Your guide to preimplantation genetic testing

What is preimplantation genetic testing (PGT)?

Preimplantation genetic testing checks the cells of an embryo (fertilised egg) for a genetic condition or chromosome abnormalities. This helps find embryos that do not have these issues. An unaffected embryo can then be placed in the uterus (womb) to try to start a pregnancy. This reduces the risk of passing these conditions on to your child.

There are 2 types of PGT available through Public Fertility Care at the Women's:

- PGT-M: preimplantation genetic testing for monogenic conditions
- PGT-SR: preimplantation genetic testing for structural rearrangements.

PGT-M: Preimplantation genetic testing for monogenic conditions

This test is an option for individuals or couples who have a higher chance of having a child with an inherited genetic condition caused by a single faulty gene.

You might consider this test if:

- Both you and your reproductive partner are carriers of a genetic condition that only appears if both of you pass it on. This is called an autosomal recessive genetic condition.
- Either you or your reproductive partner has a genetic condition that could be passed on to your child. This is called an autosomal dominant genetic condition.
- You're female, or a person with XX chromosomes, and you carry a genetic condition linked to the X chromosome. This is called an X-linked genetic condition.

PGT-SR: Preimplantation genetic testing for structural rearrangements

This test is an option for individuals or couples with changes in their chromosomes called 'rearrangements'. This can cause repeated miscarriages or increase the chance of having a baby with a physical or intellectual disability.

These changes include:

- 2 chromosomes swapping pieces (reciprocal translocation)
- 2 chromosomes joining together (Robertsonian translocation)
- a piece of a chromosome breaking off, turning upside down, and reattaching to the chromosome (chromosome inversion)
- other complex changes in chromosomes.

How is PGT done at The Women's?

First, you'll need to have some fertility tests from your doctor (GP) and Public Fertility Care (PFC).

Then, you'll meet with a genetic counsellor or clinical geneticist to talk about PGT and how it works.

For PGT-M, you and/or your reproductive partner need to have already had genetic tests to identify the specific genetic condition and the exact change in the gene. The lab will then design a custom test to check your embryos for that condition. This is called a feasibility study. A feasibility study involves:

- getting DNA samples from you and/or your reproductive partner
- getting DNA samples from close family members, such as children, parents, siblings, or DNA saved from a past pregnancy.

This process usually takes 4 to 6 weeks.

Once the genetic test is ready, the in-vitro fertilisation (IVF) process can begin to create embryos for testing.

In rare cases, a genetic test cannot be made to test the embryos.

For PGT-SR, you usually don't need a feasibility study or samples from other family members. However, the lab will need to review the information about the chromosome rearrangement to decide if PGT-SR is suitable.

In-vitro fertilisation (IVF)

IVF begins with hormone injections to help the ovaries produce eggs. A short surgical procedure is then used to collect the mature eggs.

In the lab, the eggs are mixed with sperm to fertilise them and create embryos.

Embryo testing

After embryos grow in the lab for 5 to 7 days, we take a few cells from each to test for the genetic condition or chromosome abnormality. This is preimplantation genetic testing (PGT).

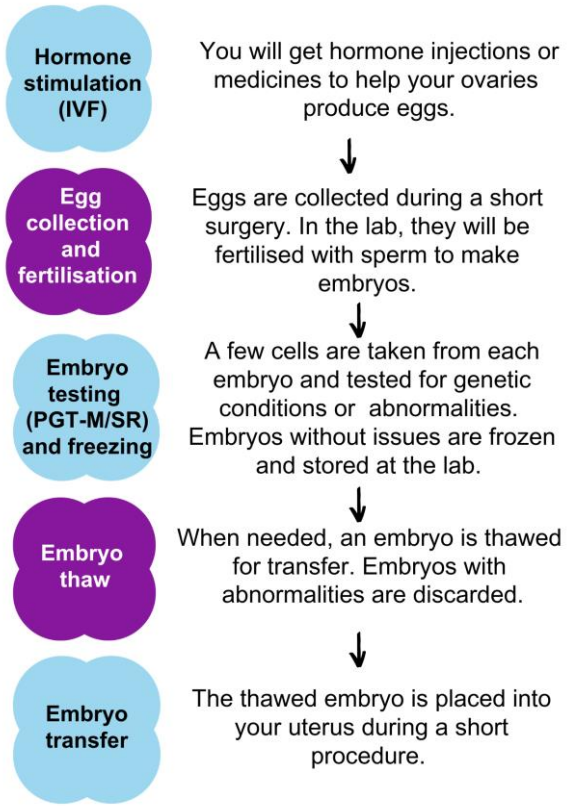
We send these cells to an external lab for testing. The embryos are frozen while we wait for the results, which can take about 4 weeks.

With PGT-M, we select an embryo that does not have the genetic condition. This embryo is then thawed and put into the uterus to try to start a pregnancy.

With PGT-SR, we select an embryo without a chromosome abnormality. This embryo is thawed and put into the uterus to try to start a pregnancy.

If we have more than 1 unaffected embryo, we will keep them frozen for you to use in the future. Embryos with the genetic condition or a chromosome abnormality are discarded.

Here's a brief overview of the process.



How accurate is PGT?

PGT-M analysis is very accurate and usually finds more than 99 in 100 (>99%) affected embryos.

PGT-SR is also very accurate and finds over 97 in 100 (>97%) affected embryos.

But, with both PGT-M and PGT-SR, there's still a small chance the testing could be wrong. Because of this, it is recommended to do another test during pregnancy to make sure the baby is not affected by the genetic condition or chromosome abnormality.

This second test is called prenatal testing or prenatal diagnosis. Prenatal testing is regularly offered, but it is up to you to decide if you want to have it done.

There are 2 types of prenatal testing: chorionic villus sampling (CVS) and amniocentesis.

For both tests, a doctor uses a needle to collect cells.

Chorionic villus sampling (CVS) takes a small piece of tissue from the placenta. It can be done between 11 and 13 weeks of pregnancy.

Amniocentesis takes a small amount of amniotic fluid. It can be done starting at 16 weeks.

Both tests have a small risk of miscarriage. This is less than 1 in 100 (<1%).

Non-invasive prenatal testing (NIPT) may be an option for screening for some chromosome abnormalities, but it does not check for single gene conditions.

NIPT is not as accurate as CVS and amniocentesis. To do this test, a small amount of blood is taken from your arm and sent to the lab for testing.

Extra testing for chromosome abnormalities

When embryos get PGT-M testing, they also get checked for extra or missing chromosomes (called chromosome aneuploidy).

Embryos that have PGT-SR testing to look for chromosome abnormalities related to a parent's chromosome rearrangement are also tested for other chromosome aneuploidies.

Chromosome aneuploidy is a common cause of early miscarriage. It can also be present in ongoing pregnancies. An example of a condition caused by chromosome aneuploidy is Down syndrome.

The chromosome testing completed at the time of PGT is not 100% accurate. It also does not find all types of chromosome abnormalities.

It's a good idea to consider other prenatal screening or testing for chromosome conditions.

This can include non-invasive prenatal testing (NIPT), first trimester combined screening, chorionic villus sampling (CVS) and amniocentesis.

Other things to consider

- Having IVF with PGT does not always lead to pregnancy. Sometimes, the eggs collected might not be good quality, or the embryos tested might not be healthy. Even if an embryo is transferred into the uterus, it may not grow into a pregnancy.

- PGT tests look for the specific genetic or chromosome issues that a person or their partner are at high risk for. It also checks for some common chromosome aneuploidies. However, it doesn't test for every possible condition. All pregnancies have a 3 to 4 in 100 (3 to 4%) chance of having a baby with a birth defect or a medical issue that appears in the first year.
- The PGT and IVF process can take several months to complete.
- Some of the medicines and hormone injections might cause side effects.
- Some people might find the process emotionally difficult.

For more information

If you have any questions about preimplantation genetic testing, please ask your doctor or nurse at your next appointment.

Useful contacts

The Women's

Public Fertility Care

Ph: 03 8345 3200
E: reproductiveservices@thewomens.org.au
Monday to Friday, 8.30am to 5pm

Genetic Counselling Service

P: 03 8345 2180
E: pfsgenetics@thewomens.org.au

Other services

Victorian Assisted Reproductive Treatment Authority (VARTA)

VARTA helps people learn how to increase their chances of having a baby. Visit their website for information on fertility, fertility treatments, surrogacy and donor conception.
varta.org.au

Centre of Perinatal Excellence (COPE)

Provides information and support to reduce the impacts of emotional and mental health issues before and after childbirth.
cope.org.au

Antenatal and Postnatal Psychology Network

Connect with Victorian psychologists who can help you with pregnancy and early parenting challenges. Visit their website to find a psychologist.
antenatalandpostnatalpsychology.com.au

Family Violence Support

1800 Respect National Helpline

You can get help if you have experienced sexual assault, domestic or family violence and abuse.

You can call any time of day or night.

1800 737 732

1800respect.org.au

Do you need an interpreter?

You can ask for an interpreter if you need one.