VARTA
Victorian Assisted Reproductive Treatment Authority

Pre-implantation genetic testing explained

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Pre-implantation 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 **pre-implantation genetic diagnosis (PGD).**

How is PGT done?

Hormone stimulation

Fertility drugs are given to develop a number of 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 are distinguishable: the cells that will develop into the placenta and the cells that will become the baby.
A few cells are removed from the future placenta for genetic testing to minimise the risk that the developing fetus is harmed.

PGT

Genetic testing is performed on the cells that are removed.
The time it takes to get PGT results can vary from days to weeks and depends on the method used and where the testing takes place.
Embryos are frozen until test results are available.

Embryo transfer

If the test shows that there is one or more normal embryos, one is thawed and transferred to the woman's uterus. Any remaining normal embryos will be kept frozen for transfer later if the first transfer does not lead to a pregnancy.

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Who needs which test?

PGT for monogenic conditions (PGT-M)

Some individuals 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 when they have a child affected by the condition or if they have a family member who is affected.

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 individuals 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.

Advantages and disadvantages of PGT-M and PGT-SR

It is important to know that **PGT does not guarantee the birth of a healthy baby.** Women who become pregnant are advised to undergo 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 for you, please speak to your doctor or genetic counsellor.

Advantages

- For people who are known to carry a faulty gene or a chromosomal rearrangement, it reduces the risk of having a child with a serious health condition.
- For women who have had previous miscarriages 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 with a fetus affected by a genetic or chromosomal condition.[1]

Disadvantages

- PGT requires IVF treatment, which fertile people do not otherwise need.
- The cost of PGT is not covered by Medicare and is in addition to the costs of IVF.
- PGT only tests for specific conditions, so it does not eliminate the risk of a child having another condition that the test does not cover.
- Embryos may not survive the biopsy procedure.[2]
- Due to technical challenges, there is a small chance that 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.

References

[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.

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Where can I go for more information?

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

There are also support networks that you can join.

Please see the VARTA website for a list of these support networks: **www.varta.org.au/resources**

For more information regarding PGT

Australian National Infertility Network
www.access.org.au/wp-content/uploads/2016/08/42-pgd.pdf

VARTA information booklet on PGT
www.varta.org.au/sites/varta/files/What%20is%20PGD%20-%20brochure.pdf

Centre for Genetics Education
www.genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-29-preimplantation-genetic-diagnosis-pgd

VARTA
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