VARTA
Victorian Assisted Reproductive Treatment Authority

Preimplantation genetic testing (PGT)

Preimplantation genetic testing (PGT) is a technique used in IVF to help people reduce their risk of passing on a known genetic condition. There are two types of PGT:

- PGT for monogenic/single gene defects (**PGT-M**) is used to identify embryos that are not affected by a 'faulty' gene that can lead to disease.
- PGT for chromosomal structural rearrangements (**PGT-SR**) is used to identify embryos that have the correct amount of genetic material.

PGT-M and PGT-SR are also known as preimplantation genetic diagnosis (**PGD**). They are different to **PGT-A** (preimplantation genetic testing for aneuploidy) which is covered in another VARTA factsheet.

How is PGT done?

Hormone stimulation	Fertility drugs are given to develop eggs (stimulated cycle).
Egg retrieval	Eggs are retrieved from the ovaries and sperm are added to the eggs to create embryos.
Embryo development and biopsy	Embryos are grown in the laboratory for 5 - 6 days. At this time, two types of cells can be seen: the cells that will develop into the placenta and the cells that will become the foetus. A few cells are removed from the future placenta for genetic testing to minimise the risk of harm to the developing foetus.
PGT	Genetic testing is performed on the cells that are removed. The time it takes to get PGT results varies from days to weeks, depending on the method used and where the testing is done. Embryos are frozen until test results are available.
Embryo transfer	If the test shows an embryo is normal it is thawed and used for transfer into a patient's uterus to attempt a pregnancy.

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Advantages and disadvantages of PGT-M and PGT-SR

PGT does not guarantee the birth of a healthy baby. People who become pregnant are advised to have prenatal testing (e.g. DNA testing or chorionic villus sampling) to confirm PGT results.

Here are some of the advantages and disadvantages of embryo testing. For more detailed information and to find out if PGT is right for you, speak to your doctor or a genetic counsellor.

Advantages	Disadvantages

• People who carry a 'faulty' gene or chromosomal rearrangement can reduce the risk of having a child with a serious health condition.
• For people with a history of miscarriage due to a known structural rearrangement in their chromosomes, PGT-SR reduces the risk of miscarriage.
• It reduces the risk of having to make difficult decisions later, including whether to terminate or continue a pregnancy if a foetus has a genetic or chromosomal condition.[1]
	• PGT requires IVF treatment, which fertile people do not otherwise need.
	• The cost of PGT is not covered by Medicare and comes on top of the costs of IVF.
	• PGT only tests for specific conditions, so it does not eliminate the risk of a child having another condition not covered by the test.
	• Embryos may be damaged in the biopsy procedure.[2]
	• Due to technical challenges, there is a small chance the test results may not reflect the true health of the embryo.[3]
	• In the case of chromosomal rearrangements, the embryo may contain a mixture of cells with normal and abnormal chromosome arrangements. This is called mosaicism. This can cause a false positive or false negative PGT result.[4]
	  ○ **A false positive result** means that the few cells that are tested show abnormalities, while the remaining cells may be normal. Based on PGT alone, an embryo that may have led to the birth of a healthy baby may be discarded.
	  ○ **A false negative result** means that the few cells that are tested are normal while the remaining cells may have abnormalities. Based on the test result, a chromosomally abnormal embryo may be transferred.
	• Embryos may not survive the thawing process.
	• Sometimes none of the embryos are suitable for transfer due to a genetic abnormality. Your doctor will discuss your results and options with you.

[1] Lamb, B., Johnson, E., Francis, L., Fagan, M., Riches, N., Wilson, A., Johnstone, E. (2018). ***Pre-implantation genetic testing: decisional factors to accept or decline among in vitro fertilization patients***. Journal of Assisted Reproduction and Genetics, 35(9), 1605-1612.

[2] Cimadomo, D., Capalbo, A., Ubaldi, F. M., Scarica, C., Palagiano, A., Canipari, R., & Rienzi, L. (2016). ***The impact of biopsy on human embryo developmental potential during preimplantation genetic diagnosis***. BioMed research international, 2016.

[3] Brezina, P. R., & Kutteh, W. H. (2015). ***Clinical applications of preimplantation genetic testing***. BMJ, 350, g7611.

[4] Victor, A.R., Tyndall, J.C., Brake, A.J., Lepkowsky, L.T., et al. (2019). ***One hundred mosaic embryos transferred prospectively in a single clinic: exploring when and why they result in healthy pregnancies***. Fertility and Sterility, 111(2):280-93.

Who are these tests for?

PGT for monogenic conditions (PGT-M)

Some people carry a 'faulty' gene that may not affect them but can cause severe genetic conditions in their offspring (e.g. cystic fibrosis or Huntington's disease). They may become aware of this through pre-pregnancy genetic carrier screening, if they have a child affected by the condition, or if a family member is diagnosed.

A PGT-M test can be designed for almost any genetic disorder caused by a single 'faulty' gene, as long as the location of the gene causing the disorder is known.

PGT for structural rearrangements (PGT-SR)

Some people have chromosomal rearrangements that do not affect their health but can affect their chance of having a healthy baby. They may become aware of this as a result of chromosomal testing, which is often recommended after repeated miscarriages or the birth of a child with a chromosomal abnormality.

PGT-SR testing can be used for any chromosome rearrangement that a parent is known to have and helps identify embryos with the correct amount and arrangement of chromosomal material.

Where can I get more information?

Your fertility specialist and genetic counsellor can answer any questions you have.

For more information:

- Access Australia: Preimplantation Genetic Diagnosis (PGD)
- Centre for Genetics Education: Preimplantation Genetic Diagnosis (PGD)