Preimplantation genetic testing (PGT)

Preimplantation genetic testing (PGT) refers to a range of techniques that are used to test high risk embryos for gene disorders or chromosomal abnormalities.

PGT is not part of a routine assisted reproductive technology treatment cycle and may only be approved in certain circumstances.

The Reproductive Technology Council (external site) may approve PGT if:

- the embryo is for use in the reproductive treatment of a woman; and
- based on existing scientific and medical knowledge:
  - the procedure is unlikely to leave the embryo unfit for implantation; and
  - where, for genetic testing, there is a significant risk of serious genetic abnormality or disease being present in the embryo.

Licensed fertility providers must ensure that a separate consent is obtained for any PGT procedure that requires the approval of the Reproductive Technology Council.

Preimplantation genetic testing for aneuploidy (PGT-A)

Preimplantation genetic testing for aneuploidy (PGT-A) was previously known as preimplantation genetic screening (PGS).

A genetic test that checks embryonic cells collected via in vitro fertilisation (IVF) to assess for chromosomal abnormality and identify embryos that may fail to implant, lead to miscarriage or result in a child with a chromosomal abnormality.

PGT-A aims to identify embryos that are:

- Euploid: with the correct number of chromosomes
- Aneuploid: with missing or additional chromosomes
- Mosaic: with both euploid and aneuploid cells.

PGT-A may increase the chance of embryo implantation and reduce the likelihood of miscarriage or birth of a baby with a chromosomal abnormality.

PGT-A is an IVF procedure and will incur out of pocket costs over and above a routine cycle of fertility treatment. There is limited evidence of the effectiveness of PGT-A in general use. There is some evidence that use in older women may reduce miscarriages, though this has not been shown to increase the number of births.

Approval for PGT-A is required from the Reproductive Technology Council (external site). The Reproductive Technology Council has issued a blanket approval for PGT-A where the woman providing the eggs:

- is greater than 35 years of age;
- has experienced two or more miscarriages;
- has experienced two or more failed IVF attempts after embryos transfer; or
- has been referred by a geneticist and has a family history of aneuploidy.

Preimplantation genetic testing for monogenic disorders (PGT-M)

A genetic test that checks embryonic cells collected via in vitro fertilisation (IVF) to identify embryos with a specific genetic condition that affects a single gene. PGT-M may be used when the parents are known carriers of a genetic condition. In these cases, the embryos are tested for the specific mutations carried by the parents. Carrier embryos or embryos without the genetic mutation are then considered for transfer. PGT-M does not screen embryos for a panel of genetic conditions.

PGT-M may be appropriate when:

- a person is affected by a genetic condition;
- a woman is a carrier of an X-linked condition; or
- a person and their partner or donor are both carriers of the same genetic condition.

The Reproductive Technology Council (external site) has considered that conditions suitable for PGT-M include Huntington's Disease, cystic fibrosis, sickle cell anaemia and Duchenne Muscular Dystrophy. Other conditions may be tested for on a case by case basis.

PGT-M may reduce the risk of miscarriage or the likelihood of the birth of a baby with the specific genetic condition.

Approval for the procedure is required from the Reproductive Technology Council. Support from a clinical geneticist or genetic counsellor is required.Â The geneticist or counsellor must have assessed the risk and seriousness of the condition.

Preimplantation genetic testing for structural rearrangements (PGT-SR)

A genetic test that checks embryonic cells collected via in vitro fertilisation (IVF) to assess for chromosomal structural rearrangements and identify embryos that may be unsuitable for use in assisted reproductive procedures.

Chromosomal structural rearrangements include:

- dislocations;
- translocations;
- inversions;
- deletion;
- duplications.

If the chromosomal set is complete the structural rearrangement is known as balanced. Where there is missing or extra chromosomal information the structural rearrangement is known as unbalanced.

Chromosomal structural rearrangements (particularly deletions and duplications) can cause significant genetic abnormalities or disease. However, many balanced structural rearrangements do not have an immediate effect but can impact on the carrier's ability to conceive.

PGT-SR may be used where there is a known structural relocation in the gamete provider.

Approval for PGT-SR is required from the Reproductive Technology Council (external site).

Saviour siblings

The term "Saviour sibling" refers to the use of preimplantation genetic testing (PGT) in conjunction with human leukocyte antigen typing to select an embryo, created through IVF, that is able to provide tissue or stem cells to treat a life threatening medical condition in a sibling.

In Western Australia there is no specific provision that permits access to IVF or PGT for a family that is seeking to have a child that can act as a saviour sibling.

To access to PGT for this purpose a person must have:

- a medical or genetic reason to access IVF; and
- a risk of genetic disease in the embryo.

Approval of the Reproductive Technology Council (external site) is required to access PGT for this purpose.

Sex selection

"Sex selection" in assisted reproductive technology refers to a genetic test that checks embryonic cells, collected via IVF, to identify embryos of a particular sex. Embryos of the desired sex are then considered for transfer.

Sex selection for non-medical reasons is prohibited in Western Australia.

Sex selection may only be approved to avoid the transmission of a sex-linked genetic abnormality or disease.

More information

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