PGT genetic testing: uses, benefits, and risks

What is PGT genetic testing?

Preimplantation genetic testing is an embryo testing strategy used during IVF. Eggs are retrieved after ovarian stimulation, fertilized in the laboratory, and cultured for several days, often to the blastocyst stage. A small number of cells is then biopsied, usually from the trophectoderm, the cell layer that contributes to the placenta. The embryo is typically cryopreserved while genetic analysis is performed.

PGT is not a single test. It is an umbrella term for several different analyses with different goals. The American College of Obstetricians and Gynecologists describes three major categories: PGT-A, PGT-M, and PGT-SR. Each is designed for a different clinical question, and choosing the correct type matters.

Because PGT tests only sampled cells from an early embryo, results are highly informative but not absolute. The result may guide embryo selection, but it cannot diagnose every possible genetic condition, exclude all birth defects, or ensure implantation. IVF success also depends on uterine factors, endometrial receptivity, embryo development, sperm and egg quality, and many other variables.

Types of PGT: PGT-A, PGT-M, and PGT-SR

PGT-A, or preimplantation genetic testing for aneuploidy, evaluates embryos for abnormal numbers of chromosomes. Human embryos typically have 46 chromosomes. Aneuploidy means there is an extra or missing chromosome, such as trisomy or monosomy. These abnormalities are common in early embryos, especially as maternal age increases, and are associated with implantation failure, miscarriage, or chromosomal conditions.

PGT-M, or preimplantation genetic testing for monogenic disorders, is used when there is a known risk of a specific single-gene condition in a family. Examples may include autosomal recessive, autosomal dominant, or X-linked conditions, depending on the genetic diagnosis and inheritance pattern. PGT-M usually requires advance test development, including genetic reports and sometimes DNA samples from relatives, so the laboratory can track the familial variant accurately.

PGT-SR, or preimplantation genetic testing for structural rearrangements, is considered when one partner carries a balanced chromosomal rearrangement, such as a reciprocal translocation, Robertsonian translocation, inversion, or other structural change. A carrier may be healthy but produce embryos with unbalanced chromosomal material, increasing the risk of miscarriage or an affected pregnancy. PGT-SR aims to identify embryos with balanced or normal chromosomal content suitable for transfer.

Some IVF cycles combine more than one PGT approach, for example PGT-M plus chromosomal screening. Whether this is useful depends on the specific indication, embryo number, laboratory platform, and counseling about what each result can and cannot show.

When PGT may be considered

PGT is most clearly useful when there is a defined genetic or chromosomal risk. Couples or individuals may consider PGT-M if genetic testing shows they carry a pathogenic variant that could be passed to a child. This is particularly relevant for people with a prior affected child, a known family history, carrier screening results showing shared recessive risk, or a personal diagnosis of an inherited condition.

PGT-SR may be discussed when karyotype testing identifies a balanced translocation or other rearrangement in one partner. This situation can be emotionally difficult because the carrier may feel entirely well yet experience recurrent pregnancy loss or infertility. A genetic counselor can help estimate reproductive risks and explain how embryo testing might change the probability of transfer, miscarriage, or live birth.

PGT-A is more debated. It may be considered in selected situations, such as advanced maternal age, recurrent pregnancy loss, repeated IVF failure, or a large number of embryos where chromosomal screening may help prioritize transfer. However, ACOG notes that routine use of PGT-A in all infertile patients is not currently supported by sufficient evidence. The potential value varies by age, embryo number, prognosis, and clinic-specific practices.

PGT is only available in the context of IVF. That means it may involve ovarian stimulation, egg retrieval, fertilization, embryo culture, biopsy, freezing, and embryo transfer. For some patients, the first step is understanding IVF medications and ovarian response; related fertility treatment topics such as gonadotropin injections and ovarian stimulation may be part of the conversation with the care team.

Potential benefits of PGT

For patients with a known monogenic disorder risk, the most important benefit of PGT-M is the opportunity to reduce the chance of transferring an embryo affected by that specific condition. It can help some families avoid repeated pregnancy terminations or the recurrence of a serious inherited disease, although confirmatory prenatal testing may still be advised.

For carriers of structural chromosomal rearrangements, PGT-SR may help identify embryos that are chromosomally balanced or normal, reducing the likelihood of transferring embryos with unbalanced chromosomal content. This may lower the risk of miscarriage related to that rearrangement, although it cannot prevent all causes of pregnancy loss.

PGT-A may help select embryos with a euploid chromosome complement for transfer. In selected groups, this can reduce the number of transfers of

embryos unlikely to implant or likely to miscarry because of aneuploidy. It may also reduce the risk of multiple pregnancy when used to support single embryo transfer, because clinicians may feel more confident transferring one embryo with reassuring results.

Other possible benefits include more efficient embryo prioritization, shorter time to pregnancy for some patients, and more information for planning future embryo transfers. For people who have endured repeated losses or uncertainty, having additional embryo-level information can feel empowering. At the same time, the emotional benefit depends heavily on how results are explained and whether expectations are realistic.

Limitations and accuracy issues

PGT is powerful, but it is not a guarantee. One central limitation is mosaicism. An embryo may contain more than one cell line, such as some euploid cells and some aneuploid cells. Because the biopsy samples only a few trophectoderm cells, the result may not perfectly represent the inner cell mass that becomes the fetus. Mosaic results can be difficult to interpret and may require detailed counseling.

False-positive and false-negative results are possible. Laboratory error is uncommon in high-quality programs but cannot be reduced to zero. Biologic factors, DNA amplification issues, contamination, allele dropout in single-gene testing, or limitations of the platform can affect interpretation. PGT-M is usually designed for a specific familial variant; it does not screen the embryo for every genetic disease unless other testing is added.

PGT-A typically evaluates chromosomal copy number, not all gene-level changes. A euploid PGT-A result does not rule out single-gene disorders, congenital anomalies, placental problems, or pregnancy complications. Similarly, PGT-M for a particular condition does not necessarily mean the embryo has normal chromosomes unless aneuploidy testing is also performed.

Another important limitation is that testing may leave no embryos recommended for transfer. This result can be devastating, especially after a physically and financially demanding IVF cycle. Sometimes the issue reflects true embryo genetics; sometimes it reflects low embryo number, maternal age-related

aneuploidy, sperm or egg factors, or chance. Counseling before the cycle should include what will happen if results are inconclusive, mosaic, abnormal, or if no embryo is available.

Risks and burdens to consider

PGT itself requires embryo biopsy, and modern biopsy techniques are generally considered safe in experienced laboratories. Still, embryo manipulation is not risk-free. Some embryos may not tolerate biopsy or freezing and thawing, and outcomes may vary by embryo quality and lab expertise. Patients should ask their clinic about blastocyst survival rates, biopsy practices, reporting categories, and how mosaic or inconclusive results are managed.

The medical risks are tied to IVF as a whole. Ovarian stimulation can cause bloating, discomfort, and rarely ovarian hyperstimulation syndrome. Egg retrieval involves sedation and carries small risks such as bleeding, infection, or injury to nearby structures. Embryo transfer is usually low risk, but pregnancy after IVF still requires standard obstetric care.

Financial burden is often substantial. PGT may add costs for biopsy, laboratory analysis, genetic test development, embryo freezing, storage, and future transfers. Insurance coverage varies widely and may distinguish among PGT-A, PGT-M, and PGT-SR. Some policies cover testing only for specific indications, while others consider certain uses investigational.

Emotional and ethical burdens deserve equal attention. Patients may face decisions about embryos reported as affected, aneuploid, mosaic, or inconclusive. Some people struggle with embryo disposition, religious concerns, family expectations, or grief when results are not what they hoped. Genetic counseling and mental health support can be valuable before and after testing.

What happens after a PGT result?

After results return, the fertility team typically reviews which embryos are recommended for transfer, which are not, and which require additional discussion. Embryo selection usually considers both genetic results and embryology grading. A euploid embryo with good development may be prioritized, but grading and genetics do not predict success perfectly.

If PGT-M or PGT-SR was performed, the report may classify embryos by whether they are unaffected, carriers, affected, balanced, unbalanced, euploid, aneuploid, mosaic, or inconclusive, depending on the test. The terminology can be confusing, and it is reasonable to request a dedicated session with a genetic counselor to review the report line by line.

Pregnancy testing and obstetric care proceed as they would after other IVF transfers. Importantly, PGT does not replace prenatal screening or diagnostic testing. Many clinicians still offer noninvasive prenatal screening, ultrasound evaluation, chorionic villus sampling, or amniocentesis depending on age, history, PGT type, and patient preference. Diagnostic prenatal testing is the standard way to confirm fetal chromosomal or genetic status during pregnancy.

If transfer does not lead to pregnancy, it does not mean the PGT result was wrong. Even chromosomally reassuring embryos may fail to implant because pregnancy probability is never 100 percent. This can be painful, but it is a known limitation of reproductive biology rather than a personal failure.

Questions to ask your fertility clinic and genetic counselor

Which type of PGT are you recommending: PGT-A, PGT-M, PGT-SR, or a combination?
What specific condition, variant, or chromosomal finding is being tested?
How will the laboratory report euploid, aneuploid, mosaic, carrier, affected, balanced, or inconclusive embryos?
What are the clinic’s embryo biopsy, freezing, thaw survival, and live birth outcomes for patients with a similar prognosis?
What are the costs, insurance requirements, and expected timeline, including test development for PGT-M?
What options will we have if all embryos are abnormal, mosaic, or inconclusive?
Will prenatal screening or diagnostic testing still be recommended if pregnancy occurs?

These questions are not only technical. They help ensure that the testing plan matches your goals, risk tolerance, and values. A thoughtful consent process should include possible outcomes before embryos are created, not only after results arrive.