

This Fact Sheet describes a pre-pregnancy testing option called pre-implantation genetic diagnosis (PGD). With the use of *in vitro* fertilisation (IVF), PGD is a diagnostic test performed on an embryo prior to implantation in the uterus.

### In summary

- PGD involves testing for certain genetic conditions in an embryo created using *in vitro* fertilisation (IVF) prior to transferring it to the uterus and allowing it to develop normally
- Only those embryos that do not have the specific genetic condition that was tested for will be transferred into the woman's uterus
- In Australia, PGD is currently only offered in the private health setting.

### WHAT IS PRE-IMPLANTATION GENETIC DIAGNOSIS (PGD)?

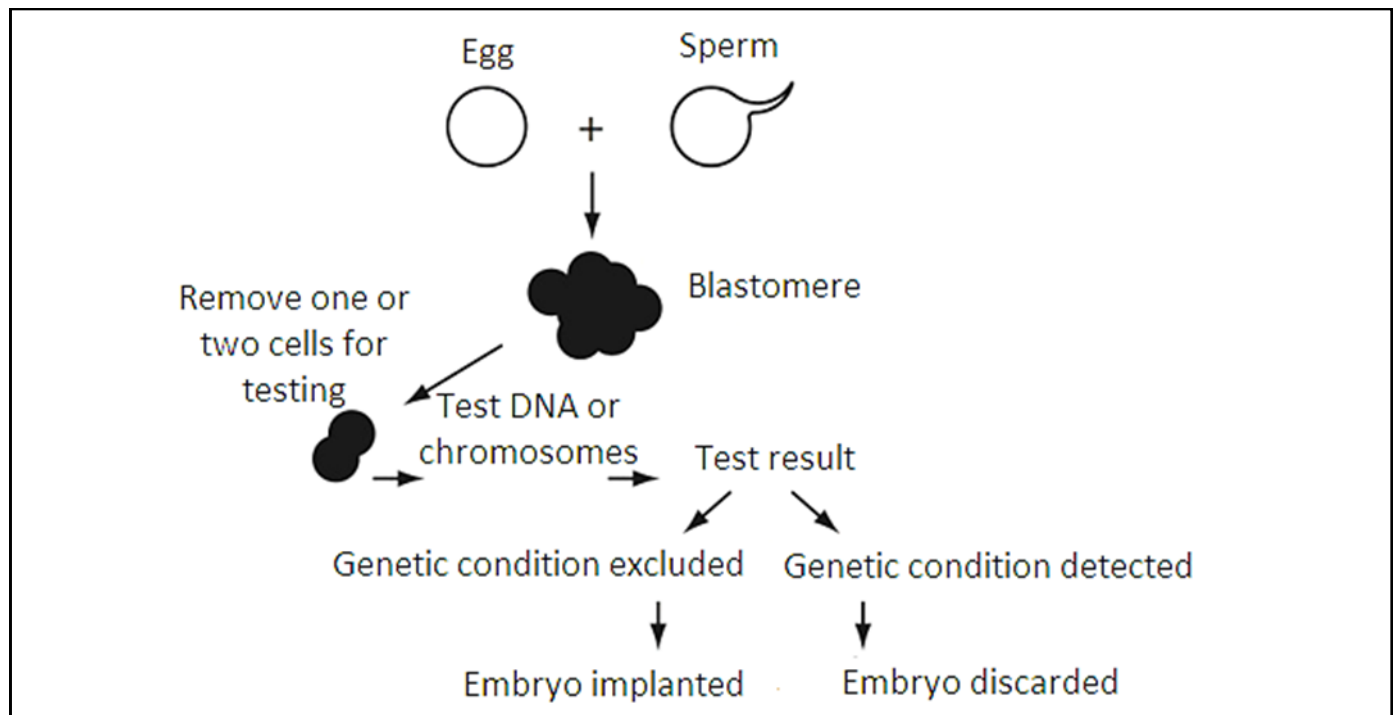
Pre-implantation genetic diagnosis (PGD) is a technique that is currently only available in the private setting. It is a very specialised technique that can help couples who are at increased risk of having a child with a genetic condition avoid doing so without the need for decisions regarding termination of an affected pregnancy.

PGD involves testing an embryo that has been created using *in vitro* fertilisation (IVF), prior to transferring it to the uterus and allowing it to develop normally.

### HOW IS PGD PERFORMED?

Hormones are given to the woman to stimulate her ovaries and enable the collection of a number of eggs (called oocytes). After the eggs are removed, the eggs are fertilised in the laboratory with sperm. Those eggs that are successfully fertilised divide and multiply to form a developing embryo called a blastomere. After three to five days, one or two cells are removed in order to test for the specific genetic condition in question. The removal of these cells does not appear to harm the developing embryo (*Figure 29.1*).

Only those embryos that do not have the specific genetic condition tested for will be transferred into the woman's uterus.



**Figure 29.1:** The PGD process. In some laboratories, the embryo is allowed to grow for up to five days so that cells are removed from the blastocyst, rather than the blastomere

Usually, no more than one embryo will be transferred to the uterus at any one time to avoid the possibility of multiple births.

In some IVF units, unaffected embryos that are not used can be frozen for transfer in another cycle.

#### **WHAT ARE THE ADVANTAGES AND DISADVANTAGES OF PGD?**

Success rates for having a child from an IVF cycle followed by PGD vary from IVF centre to centre but tend to follow standard IVF success rates. Therefore a pregnancy and an unaffected child cannot be guaranteed using this technique

Like any IVF procedure, stress and often disappointment can accompany PGD. Couples will need to balance the financial and emotional cost

of the IVF procedure followed by PGD with that of termination of an affected child conceived naturally.

For couples with a moral or religious objection to pregnancy termination and who also have a risk of having a child with a genetic condition, this technique may provide the opportunity to have an unaffected child.

In others, PGD may be a preferred option over prenatal testing in a naturally conceived pregnancy. It can also eliminate the possibility of repeated miscarriages for couples where one partner carries a chromosomal translocation.

In Australia, PGD is currently only offered in the private setting. Genetic counselling is important before considering PGD.