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.

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.

**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