CYSTIC FIBROSIS NZ FACT SHEET

Cystic fibrosis carrier screening

Statement approved by the Genetic Health Service March 2017

Approximately 1 in 25 of the Caucasian population are carriers of cystic fibrosis (CF). Population-based screening for CF involves individuals having genetic testing to determine if they are carriers of a mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene that causes CF. This can be performed in pregnancy or preferably earlier, during reproductive planning.

If an individual is shown to be a CF carrier, then their partner can be tested to see if (s)he is also a carrier. Cystic fibrosis only occurs when both parents are a CF carrier. If both parents are a carrier, it doesn't necessarily mean their baby will have cystic fibrosis – this is only one of the possibilities and has a 1 in 4 chance of occurring. The other possibilities are that the child will be born a carrier like their parents (but not have the condition), or they will be born without cystic fibrosis and will not be a carrier either.

If both parents are carriers then they will be offered genetic counselling to discuss their reproductive options. One of the options is to have prenatal testing which is genetic testing of an established pregnancy at about 11–12 weeks gestation via CVS or 15–16 weeks gestation via amniocentesis.

If prenatal testing is performed in the pregnancy and the foetus is shown to have cystic fibrosis, then the couple has the difficult decision of whether or not to continue the pregnancy. If the baby is shown to be a carrier, this can be helpful for them to know so as not to raise anxiety at the time of newborn screening (which can show a positive result for CF carriers) and also to have this information for their child's future reproductive implications.

Another option for couples who are both CF carriers is to consider preimplantation genetic diagnosis (PGD). This involves taking the couple through IVF treatment to create embryos that can then have a couple of cells removed and tested genetically. Only embryos that do not have CF are selected to be transferred into the female's womb. Thus, any resulting pregnancy will not be affected with CF.

In the Australian state of Victoria, population-based screening for CF carriers is offered to pregnant women although not all decide to take up this offer. Screening identifies the majority of CF carriers. However, the screening test is designed to identify only the most common CF causing alterations in the gene, but it does not screen for the more than 2000 gene changes that have been shown to cause CF. While screening cannot identify all CF carriers, it does significantly reduce the risk of having an affected child for couples who are shown not to be carriers.

In Australia, 67 percent of parents identified as carriers through their screening programme, decided to use prenatal diagnosis when having more children.

A study of 71 CF health professionals in Australia found there was moderate support for population CF carrier screening. Opposition was associated with fear of discrimination

against carriers and limitations on predicting the severity of the condition before a CF child is born, since some people with cystic fibrosis do have a less severe form of the disorder.

There is no free population-based screening programme for prospective parents in New Zealand. However, the test can be sought through private clinics. These private clinics charge around $500 for an initial consultation and carrier testing for the most common CF mutations, as well as testing for two other common genetic disorders (Spinal Muscular Atrophy and Fragile X). Clinics can be contacted directly by prospective parents to arrange an appointment.

If you or your partner have a family history of CF; for example, someone who has CF or is a CF carrier, you can access free CF carrier testing from your GP. It is important to tell your GP the exact gene alteration/mutation identified in your family member to ensure your test is as accurate as possible. If both members of a couple are identified as CF carriers, then they can be referred for publicly funded genetic counselling to Genetic Health Services NZ.

Cystic Fibrosis Victoria's Position Statement on carrier screening supports *individuals having an opportunity to make informed life and reproductive choices as a result of carrier screening.*

Cystic Fibrosis New Zealand *recommends people interested in understanding their risks of being a carrier for a CF gene contact their GP or the NZ Genetic Health Service.*

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