fertilized until the eighth week in pregnancy.

Fetus – An unborn baby more than eight weeks after conception.

In vitro fertilization (IVF) – The process of combining eggs and sperm outside the body in a clinic or laboratory. Once an embryo forms, it can be placed in the uterus.

Monogenic disease – A genetic condition caused by a mutation in one gene.