REPRO

Reproductive options for people with genetic conditions

If you have a genetic diagnosis confirmed, and you are planning a pregnancy, there are reproductive options available to you.

These are some of your options:

1. Conceiving naturally and having no genetic testing. This means accepting the chance of passing on the genetic condition to your child.
2. Conceiving naturally and having testing early in pregnancy (**prenatal testing**).
3. Conceiving via **in-vitro fertilisation (IVF)** with **pre-implantation genetic testing (PGT)** and only selecting unaffected embryos.
4. Conceive using donor eggs or sperm to avoid passing on the genetic condition.

Each option is a personal decision. This information sheet is intended as a guide only, and you may wish to discuss your options further with a genetic counsellor.

Testing before conception

In-vitro fertilisation (IVF) with pre-implantation genetic testing (PGT)

This method aims to prevent passing on known disease-causing gene changes (also called pathogenic variants) to future children.

Embryos are created in a laboratory, from eggs and sperm from you and your partner. The embryos are then screened for known pathogenic variants. Only unaffected embryos are available for transfer during an IVF cycle.

PGT requires a referral from a genetics service. There are typically out-of-pocket costs, although there are publicly-funded fertility clinics available in Melbourne.

Testing during pregnancy

Prenatal testing

Prenatal testing looks for specific gene changes in your baby, early in the pregnancy. It does not reduce the risk of passing on the genetic condition, but it may inform care and decision-making during a pregnancy.

There are two type of prenatal tests:

- Chorionic villus sampling (CVS) – performed between 11-13 weeks, where a small sample is taken from the placenta
- Amniocentesis – performed from 15 weeks, where a sample of the amniotic fluid that surrounds the baby is collected

Both procedures come with a small risk of miscarriage. Therefore, diagnostic testing is usually only done when there is a clinical need (for example, if you are considering terminating an affected pregnancy).

Prenatal testing can be arranged through the public health system (with few out-of-pocket costs) or through a private provider.

Monash Health · Austin Health · The Royal Melbourne Hospital · The Royal Children's Hospital Melbourne

This information sheet was produced through the Scaling Up Genomics in Nephrology project funded by the Melbourne Genomics Health Alliance. Visit **kidneygenomics.org.au** for more information.

Other screening tests

There are additional screening tests available to the general public. These may look for a number of other genetic conditions during a pregnancy.

Screening tests include:

- reproductive carrier screening
- non-invasive prenatal testing (NIPT)
- ultrasound screening
- first- and second-trimester combined screening

Your GP, obstetrician or a genetics service can discuss these options with you.

Further information about reproductive genetic testing

- You can ask your treating team to arrange for you to speak to a genetic counsellor or genetics service.
- The Centre for Genetics Education, resources under the 'Pregnancy' tab – at **genetics.edu.au**

For more information about public IVF services

- Monash Health Fertility services – at **monashhealth.org**
- The Royal Women's Hospital Public Fertility Care Service – at **thewomens.org.au**

Private options are also available.

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