Cystic Fibrosis (CF)

What is Cystic fibrosis (CF)?

Cystic Fibrosis (CF) is the most common serious inherited (genetic) condition in our community.

CF is an inherited disorder, caused by mutations in a particular gene pair. In Australia, about one child in 2500 is born with CF.

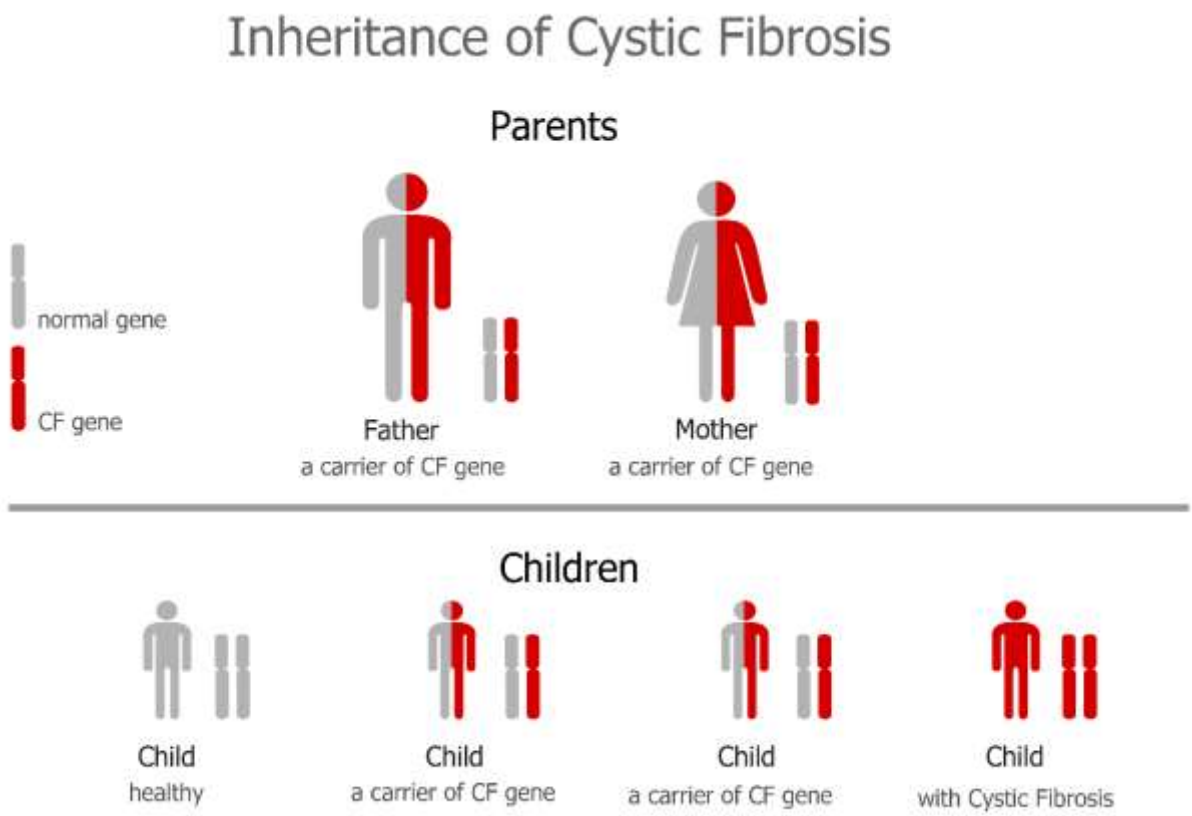
People who carry the CF mutation are 'unaffected' – that is, they show no symptoms of CF – and are usually not aware that they carry a mutation that can cause CF. **There are likely to be about one million carriers of CF in Australia**.

All newborns in Australia have a 'heel prick test' – this tests for a number of conditions, one of which is CF. However, the heel prick test does not detect all cases of CF. The definitive test is known as the 'sweat test' since people with CF have a high concentration of salt in their sweat.

Symptoms

People with CF have very thick and sticky mucus secretions that affects the function of their lungs and digestive system. People with CF can have lifelong coughing and shortness of breath with persistent lung infections. CF also causes poor growth, diabetes, liver problems, and osteoporosis.

The treatment of CF includes regular physiotherapy to help clear lung secretions, antibiotics, and enzyme supplements to help digestion.



What is the cystic fibrosis gene?

The CF gene makes a protein that controls the flow of salt in and out of cells that line the lungs and the digestive system. In CF the mucus glands secrete too much salt and not enough water, making secretions in the airways sticky and prone to infection.

Cystic fibrosis results from both parents carrying a mutation in one of their two copies of the CF gene. If both healthy parents carry a fault in the cystic fibrosis gene, each child has a one in four (25%) chance of inheriting two faulty genes, one from each parent and therefore having cystic fibrosis. The parents are healthy gene carriers because they carry one faulty copy of a gene and one normal copy. Their normal copy of the gene keeps them healthy and compensates for the faulty copy of the gene.

Over 1000 different changes – mutations – have been found in the CF gene, however, most carriers have one common mutation called ΔF_{508} (pronounced 'delta-F-508'). More than 90% of people with CF carry at least one ΔF_{508} mutation.

While there have been great improvements in the length and quality of life for people with CF, it continues to be a serious condition.

Carrier parents have a number of options when considering pregnancy…

- Some people will choose to take a chance and hope that the baby does not have CF.
- Some may choose not to have any children, or to have any more children after an affected child.
- Many choose to have testing in early pregnancy to determine whether the baby has inherited two CF genes and will therefore have CF.
- Other couples will consider using the technique called pre-implantation genetic diagnosis (PGD). This is where the baby is conceived by an in-vitro fertilisation (IVF) procedure and embryos are tested for CF before they are implanted: only embryos that do not carry CF are implanted into the woman's uterus.