Genetic Testing (PGD)

The method of genetic screening used in In Vitro Fertilisation (IVF) is Pre-implantation Genetic Diagnosis **(PGD)**.

PGD: PGD is generally used to detect anomalies in the number or structure of the chromosomes of embryos.

When embryos have abnormal chromosomes, this may prevent implantation to the uterine lining, lead to pregnancy loss, or result in the birth of a child with physical and/or mental disabilities.

PGD may help prevent these adverse outcomes by identifying affected embryos as they are developing in the laboratory prior to being transferred back to the patient's uterus during the IVF cycle.

Aneuploidy

Normal human cells (embryonic cells) contain 46 chromosomes in 23 pairs. We receive 23 chromosomes from each parent.

If an error occurs, and the egg or sperm has an extra or missing chromosome, the embryo created will also have an extra or a missing chromosome resulting in a condition called aneuploidy.

The most common aneuploidy is Down syndrome or Trisomy 21 (three 21 chromosomes). Other common aneuploidies are Klinefelter syndrome, Trisomy 13 and Trisomy 18.

A) Genetic Screening

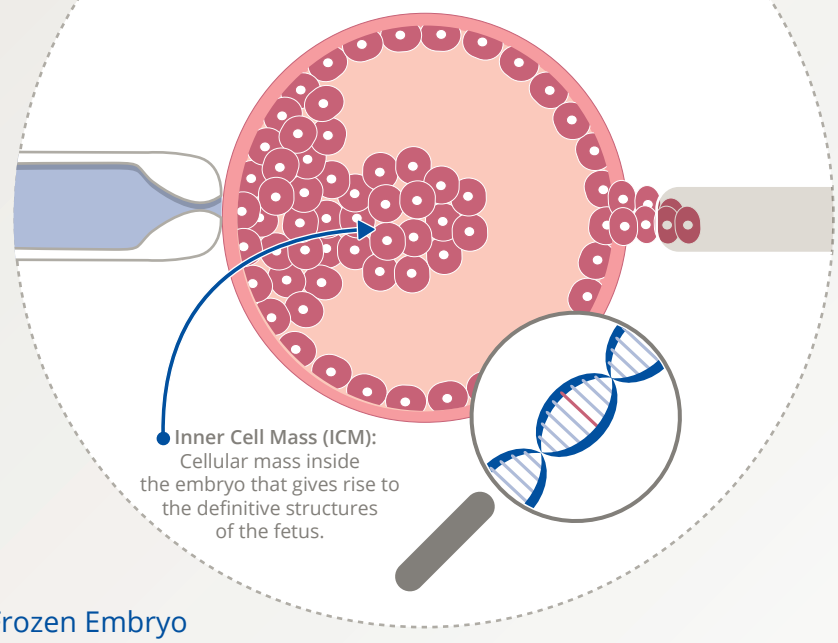
The embryo created through IVF is tested at the blastocyst stage of development. The genetic makeup of the embryo is analysed to detect possible genetic defects. The genetic screening can be performed either on the eggs or on the embryos. The PGD team of doctors, geneticists and embryologists will decide which one will be analysed.

B) Embryo Biopsy

To test an embryo, some embryonic cells are removed via a microscopic opening made in the outer shell of the embryo. The embryo is then frozen and kept in storage while the cells are analysed.

Embryo Biopsy

The embryo biopsy is performed leaving the embryo largely intact and without loss of Inner Cell Mass (ICM).



Inner Cell Mass (ICM): Cellular mass inside the embryo that gives rise to the definitive structures of the fetus.

Advanced Embryo Selection (AES) using array Comparative Genomic Hybridisation (CGH)

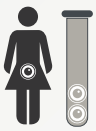
During CGH testing all chromosomes including X and Y are analysed and compared to a sample of 'normal' DNA.

A computerised scanner interprets the results of the slides which are then verified by a trained scientist.

The CGH test can detect chromosomal genetic disorders such as Down syndrome and Edwards syndrome.

C) Frozen Embryo Transfer Cycle (FET)

The embryo chosen is transferred. Generally, only one embryo is transferred, in exceptional cases two. During a Frozen Embryo Transfer Cycle, a cryopreserved embryo is thawed and transferred into the appropriately prepared uterus.



D) Embryo Storage

The remaining 'normal' embryos (already frozen) will remain in storage for future use.

Who is CGH recommended for?

Patients with:

- Recurrent miscarriage.
- Advanced maternal age.
- A family history of chromosomal disorders.
- Previous unsuccessful IVF cycles.
- Any IVF patient wishing to increase their chance of a successful cycle.

What are the potential benefits?

- Reducing the risk of a liveborn child with a chromosomal abnormality.
- Improving IVF rates by prioritising chromosomally 'normal' embryos for transfer.
- Reducing the incidence of miscarriage.
- Reducing the number of IVF cycles necessary to achieve a successful outcome.
- Supporting elective single-embryo transfer to reduce multiple pregnancies.

ML66 0216 0216

1300 354 354 | cityfertility.com.au

City Fertility Centre
your partners in life