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Sonic Genetics

Information for patients

Carriers of cystic fibrosis

The healthcare professional responsible for your care has given you this leaflet because you have been identified by reproductive carrier screening as a carrier of cystic fibrosis. This fact sheet contains some information about this genetic disorder. We recommend that you also discuss your test result with your doctor or genetic counsellor.

What is cystic fibrosis?

Each year, one in 3,500[1] babies in Australia is born with cystic fibrosis (CF). This inherited genetic disorder is more common among children 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 mucus in these organs to become thick and sticky. In the lungs, bacteria build up in this thick mucus, causing repeated infections and lung damage. The mucus in the pancreas stops the enzymes it produces reaching the gut, causing problems with digestion. Salt also builds up in the sweat glands and the body loses excess salt through sweat. The treatment for CF involves daily physiotherapy to clear the mucus from the lungs, antibiotics for infections, and medicines that contain the enzymes normally produced by the pancreas.

Babies are routinely tested for CF a few days after birth as part of the Newborn Screening Test. To confirm if the baby has inherited CF, their sweat is tested to see if it is high in salt. This early diagnosis means that treatment can be started promptly, leading to a better quality of life, although there is still a reduced life expectancy.

Genetic disorders can be 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. In most people, both copies of the CF gene are working as they should.

About one in 30[1] Australians of Caucasian ancestry will be born with one CF gene that is working normally and one that is faulty. These people are carriers of CF. The cells need only one copy of the CF gene to be working normally to stop the mucus becoming thick, and so carriers of CF do not develop CF. A person will develop CF only when both of their CF genes are faulty.

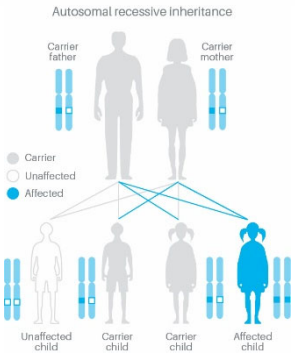
What does being a CF carrier mean for me and my child?

A carrier of CF has one faulty CF gene and one normal CF gene. The presence of the normal CF gene is sufficient to ensure that a carrier will not develop CF. There are some men who are carriers of CF and who also have a variation in the normal CF gene that is not sufficient to cause CF but can lead to infertility; this uncommon finding would be discussed by your doctor.

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

Information for patients

How does a child inherit CF?



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

In the Figure shown, both parents are carriers of CF, having one normal 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 inherit leading to three possible outcomes, that is, 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). The sex of the child bears no relevance to the inheritance of this disease.

What if both parents are carriers of CF?

If you are not pregnant, you and your partner have time to think about the carrier test result when discussing your options for family planning. These options include testing the baby in early pregnancy for CF (prenatal testing). Another option is using IVF and then testing the embryo for CF before implantation. There are other options, such as using a donor egg or sperm, or having no testing for CF at all. You should discuss these options with your doctor.

If you are pregnant, your options include prenatal testing, or no testing for CF. You should discuss these options with your doctor.

Hearing about your carrier result will usually be unexpected. Some people find this information confronting or confusing at the very time that decisions need to be made. You need information and support when making these decisions.

Your doctor may suggest that you discuss these issues with a genetic counsellor, a healthcare professional who can help you and your partner make an informed decision. Sonic Genetics offers free genetic counselling for couples where both partners are identified as carriers. Your doctor can refer you for this service; conditions apply and can be discussed with your doctor.

Your doctor or genetic counsellor will not tell you what to do: the right decision is what you decide is best for you and your family.

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

If one parent is a carrier there is a one in two chance (50%) of passing the faulty gene to your child. However, the child is very unlikely to be affected, as the child will almost certainly inherit a normal CF gene from the other parent.

There are rare situations in which a carrier test does not detect a person's faulty CF gene. This means that the carrier test result is 'normal' even though the person is actually a carrier. The reason for this is that there are many possible variations in the CF gene that can make it faulty. Our reproductive carrier screen looks for the commonest 50 variations that occur in Australia. There is a small chance that the test has not found one of the rare variations that can make the CF gene faulty. If one parent is known to be carrier, and no CF variant has been detected in the other parent, the possibility of having a child with CF is very low (about one in 1,000), but the chance is higher than in the general population (one in 3,500[1]). You should discuss this result with your doctor or genetic counsellor.

Your test result is important for your relatives

If you are a carrier, you probably inherited the faulty gene from one of your parents. That means that close family members, such as brothers, sisters and cousins, may also be carriers. We recommend that you tell them about your result and the availability of reproductive carrier screening so that they can make their own informed decisions about family planning.

Further information is available at The Centre for Genetics Education, genetics.edu.au

[1] Archibald A, Smith M, Burgess T, et al. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. *Genet Med*. 2018; **20**(5):513-526

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