

# Monash IVF Fact Sheet

## Preimplantation Genetic Diagnosis (PGD)



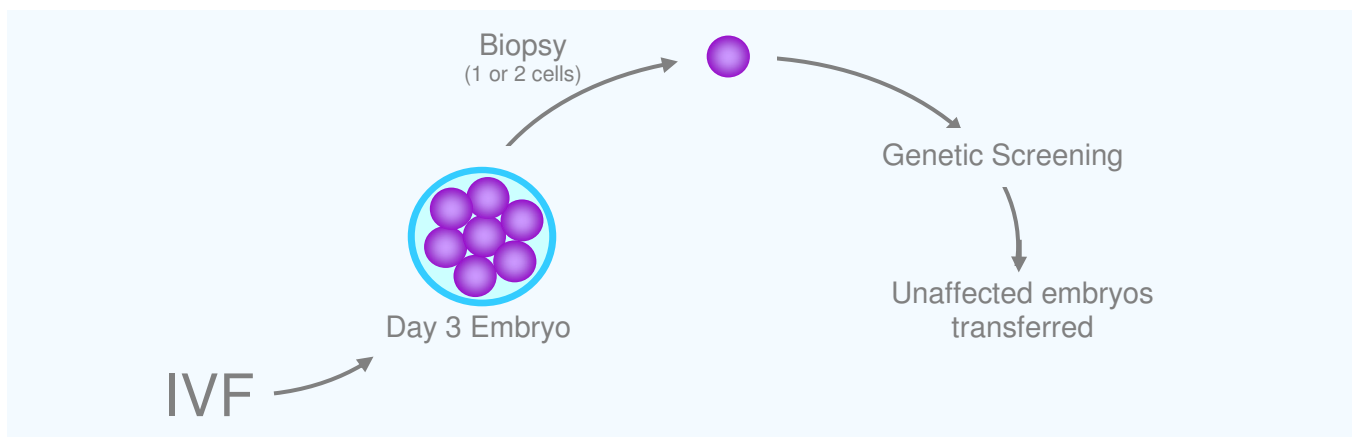
### Key points:

- PGD involves screening in vitro fertilization (IVF) generated embryos for genetic conditions prior to embryo transfer.
- PGD is available for single gene disorders, translocations, chromosomal aneuploidy or X-linked disorders.
- Only embryos that are unaffected will be considered suitable for transfer. Embryos that are affected or have a chromosome abnormality will not be considered suitable for transfer.
- PGD is NOT 100% accurate. Confirmatory prenatal diagnosis is highly recommended if a pregnancy is achieved following PGD.

### What is preimplantation genetic diagnosis (PGD)?

PGD is a reproductive option for couples at risk of passing a specific genetic disease or chromosome imbalance to their children. PGD involves screening IVF generated embryos for genetic conditions prior to embryo transfer, with only unaffected embryos transferred to the uterus. This provides the opportunity to screen embryos for genetic conditions before a pregnancy is established (Figure 1).

Figure 1: Preimplantation Genetic Diagnosis



Couples choose PGD over prenatal diagnosis for many reasons including objection to termination of pregnancy, loss of a child from the genetic disease, recurrent implantation failure or recurrent miscarriage.

PGD testing may be appropriate for:

- Couples at risk of passing a single gene disorder on to their children
- Couples at risk of having children with a particular X-linked disorder
- Couples where one partner carries a balanced chromosomal rearrangement
- Couples where one partner has an altered sex chromosome complement (eg: XXY)
- Couples with advanced maternal age (>36 years of age)
- Couples who have experienced recurrent implantation failure
- Couples who have experienced recurrent miscarriage
- Couples who have previously had a pregnancy with a chromosomal abnormality

Genetic counselling is an important step to ensure that PGD is the right option for each couple.

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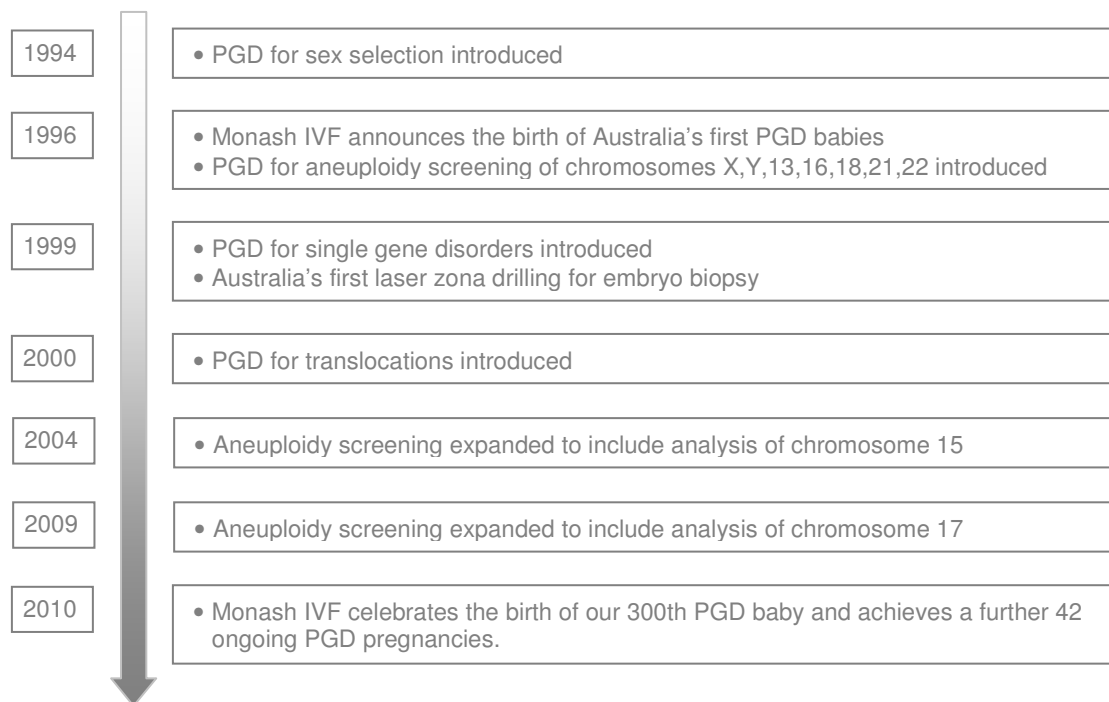


### PGD at Monash IVF

Monash IVF has offered PGD since 1994 and is one of the few centres in Australia that specialises in this area of reproductive medicine. All of our PGD testing is performed in house at Monash IVF Clayton by a highly specialised genetics team, ensuring the highest quality of care to patients. The genetics team is responsible for providing a specialised PGD service not only to our own patients but also to patients undergoing IVF cycles at numerous IVF clinics throughout Australia and New Zealand. While the main PGD laboratory is located at Clayton in Melbourne, Australia, embryo biopsy can be performed away from the PGD laboratory and the embryonic cells (called blastomeres) sent by courier to Clayton. Centralising the PGD testing enables clients to access the highest levels of expertise without having to leave their home state.

Monash IVF has consistently been at the forefront of PGD technology. Some of our milestones are listed in Figure 2.

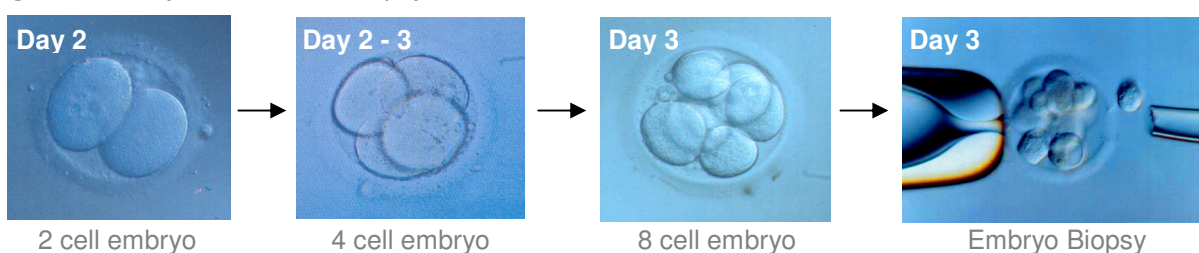
Figure 2: Key PGD milestones at Monash IVF



### What does PGD involve?

All couples requesting PGD must first undertake an IVF cycle to stimulate the woman's ovaries to produce a number of eggs. These eggs are collected and fertilised using the male partner's sperm. The resulting embryos are cultured in the laboratory. Embryo biopsy is performed on Day 3 after egg collection (Figure 3). Embryos that have developed to at least 5 cells are suitable for biopsy. A hole is drilled in the outer shell of the embryo and 1 or 2 cells are removed for genetic analysis.

Figure 3: Embryo Culture and Biopsy



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The embryos are kept in culture while the testing of the biopsied cells proceeds. Genetic test results are usually obtained within 24 hours of embryo biopsy. A qualified PGD scientist will discuss the PGD results with the couple at the completion of testing. When available, one or two unaffected embryos can be transferred on Day 4 or Day 5. When a number of embryos are identified as being genetically suitable for transfer, morphological criteria are used to determine the best embryo/s for transfer. The couple's IVF nurse will organise a pregnancy test to be performed on Day 16.

Surplus unaffected embryos will be grown in culture to Day 5 or Day 6. If they reach an appropriate stage of development (ie: form a good quality blastocyst) they will be frozen. These embryos may be used in a subsequent IVF cycle if the couple do not achieve a pregnancy with the fresh embryo/s.

Embryos that are affected or have a chromosome abnormality are discarded or donated to research with the couple's consent.

#### Accuracy of diagnosis

Couples should be aware that the results obtained from PGD are NOT 100% accurate. At best the accuracy of the test is approximately 90% if PGD has been performed for chromosome screening or up to 98% if PGD has been performed for a single gene disorder. Consequently, there may be up to a 10% error rate associated with any test.

Many factors influence the accuracy of PGD testing including:

- The type of PGD test being performed.
- The cell biopsied from the embryo. It is possible that the cell biopsied is NOT representative of the entire embryo. This may lead to a misdiagnosis.
- The number of cells biopsied from the embryo. The accuracy is higher if results are obtained from two cells rather than one cell.
- The clarity of results obtained. Some PGD results are more difficult to interpret than others. If the results are not clear a reduced accuracy may be reported.

Every effort is made to ensure that the PGD test offered has the highest possible accuracy using the currently available technology. However, given that PGD is not 100% accurate, **confirmatory prenatal diagnosis is highly recommended.**

#### Other important information

- **Embryo biopsy does not appear to affect embryo development.**  
Embryo biopsy has been performed extensively at Monash IVF and other major IVF clinics throughout the world. Follow up studies at Monash IVF have shown that the procedure is safe and does not appear to have any adverse affect on the embryo's potential to implant and develop normally.
- **Only embryos that are good quality and adequately developed can be biopsied.**  
Embryos must have at least 5 cells by Day 3 to be suitable for biopsy (ideally they should have 6 to 8 cells on Day 3). One or two cells will be biopsied from the embryo depending on the number of cells in the embryo and the type of testing required.
- **It may not be possible to obtain a result from every embryo.**  
Due to the complexity of the genetic tests, an inconclusive result may be obtained for some or all embryos. In this case, the embryo/s will be grown in culture to Day 5 or Day 6. If the embryo/s reach an appropriate stage of development (ie: form a blastocyst) they will be frozen. In some situations it may be possible to perform a second biopsy procedure (called blastocyst biopsy) on these embryos to try to obtain a conclusive result. If this is possible and unaffected embryos are identified, these can be transferred in a frozen embryo transfer cycle.

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- **PGD only tests for the condition of interest.**

PGD tests are specifically designed to detect genetic abnormalities related to the clinical indication for referral (ie: single gene disorder, translocation, aneuploidy screening or sex selection). The test does not give any information relating to other genetic conditions or abnormalities.

#### What are the costs?

Information relating to the cost of PGD is available from Monash IVF. Please note that additional costs will be incurred for IVF.

#### How to get started

Those interested in PGD should discuss this procedure with their IVF doctor or with a member of the genetics team at Monash IVF. Genetic counselling may help couples decide whether or not PGD is the right option for them.

Monash IVF offers a PGD clinic to couples at genetic risk. The PGD clinic is a free service that involves a one-on-one consultation with a clinical geneticist and/or genetic counsellor. During this one hour appointment the genetic specialists will:

- Thoroughly review the genetic history.
- Provide the couple with information regarding the PGD process at Monash IVF.
- Answer any questions the couple have in relation to their specific genetic condition and the PGD process.
- Arrange for any further clinical and DNA testing to confirm genetic status.
- Arrange for collection of blood samples for test development (if appropriate).
- Offer guidance and support to alleviate any anxiety.

Following counselling the couple should be aware of the relative risks of embryo screening and possible outcomes. This will place them in a sound position to make an informed decision about PGD. To make an appointment or to obtain further information regarding the PGD clinic at Monash IVF, please contact our genetic counsellor on +61 03 9590 8336.

#### Quality systems

Monash IVF employs a very high standard of quality assurance. Through the application of quality systems the laboratory provides standards of excellence in quality service, care and advice.