

Cystic fibrosis

Cystic fibrosis is a genetic condition, which affects many parts of the body.

People with cystic fibrosis can have a range of different problems. The most common ones are lung disease (thick mucus that is difficult to cough up and lung infections), a problem with the pancreas (food is not digested and absorbed properly), loss of excessive amounts of salt in the sweat and, in males, infertility.

Other less common problems include bowel obstruction, diabetes and liver disease.

Cystic fibrosis is a serious condition which usually shortens the life of the person affected. As recently as two decades ago, a person with cystic fibrosis was unlikely to reach their twenties. Now, with improved treatment, the average lifespan is about 30 years and is expected to increase even further.

Cystic fibrosis comes about as a result of an alteration to the CFTR gene on chromosome 7. There are about 1000 known CFTR gene alterations.

Cystic fibrosis is an autosomal recessive condition (see fact sheet on '[How do genetic conditions occur?](#)'), which means that somebody with one altered gene will be a carrier and somebody with two altered genes will have the condition.

An altered gene is found in 1 in 25 people of northern European ancestry and is slightly less common in people of southern European or Middle Eastern ancestry. An altered gene is rare in people of African or Asian ancestry.

If two people who are carriers have children, then each child has a 1 in 2 chance of being an unaffected carrier, a 1 in 4 chance of having the condition; and a 1 in 4 chance of not having the altered gene and not being affected.

Newborn screening tests (see fact sheet on '[Newborn screening](#)') aim to detect cystic fibrosis. They do so in more than 90 out of 100 cases.

But the newborn screening tests are not perfect and 5 or 10 in every 100 people with cystic fibrosis do not have the condition picked up on those tests.

The main test for cystic fibrosis is called a sweat test. This measures the salt content of sweat. Children with the condition have a higher than normal amount of salt in their sweat.

What about other family members?

If someone in the family has cystic fibrosis, then both parents are automatically carriers for the altered gene for cystic fibrosis.

All members of the close family – parents, brothers, sisters, children, uncles and aunts – should see their doctors to discuss cystic fibrosis.

If the particular gene alteration causing the cystic fibrosis in the family is known, then blood tests to detect carriers will be highly accurate.

If the particular gene alteration is not known, it may still be possible to do a carrier test, but it will be more difficult.

If someone is a carrier for cystic fibrosis and they and their partner are planning to have children, the partner should consider having carrier testing before pregnancy. Testing will pick up only about 85 in 100 carriers for cystic fibrosis – the test can not check for all 1000 gene alterations. If no gene alteration is found, the chance that the partner is a carrier will be much lower than before the test, but the chance will not have been removed altogether.

Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Cystic Fibrosis Australia at <http://www.cysticfibrosis.org.au>
- Cystic Fibrosis Medicine at <http://www.cysticfibrosismedicine.com>
- The Lung Foundation at <http://www.lungnet.org.au>
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- Better Health Channel at <http://www.betterhealth.vic.gov.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- HealthInsite at <http://www.healthinsite.com>
- MedicineNet at <http://www.medicinenet.com>
- MyDr at <http://www.mydr.com.au>
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email gtis-australia@unimelb.edu.au or visit Biotechnology Australia's website at <http://www.biotechnology.gov.au>