

This article was written by Cindy Zaitsoff, Senior Genetic Counsellor
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When both individuals in a couple are carriers of cystic fibrosis (CF), there is a one in four chance that any pregnancy they conceive together could be affected with CF.

The aim of preimplantation genetic diagnosis (PGD) is to help a couple conceive a pregnancy that would not be affected with CF. To do this, IVF technology is used to create embryos for a couple. This involves the female taking medications to stimulate her ovaries to produce a large number of eggs. These eggs are then fertilised with the male's sperm. Three to five days later, either a single cell or a number of cells are removed from each embryo and tested for their specific gene changes in the CFTR gene. Only embryos that are unaffected are chosen for transfer into the female partner's womb. These embryos may or may not be carriers of CF. In New Zealand, only one embryo is transferred at a time and any other unaffected embryos are frozen for later use.

Some couples who have IVF treatment have great success with few obstacles. However, some couples face numerous challenges and may not be successful after many attempts. The same is true for PGD. At the start of a PGD cycle, there is about a 25% chance that the couple will have a baby from that cycle. The further they get through the cycle, the greater the chance for success. If a healthy embryo is transferred, the chance of having a baby increases to about 35-45%.

There are a number of steps for a couple to complete before they can start a cycle of PGD. The first step is to meet with a genetic counsellor at their local Genetic Services Hub to discuss the genetics, genetic testing, PGD process and whether or not this is the best option for the couple. It is essential that both individuals in the couple have had genetic testing and it is known what type of gene mutation they each carry. The couple are then referred to the fertility clinic.

There is public funding for a limited number of cycles of PGD each year in NZ and there is a substantial waiting list. There are some criteria that a couple need to meet to be eligible for funding: the female partner must be less than 40 years when referred, must be a non-smoker and have a BMI of under 32. Each eligible couple receives funding for two cycles, unless they have a baby born following the first cycle. If a couple wishes to access PGD quicker, they have the option of paying privately. The cost of a cycle varies but starts from approximately \$16,500.

Pre-implantation genetic diagnosis is 95-98% accurate for each embryo. This means that there is a small chance that an embryo transferred could actually be affected with CF. There is the option of having a CVS or amniocentesis in pregnancy to confirm the diagnosis in the pregnancy.

If you are interested in exploring whether or not PGD is a good option for you, please ask your GP to refer you and your partner to Genetic Health Services NZ.

Genetic Health Service contact numbers

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Toll Free: 0800 476 123

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