Preimplantation Genetic Diagnosis (PGD) & Screening (PGS) with IVF

Embryos can be tested for a serious genetic disorder using PGD, or they can be screened for the correct number of chromosomes using PGS. Only normal embryos are transferred.

Who can have PGD?
In New Zealand PGD is permitted for:
- Serious conditions where a child has at least a 1 in 4 chance of inheriting the condition.
- Chromosome abnormalities associated with recurrent miscarriage or advanced maternal age.
- Ethics Committee approval is required for other reasons, such as ‘saviour siblings’.
PGD can’t be used for social reasons such as gender selection in New Zealand.

Who can have PGS?
Anyone having IVF or ICSI can consider having PGS. PGS is usually not possible if you are also having PGD, since different techniques are used to analyse the embryo’s DNA. PGS may be more suitable for:
- People with many good quality embryos, to provide extra embryo selection.
- Women aged 36 and older, because the chance of having the wrong number of chromosomes in an embryo increases from the age of 36.

How does PGD work?
People wanting PGD undergo IVF in the usual way. On day 3 of embryo development, a small hole is made in the soft shell of the embryo (called the Zona Pellucida) using a laser. One or more cells are gently removed from each embryo, usually on day 5 (called embryo biopsy), and then the embryos are frozen. The cells are sent to a PGD laboratory for analysis. We commonly use Canterbury Health Laboratories in Christchurch or Monash IVF in Melbourne. The PGD lab tests the cells from each embryo and tells us which embryos are normal. Normal embryos can be thawed and transferred to the woman later.

When is PGD used?
PGD is used by people who have a serious genetic disorder that could be passed on to their children. Disorders can be divided into two types:
- Single gene defects. These disorders are caused by a change in a single gene. Examples include Cystic Fibrosis, Huntington’s Disease, Beta-thalessemia, Fragile X and Spinal Muscular Atrophy.
- Chromosome abnormalities. These arise from either having the wrong number of chromosomes, such as trisomy 21 which causes Down Syndrome, or having rearrangements of parts of chromosomes, called translocations.

Getting prepared for PGD
In addition to getting prepared for IVF treatment itself, there are some extra steps when you are using PGD.
• **Genetic counselling** Genetic counselling is offered by the Regional Genetic Services. It covers the pattern of inheritance of the family’s genetic condition, the chance of a child inheriting the condition, the impact of the condition on a child and their family, the alternatives to PGD and the implications of using PGD.

• **Clinic counselling** You will also need to see a Fertility Associates counsellor before PGD treatment to explain the issues that accompany IVF treatment.

• **Feasibility studies** For most disorders, the PGD lab will want to check that they can reliably detect the disorder for the particular family. This is called a feasibility study. It involves sending blood samples from the man and woman, and sometimes from other close family members, to do a trial run using blood cells instead of embryo cells. It may take several months for the PGD lab to identify suitable genetic markers for the family.

• **Planning ahead** Treatment is usually planned 2–3 months ahead to give the PGD lab time to buy and test the genetic markers.

• **Consent** In addition to the standard consent for IVF, there is a Fertility Associates consent form for PGD, and a consent form from the PGD laboratory too.

• **Paying for PGD** Most PGD is publicly funded, covering up to 2 cycles of treatment. We will advise you of the cost well ahead if you are having private treatment. If treatment is stopped before PGD, the PGD labs usually charge a cancellation fee to cover the costs of their materials.

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**PGS & PGD risks and problems**

• **Embryos unsuitable for biopsy** Some embryos may not be suitable for embryo biopsy because they do not reach the right stage of development. These embryos are very likely to be abnormal.

• **Embryo damage** About 1–2% of embryos are damaged during embryo biopsy.

• **Transport problems** It is possible that samples may be lost or damaged during transport to the PGD lab. Fertility Associates takes responsibility for the steps that take place in our clinic. The courier is responsible for the transport of cells, and the PGD lab for the PGD analysis and giving results. Our fees do not cover any insurance to cover loss or delay of cells during transport. We can put you in touch with the courier company if you wish to consider insurance.

• **No result or an inconclusive result** Sometimes the enzyme and chemical reactions in PGD or PGS testing do not occur as expected. About 5–10% of embryos have an inconclusive result.

• **Wrong diagnosis** Although PGD/PGS laboratories use clever strategies to reduce the chance of misdiagnosis, PGD and PGS is only 95% accurate. Consequently, we strongly advise people to follow up PGD and PGS with pre-natal diagnosis using CVS or Amniocentesis when they become pregnant. The chance of misdiagnosis by CVS or Amniocentesis is much lower because many more cells are tested. Prenatal diagnosis is about 99% accurate.

• **Correct diagnosis, but unrepresentative cells** Sometimes some cells in the embryo have the right number of chromosomes while other cells have an abnormal number. When this happens, the cells biopsied may not represent the cells left behind.

• **Correct diagnosis, but other abnormalities** PGD only tests for the particular genetic disorder, and PGS only screens for the correct number of chromosomes. It is possible for an embryo to have other genetic abnormalities.

• **No normal embryos** Sometimes all the embryos tested are abnormal so there are no embryos available for use.

• **Biopsied embryos stop developing** Some embryos may be normal but stop developing and are therefore not suitable for freezing or transfer. It is possible that this could happen to all the embryos.

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**When is PGS used?**

PGS is used to screen embryos created during routine IVF treatment. Many embryos look normal but have the wrong number of chromosomes (called aneuploidy). PGS offers a way of better selecting which embryo to transfer. Embryos with the wrong number of chromosomes cannot give rise to a normal pregnancy.

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**Getting prepared for PGS**

You need to decide whether to use PGS when you start your treatment cycle. No special preparation is needed. All PGS is privately funded. In addition to the standard consent for IVF, there is a Fertility Associates consent form for PGS, and a consent form from the PGS lab too.
WE FEEL LIKE such an ordinary couple, we met mid 20’s and just followed the pattern all our friends were following. Marriage, a year overseas and home to buy our home – a small first home. Then we began to save to have a child.

It was fun trying for a child … for a while, but when nothing happened the fun went out of it. Finally we decided to go to our GP who was very nice and did some tests. Nothing showed so we were sent over to Hamilton Fertility Associates.

I remember that first time sitting in the waiting room feeling so scared as we watched all these people coming in to wait and see the doctor and others with labels walking through. We did not know then but we were going to get to know the labeled ones quite well. The wait took forever. I know doctors always give the person with them the time but we were about 30 minutes late and by the time we went in were really tense. The doctor said we fell in the group of ‘unexplained infertility’. That felt so bad, if you don’t know the problem how can you fix it? Well, it got worse – not only that, but we hadn’t been trying for long enough to qualify for public funding. Anyway we were to do some more tests and if that changed it the clinic would let us know.

More day 2 blood tests, then day 21 – I was just a pincushion. Another trip to Hamilton for a semen analysis – at least that was not my turn. And then a phone call from the doctor, the sperm had enough antibodies (like little caps) that it was going to be hard for us to get pregnant ourselves.

It changed our scoring, public funding, hurray, and we would get a letter telling us what next.

Sometimes 2 weeks of waiting for a letter is forever. If we knew about waiting what we do now we would have relaxed but our lives seemed to be around the letterbox. The letter arrived and we could plan for an IVF cycle with the sperm injection. Wonderful and of course we would be one of the lucky ones, we had had our share of bad luck.

It was really scary going to Hamilton to collect drugs and have counselling. Of course we didn’t need counselling except the nurses really gave us a push. Thank goodness as we did need to sit down and talk about it all and that was our chance. We learnt a lot that day from the nurses and counsellor and met others at the education group. I am not sure how many couples go to the clinic but there are always different people in the waiting room and it is only at the blood tests you see the same people. Anyway I joined the group in our area. We meet each month, its mostly women in our group and we have a good gossip, sometimes we go for a walk or do something else. It’s always good for me to do this, as women need to talk about things.

That cycle didn’t work, so we had to use our frozen embryos. We read the book and it said a low chance of getting pregnant with frozen embryos so we were not too hopeful. 1st Frozen, 2nd Frozen, last Frozen…

We got a positive test, surely now things would be okay. We were so excited we told the world! We really wished we hadn’t when two weeks later we began to bleed and miscarried. Our emotions were all over the place. On one hand we knew we could get pregnant, on the other we would have to go through it all again. AND it was almost a repeat. We had a lot of trips over to Hamilton, firstly for the IVF cycle, and then to have the fresh embryo put back, then a review and then a frozen cycle. We are pregnant again, from a frozen embryo and the difference this time is we have two embryos remaining still, just in case.

I don’t think we will tell anyone this time until the 3 months is up, it’s too hard untelling them. We don’t plan or talk about it too much as even that feels scary. We do tick off every day and feel glad about another day without blood. Maybe, just maybe…

“Our group meets each month and we have a good gossip, sometimes we go for a walk or do something else. It’s always good for me to do this, as women need to talk about things.”