PREIMPLANTATION GENETIC DIAGNOSIS

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Important points

- While there are a number of different prenatal tests and procedures available to diagnose the development of a baby, another option where there is a risk that the baby will have a genetic condition is preimplantation genetic diagnosis (PGD)
- PGD involves testing for certain genetic conditions in an embryo created using assisted reproductive technologies (ART) such as *in vitro* fertilisation (IVF), prior to transferring it to the uterus and allowing it to develop normally
- After hormonal stimulation of the woman's ovaries, some eggs are removed and then fertilised in the laboratory with sperm
- One to two cells are removed from the embryo at the eight cell stage (after 3 days) or at blastocyst stage (after 5 days), for testing
- Only those embryos that do not have the specific genetic condition that was tested for will be transplanted into the woman's uterus
- Usually, no more than one or two embryos will be transferred to the uterus at any one time to avoid the possibility of multiple births (more than one baby in a pregnancy)
- Success rates for having a child from an IVF cycle followed by PGD varies from IVF centre to centre but tend to follow standard IVF success rates
- Like any IVF procedure, stress and often disappointment can accompany PGD. Couples will need to balance the financial and emotional burden of the IVF procedure followed by PGD with that of termination of an affected child conceived naturally
- In Australia, PGD is currently only offered in the private setting

Every woman hopes for a healthy baby. In some cases, the baby may have either a serious physical or intellectual problem.

There are a number of different prenatal (meaning before birth) tests and procedures available to diagnose the development of the baby. Each has advantages, disadvantages and limitations.

Prenatal diagnostic tests include:

1. Ultrasound (see Genetics Fact Sheet 17A)
2. Chorionic villus sampling (usually simply called CVS) (see Genetics Fact Sheet 17C)
3. Amniocentesis (see Genetics Fact Sheet 17C)
4. Cordocentesis (see Genetics Fact Sheet 17C)

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

What is Preimplantation Genetic Diagnosis (PGD)?

Preimplantation genetic diagnosis (PGD) was first reported in 1989.

It is a very specialised technique that can help couples who are at 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 assisted reproductive technology (ART) such as *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 or 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, the developing embryo contains either about eight cells (after three days) or is a blastocyst (after five days) (*Figure 18.1*)

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.

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

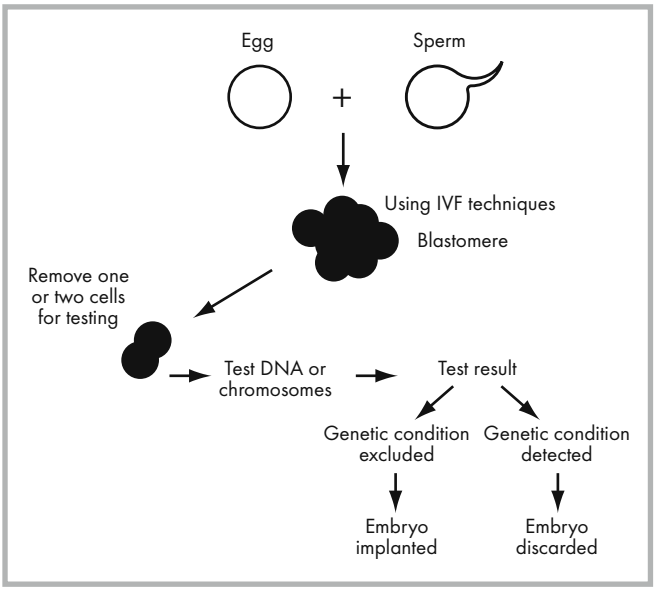

Figure 18.1. *The PGD process. In some laboratories, the embryo is allowed to grow for up to five days so that cells are removed from the blastocyst, rather than the blastomere.*

on the same day, or, in some IVF units, on day five of development.

Usually, no more than one or two embryos 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.

It is important for an IVF unit to have a high pregnancy rate to be able to offer reliable preimplantation genetic diagnosis (PGD) with a real chance of achieving pregnancy.

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 (see Genetics Fact Sheet 3) is important before considering PGD.

Other Genetics Fact Sheets referred to in this Fact Sheet: 3, 17A, 17C

Information in this Fact Sheet is sourced from:

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