Fact sheet **29**

PRE-IMPLANTATION GENETIC DIAGNOSIS (PGD)

This fact sheet talks about a pre-pregnancy testing option called preimplantation genetic diagnosis (PGD). Using *in vitro* fertilisation (IVF), PGD is a genetic test performed on a developing embryo before a pregnancy starts.

IN SUMMARY

- PGD involves testing for specific genetic condition/s in an embryo made using *in vitro* fertilisation (IVF), before transfer to the uterus (womb) and allowing a pregnancy to develop in the usual way
- Only those embryos that do not have the specific genetic condition that was tested for will be transferred to try for an ongoing pregnancy
- PGS (**P**reimplantation **g**enetic **s**creening) is often used by couples who are older, have experienced recurrent miscarriage or have had a number of failed embryo transfers following IVF treatment. PGS is not checking for a specific genetic condition, but rather screens embryos to ensure that the number of chromosomes are balanced.

WHAT IS PRE-IMPLANTATION GENETIC DIAGNOSIS (PGD)?

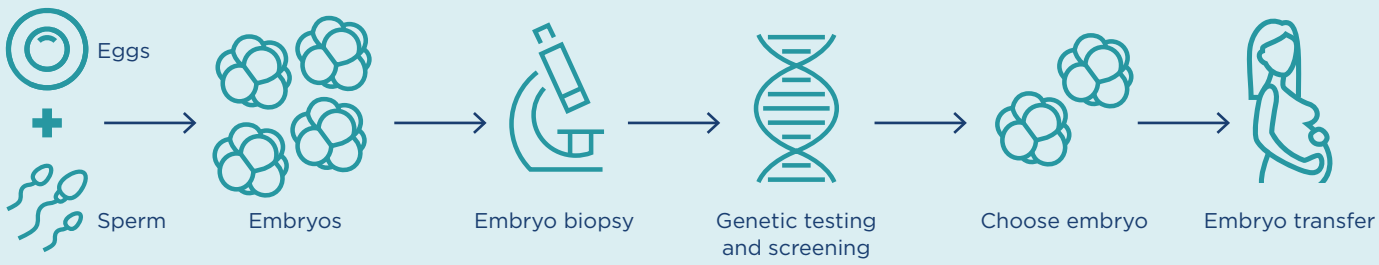
Preimplantation genetic diagnosis (PGD) is a specialised technique that can help couples who have an increased chance of having a child with a genetic condition. Some women and their partners choose have PGD with IVF rather than trying for a natural pregnancy with the option of genetic testing of an ongoing pregnancy.

HOW IS PGD PERFORMED?

Hormones are given to the woman to stimulate her ovaries and allow the collection of a number of her eggs (called oocytes). After the eggs are removed, they are fertilised in the laboratory with sperm. Those eggs that are successfully fertilised divide and multiply to form a developing embryo. When the embryo is at the blastocyst stage, a few cells are removed to test for the specific genetic condition in question. In general, the removal of these cells does not appear to harm the developing embryo (*Figure 29.1*).

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

Figure 29.1:
The PGD process. The embryo allowed to grow for five days and a few cells are removed from the blastocyst



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WHAT ARE THE ADVANTAGES AND DISADVANTAGES OF PGD?

Genetic counselling is important when considering all the options for planning a pregnancy. The advantages and disadvantages of each option are complex, and vary depending on the individual situation for a couple.

The option of IVF and PGD allows couples to reduce the chance of having a child with a specific genetic condition without having to face the difficult decision of whether to stop a naturally conceived pregnancy that has been shown to be affected by testing during pregnancy.

IVF/PGD can also reduce the chance of repeated miscarriages for couples where for example one partner carries a chromosome translocation.

As with any IVF procedure, stress and sometimes disappointment can arise when undertaking the PGD process. Success rates for having a child from an IVF cycle followed by PGD vary from IVF centre to centre (these are available from the individual IVF provider) but tend to follow standard IVF success rates.

Couples may balance the financial and emotional cost of the IVF procedure followed by PGD, with that of considering termination of pregnancy or continuing a pregnancy for a child with a genetic condition conceived naturally.

In preparation for pregnancy, reproductive genetic carrier screening is also available for women and their partners. This test screens for whether there is an increased chance for other recessive genetic conditions in pregnancy.

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