(CVS) or an amniocentesis. He or she will advise this at the confirmation of a pregnancy, through an ultrasound scan, about five weeks after IVF treatment. The recommended NIPT and other tests can identify pregnancies affected by conditions such as Down syndrome. CVS would be performed at 10-12 weeks and amniocentesis at 15-20 weeks.

Benefits of NIPT, ultrasound scan, CVS or amniocentesis:

- Confirmation of results of genetic testing
- Peace of mind

Potential Risks of CVS or Amniocentisis

- Miscarriage
- Bleeding

- **No normal embryos:** The test may find that none of the embryos are normal, in which case there will be no transfer procedure. The likelihood of this happening depends mainly on the patient's age and the number of eggs retrieved. For women aged over 40, about 10-15% of cycles result in all the embryos being abnormal, which means there is no transfer. For younger patients, the risk of all embryos being abnormal is lower, sometimes significantly, as age is the most important factor affecting a woman's prospects of falling pregnant. However, the quality of the male partner's sperm also has to be taken into account.
- **No diagnosis or partial diagnosis:** Some embryos will have no diagnosis due to the loss of the biopsied cells, or naturally poor DNA quality (often found in damaged or dying cells). Cells can regress as the embryo divides to form a blastocyst. Embryos without a result can still be transferred, at your own discretion, but the benefits of PGS/PGD will not apply in such cases. In this situation, your pregnancy will need to be carefully monitored with ultrasound examination to check the fetus's development.

What are the Alternatives to Genetic Testing?

The alternative to PGS is to undergo an IVF cycle with no genetic testing of the embryos. In this case, standard prenatal testing (CVS, amniocentesis, NIPT and ultrasound examination) for abnormalities would be carried out if a pregnancy resulted from fertility treatment.

These methods will identify pregnancies affected by chromosomal disorders such as Down syndrome, but do not increase the chances of a successful IVF cycle.

Making an Informed Decision

We hope this fact sheet has made you better informed as it is important to understand all aspects of genetic testing before considering undergoing the procedure. It's also worth remembering that general screening is provided for patients seeking extra assurance when they are not aware of any genetic disorder in their family history. However, we cannot guarantee what happens at developmental level.

The risks, benefits and alternatives of such testing should be discussed thoroughly with your genetic counsellor, treating specialist and/or the scientist performing any procedure.

Where to Now?

If you would like to include genetic testing in your treatment, or just want more information, please speak with your specialist or scientist.

Contact Us

Adelaide	1300 483 235
Brisbane City	1800 123 483
Brisbane Southside	1300 483 784
Gold Coast	1300 859 116
Melbourne City	1300 781 483
Melbourne Bundoora	1300 483 232
Sydney	1300 277 447

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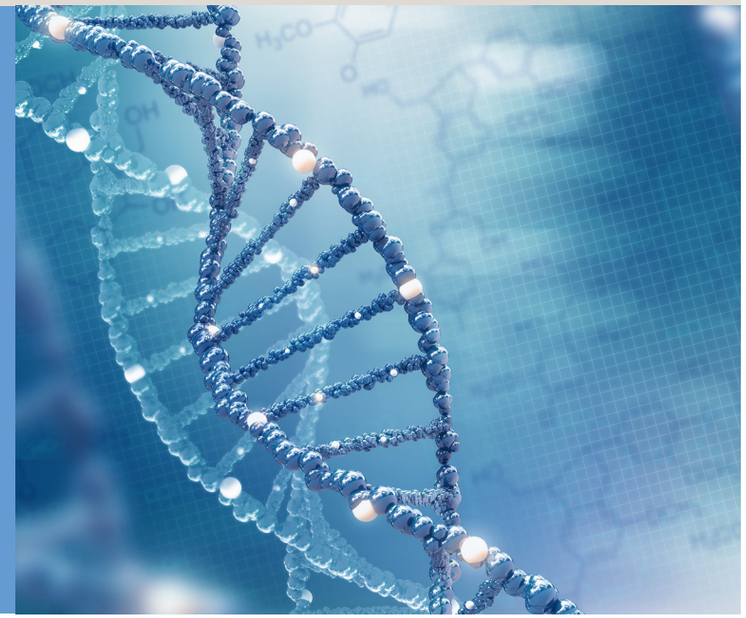
Genetic Testing

What You Need to Know



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What is Genetic Testing and why is it Required?

Genetic testing is important because abnormal chromosomes can stop an embryo from implanting on the uterine lining, or result in a miscarriage or the birth of a child with physical and/or mental disabilities.

Testing can help prevent these outcomes by identifying the affected embryos during their development in the laboratory, before they are transferred back to a patient's uterus during an IVF cycle.

At City Fertility Centre we pride ourselves on offering the latest techniques to give our patients the best care and maximise their chances of falling pregnant.

This approach includes genetic testing of IVF embryos, and this fact sheet is designed to give you a valuable insight into the process, along with its benefits, risks and alternatives.

Types of Genetic Testing

There are two methods of genetic testing:

1. **Preimplantation Genetic Screening (PGS)**
   PGS does not diagnose any specific diseases but looks more generally at the chromosomal make-up of the embryo. If an embryo contains the wrong number of chromosomes (a condition known as aneuploidy), such as when one is missing, then the cells in that embryo are missing a whole set of instructions. If there is an extra chromosome, the cells will malfunction by following those instructions more often than they should. This can result in conditions such as Down syndrome.
2. **Preimplantation Genetic Diagnosis (PGD)**
   PGD is advanced single-gene testing suitable for people who have a family history of a single-gene disorder and are at risk of producing embryos affected by that condition.

   Currently PGD allows testing for disorders including:

- Cystic fibrosis
- Beta-thalassaemia
- Thalassaemia
- Spinal Cerebral Atrophy and Spinal Muscular Atrophy
- BRCA1/BRCA2 (hereditary breast and/or ovarian cancer)

What Does the Testing Involve?

Genetic testing entails analysing an embryo's cells to detect possible chromosomal defects. The procedure follows these steps:

1. Embryos are generated within an IVF cycle, using microinjection, and are grown to the blastocyst stage of development (around day 5-6).
2. Cells called trophectoderm (placental) cells are removed from the blastocyst stage embryo and tests are carried out on those cells to analyse their genetic makeup.
3. The cells are then analysed to verify that all 23 pairs of chromosomes, including X and Y, are identified.
4. Frozen Embryo Transfer (FET): Generally, only one embryo is transferred (in exceptional cases two). During an FET, a cryopreserved embryo is thawed and transferred into the prepared uterus.
5. The remaining "normal'' embryos (already frozen) will be kept in storage for future use.

What Cannot be Tested for?

It is not possible to provide certainty that every disorder can be tested for. The main goal is to first determine whether the embryo is healthy and then plan the next step if anything abnormal is detected.

Who is Genetic Testing Recommended for?

Patients most suited to PGS are those who:

- Have experienced recurrent miscarriage.
- Have had unsuccessful IVF cycles. Please note that PGD/PGS is open to all patients and is offered under clinical guidance depending on your particular circumstances. However, patients can opt for genetic testing from the outset of their treatment.
- Have a family history of chromosomal disorders such as Down syndrome.
- Are of an advanced maternal age (38 or over).
- Want to increase their chance of a successful IVF cycle.

What are the Benefits of Genetic Testing?

PGS and PGD may increase IVF conception rates by helping to identify the embryos most likely to produce a pregnancy, allowing them to be prioritised for transfer.

Other benefits are:

- Supporting elective single-embryo transfer to reduce multiple pregnancies
- Reducing the incidence of miscarriage
- Reducing the risk of a live-born child with a chromosomal abnormality
- Reducing the number of IVF cycles necessary to achieve a successful outcome

What are the Risks?

It's important to keep in mind that adverse results may arise when attempting genetic testing. The chances of these occurring are usually minimal but risks are associated with:

- **Embryo biopsy:** The risk of damaging an embryo during removal of cells is less than 1%. However, if an embryo is damaged, it may not be suitable for transfer into the uterus.
- **The preparation of biopsied cells:** After biopsy, the removed cells are placed in a small test tube and their DNA is amplified (replicated). The cells are no longer viable in any way after this process and can only be used for the tests. Some biopsied cells (less than 2% per cycle) may not yield a test result, or the cells may be lost during transfer to the test tube. Embryos without a result from the analysis may still be transferred, but the advantages of PGS will not apply.
- **Misdiagnosis:** PGS is not 100% accurate and prenatal diagnosis is still recommended following confirmation of pregnancy. However, the risk of a clinical misdiagnosis resulting in a fetus or baby with chromosomal abnormalities after PGS is less than 2%. This is still lower than the risk of having a fetus or baby with chromosomal abnormalities without PGS/PGD. Due to the chance of misdiagnosis, as well as the presence of types of abnormalities that are not tested for, your doctor will strongly recommend a non-invasive prenatal test (NIPT), ultrasound scan, Chorionic Villus Sampling