



Preimplantation Genetic Diagnosis (PGD)

Guideline for getting access to PGD for severe Haemophilia A or B

What is PGD?

Preimplantation Genetic Diagnosis (PGD) is a procedure involving the removal of one or two cells from an embryo and subsequent genetic testing of these cells for specific genetic conditions or chromosomal disorders.

PGD involves having an IVF treatment. During the IVF cycle, one or more cells are removed from each embryo in a procedure called embryo biopsy, which typically happens on day 3 or 5 after egg collection. The biopsied cells are transported to a genetic testing laboratory, while the embryos from which the cells were obtained continue to develop in the IVF clinic. The results from the genetic testing laboratory are ready one or two days after biopsy, and are used to decide which embryos are suitable for transfer or freezing for future use. Sometimes none of the embryos are suitable for transfer.

PGD for haemophilia

There are different types of PGD. For Single Gene Disorders, such as haemophilia, a process called polymerase chain reaction (PCR) is used.

The DNA in each of the biopsied cells is replicated a million times or more using a process called multiplex fluorescent polymerase chain reaction (F-PCR). The DNA is then analysed to see what version of the gene is present in each biopsied cell. A single gene change will cause one particular line in the analysis to change (see the diagram below). Most of the single gene tests for PGD are done by Canterbury Health Laboratories in Christchurch.

Who can have PGD?

Females who know they carry a gene for severe haemophilia and who want to avoid passing haemophilia to their sons, can ask for PGD. If the woman does not know if she is a carrier, then she should ask for carrier testing. This is done by testing her blood for the haemophilia gene.

What does PGD involve?

Genetic counselling and informed consent is essential. Planning blood tests are also necessary.

Most people require a feasibility study before their first PGD cycle. The genetics lab uses a blood sample from the man and woman, and sometimes other family members, to check that the particular gene can be successfully identified using reagents available to the genetics laboratory.

When these are completed, the couple go through in vitro fertilisation (IVF) to produce embryos. Each embryo is then carefully tested to see if it has the haemophilia gene. The fertility specialist and the couple then decide which embryos to transfer to the woman, in order to become pregnant.

EMBRYOS CREATED BY IVF CAN BE TESTED FOR SERIOUS GENETIC DISORDERS USING PGD. ONLY UNAFFECTED EMBRYOS ARE TRANSFERRED.

Is PGD funded by District Health Boards?

Yes. Funding is available for two rounds of PGD per couple.

PGD in New Zealand

PGD for severe haemophilia is available in Christchurch, through the joint cooperation of Canterbury Health Laboratories, Repromed Christchurch fertility specialists and the Canterbury District Health Board Haematology Service.

Do you have to live in Canterbury to get access to PGD in Christchurch?

No. Funding for New Zealand residents is available from the Ministry of Health nationally. Therefore, anyone can ask for PGD in Christchurch, no matter where they live.

For example, in Auckland Fertility Associates offers 'Transport Preimplantation Genetic Diagnosis' in partnership with Canterbury Health Laboratories. Egg collection, in vitro fertilisation and embryo culture all take place in Fertility Associates in Auckland. On day 3 or 5 of embryo development, cells are removed from each viable embryo and sent Canterbury Health Laboratories. There the genetic screening and diagnosis is made and the results and recommendations are sent back to Fertility Associates. Our embryologists then select the appropriate embryo(s) for fresh replacement and possible cryopreservation.

How does a couple get referred for PGD in Christchurch?

Speak to your Haemophilia Treatment Centre. We recommend that couples interested in PGD begin the process of genetic testing as soon as possible to be ready for when they wish to have children as their can be waitlists for the publicly-funded PGD service.

In Christchurch, ring the Haemostasis Service at Christchurch Hospital (03 364 1246), or get in touch with Dr Mark Smith, Haematologist, CDHB 03 364 0387, mark.smith@cdhb.govt.nz