
Cystic fibrosis (CF)

Summary

- Cystic fibrosis (CF) is a life-limiting genetic disorder.
 - It mainly affects the lungs, the digestive system (the pancreas and sometimes the liver) and the reproductive system.
 - There is no cure for CF, but treatment can slow progression of the disease.
 - CF is caused by a child inheriting two copies of a changed (mutated) gene – one copy from each parent.
 - One in 25 people carry a single copy of the changed gene but do not have CF and will have no symptoms.
 - CF is usually diagnosed at birth. It is not contagious, and it occurs equally in males and females
 - Testing can now identify if you carry the CF gene change.
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Cystic fibrosis (CF) is a life-limiting genetic disorder. It impacts the whole body, but mainly affects the respiratory system (lungs), the digestive system (the pancreas and sometimes the liver) and the reproductive system.

When a person has CF, their mucus is very thick and sticky. It is difficult for people with CF to clear this mucus from their lungs. It clogs the tiny air passages and traps bacteria. This causes recurring infections and blockages, which can cause irreversible lung damage over time.

Thick mucus in the digestive system can also affect the transfer of digestive enzymes from the pancreas to the small intestine. This leads to difficulty with digesting fats and absorbing some nutrients. This means that people with CF can have problems with nutrition and need to consume a diet high in kilojoules, fats and salts.

CF is the most common life limiting genetic disorder affecting Australians today for which there is no cure.

Symptoms of cystic fibrosis

People with CF may experience:

- a persistent cough that sometimes produces thick mucus
- difficulty breathing
- wheezing
- frequent lung infections
- salty sweat – salt loss in hot weather may produce muscle cramps or weakness
- tiredness, lethargy or reduced ability to exercise
- poor growth or weight gain
- frequent visits to the toilet
- bulky, greasy poos
- diarrhoea or constipation
- poor appetite.
- CF-related diabetes
- infertility in males.

Frequency of cystic fibrosis

CF is the most common life limiting genetic disorder affecting Australians today for which currently there is no cure. A baby is born with CF every four days.

In Australia, approximately one in 25 people carry a single copy of the CF gene change. People who carry one copy of the CF gene change do not have any symptoms of the condition.

If two people carry the CF gene change and they have a child, each pregnancy will have:

- a one-in-four chance that the child will have CF
- a two-in-four chance that the child will not have CF, but will carry a single copy of the gene change (they will be a carrier of the CF gene change)
- a one-in-four chance that the child will not have CF and will not carry the gene change.

One in every 2,500 births produces a child who has CF. Approximately 3,500 people in Australia have CF.

Diagnosis of cystic fibrosis

In Australia, most babies are screened at birth for CF through the newborn screening test. This involves collection of a blood sample through a heel prick test immediately after birth. If the results of the screening test reveal very high levels of a substance called immunoreactive trypsin (IRT), CF is suspected and the DNA in the blood is then analysed for the most common gene changes that cause CF.

A sweat test may be done to measure the amount of salt (sodium chloride) in the sweat and confirm the diagnosis.

Some babies may also be diagnosed shortly after birth as a result of an intestinal blockage called **meconium ileus**. Most people who have CF are now diagnosed within the first two months of life.

Treatment for cystic fibrosis

Treatment for CF can be intensive and time consuming. At present, there is no cure for CF. Treatment aims to slow progression of the condition and can include:

- chest physiotherapy
- medications that help correct the changes to the body's cystic fibrosis transmembrane conductance regulator (CFTR) protein, which is what causes CF
- antibiotics
- inhalations via a compressed air pump and nebuliser
- enzyme replacement capsules with meals and snacks
- a well balanced diet high in protein, fat and kilojoules
- percutaneous endoscopic gastrostomy (PEG) – a procedure in which a flexible feeding tube is placed through the abdominal wall and into the stomach
- supplementary vitamins
- salt supplements
- regular exercise.

Regular attendance at a major CF clinic is beneficial and recommended.

Carrier testing for cystic fibrosis

People who carry a single copy of the CF gene change are healthy and often not aware that they are carrying this gene change. If you are planning a pregnancy, you can be tested to see if you are carrying the CF gene change.

Cystic Fibrosis Community Care has a range of information and resources available about carrier testing.

Genetic counselling and cystic fibrosis

If your child or another family member has been diagnosed with cystic fibrosis, or if CF runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand CF and what causes it, how it is inherited, and what a diagnosis means for your child's health and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If CF runs in your family, a genetic counsellor can explain what genetic testing options are available to you and

other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

Support for people and families with cystic fibrosis

Cystic Fibrosis Community Care provides a range of support programs and services for people and families living with CF in Victoria and NSW.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by CF.

Where to get help

- Your **GP (doctor)**
- Your **obstetrician**
- **Cystic Fibrosis Community Care** in Victoria Tel. **(03) 9686 1811** and NSW **(02) 8732 5700**
- **Cystic fibrosis clinics**
- **Victorian Clinical Genetics Service** Tel. **1300 118 247**
- **Genetic Support Network of Victoria** **(03) 8341 6315**

This page has been produced in consultation with and approved by:

Cystic Fibrosis Community Care

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