Sonic Genetics
Quality is in our DNA

Genetic carriers of cystic fibrosis

Information for patients

The healthcare professional responsible for your care has given you this leaflet because you have been identified by Preconception Carrier Screening as a carrier of a change in the gene that causes cystic fibrosis. This fact sheet contains more information about the particular genetic disorder mentioned in your report. We recommend that you also discuss your result with an experienced doctor or genetic counsellor.

Each year, about one in 2,500 Australian babies, boys and girls, are born with cystic fibrosis (CF). This inherited genetic condition more commonly affects people who are of Northern European or UK descent (Caucasian).

CF is due to a problem with the way salt is moved in and out of the cells of the lungs, pancreas and sweat glands. This causes the normal mucus in all or some of these organs to become thick and sticky. In the lungs, bacteria build up in this thick mucus, causing repeat infections and lung damage. The mucus in the pancreas stops the enzymes it makes reaching the gut, causing problems with digestion. The salt builds up in the sweat glands, so the body loses salt through sweat.

Treatment involves daily physiotherapy to clear the mucus from the lungs, as well as, often, antibiotics for the infection. Medicines that contain the enzymes normally produced by the pancreas to aid digestion are also needed. Babies are tested for CF a few days after birth as part of the Newborn Screening Test. To confirm if the baby actually has inherited CF, their sweat is tested to see if it is high in salt. This early diagnosis means that treatment can be started, leading to a better quality of life, although there is still a reduced life expectancy.

Genetic conditions are caused by inheriting faulty genes. Our genes are made of DNA and tell the body how to function, grow and develop. The CF gene (its scientific name is CFTR) contains the information that the cells use to stop the mucus becoming thick and sticky. Everyone has two copies of the CF gene. Most people will have both of their CF gene copies working as they should. It is only when both of their CF genes are faulty that the person has CF.

About one in 25 Australians of Caucasian ancestry will be born with one CF gene copy that is working normally and one copy that is faulty. These people are genetic carriers of CF. However, the cells need only one CF gene copy to be working normally to stop the mucus becoming thick. Genetic carriers of CF do not have any symptoms of the condition and will not develop them.

How to interpret a carrier result for cystic fibrosis

Hearing about the carrier result will usually be unexpected. It may lead to a range of physical and emotional reactions. In the midst of this emotional confusion, there are some decisions that need to be made. You need as much information and support as possible when making these decisions.

Genetic counsellors are experienced in providing support and information that will help you and your partner make an informed decision. The only right decision is the one that is best for you and your family.

If you are not pregnant, you have time to think about the carrier test result when discussing your family planning options. Among these options is testing the baby in pregnancy for CF (prenatal testing). Another is using IVF and then testing the embryo for CF before implantation. However, having no testing for CF at all is also an option.

If you are pregnant, your options include prenatal testing or no testing for CF. Again, seeking support and information when making these decisions is important.

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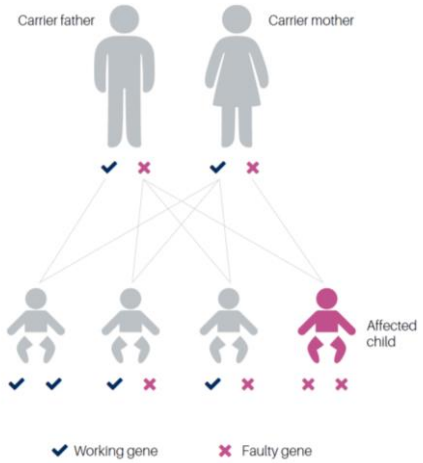
How does a person inherit CF?

For a baby to have CF, it must inherit the faulty gene from both their mother and father. Where both parents are genetic carriers of CF, there is a one in four chance (25%) in **every pregnancy** that they will each pass on the faulty CF gene copy to their baby who will then have CF.

In the Figure shown, both parents are carriers of CF, having one normal working CF gene and one faulty CF gene each. Their child will inherit one CF gene from each parent. There are four possible combinations of CF genes that the child could have, i.e. the child has two normal genes (one in four chance), or is a carrier (two in four chance), or has two faulty genes (one in four chance).



What does being a CF genetic carrier mean for me?

A genetic carrier of CF has one faulty gene and one normal CF gene which is working normally. The presence of the normal CF gene is sufficient to ensure that a carrier will not develop CF. There is a one in two chance (50%) of passing the faulty gene onto your child, but the child will not be affected if the child has inherited a normal CF gene from the other parent.

A child will develop CF only if the child has inherited a faulty gene from both parents. You have been identified as a carrier of CF. It is important to know the result of your partner's CF carrier test.

Rarely, males are found to be carriers of certain variations in their CF gene that do not make it faulty but can lead to infertility. These results should be discussed with your doctor.

What if one parent is a carrier and the other parent's carrier test is normal?

There are many variations in the CF gene that make it faulty. The carrier test looks for the most common variations. There is a small chance that the test has not found one of the rare variations that make the CF gene faulty. The possibility of having a baby with CF is still low (about one in 1,000), but the chance is higher than in the general population (one in 2,500). You should discuss this result with your doctor. Genetic counsellors are also experts in providing this information.

Your test result is important for your relatives

If you are a carrier of the faulty gene, you very likely inherited it from one of your parents. That means that your brothers, sisters and cousins, if you have any, also have a chance of being a CF genetic carrier. Telling them about the availability of the carrier test may help them make informed family planning decisions as well.

Sonic Genetics offers free genetic counselling for couples where both partners are identified as carriers for cystic fibrosis. If you are eligible for this service, your doctor may refer you for genetic counselling. Conditions apply and can also be discussed with your doctor.

Further information and contacts

The Centre for Genetics Education www.genetics.edu.au