# Cystic Fibrosis and Carrier Testing

## Information for people considering carrier testing for cystic fibrosis

## What is cystic fibrosis?

Cystic Fibrosis (CF) is a genetic condition. It usually affects people from birth and causes a number of different symptoms. The main problems it causes are with a person's lungs and with their digestion.

### Lung symptoms

People with CF have very sticky mucus in their lungs. This leads to lung infections and over time this can lead to severe damage to their lungs.

### Digestive symptoms

People with CF are also not able to secrete the enzymes into their gut that are needed to digest food properly. This means it is very hard for them to extract the nutrition they need from the food they eat.

## What causes CF?

CF is an inherited condition which means it is caused by a variant or 'mutation' in a person's genes.

Genes are the unique set of instructions inside our bodies which help make each of us an individual. We each have two copies of every gene (one from our father and one from our mother).

Individuals, who are affected by CF, have a variant in both copies of their CF gene and therefore have no working copies of the gene.

If a person has a variant in only one of their two copies of the CF gene they should not have any symptoms of CF because they have one working copy of the gene pair. These people are known as ***carriers*** of CF.

## How is CF inherited?

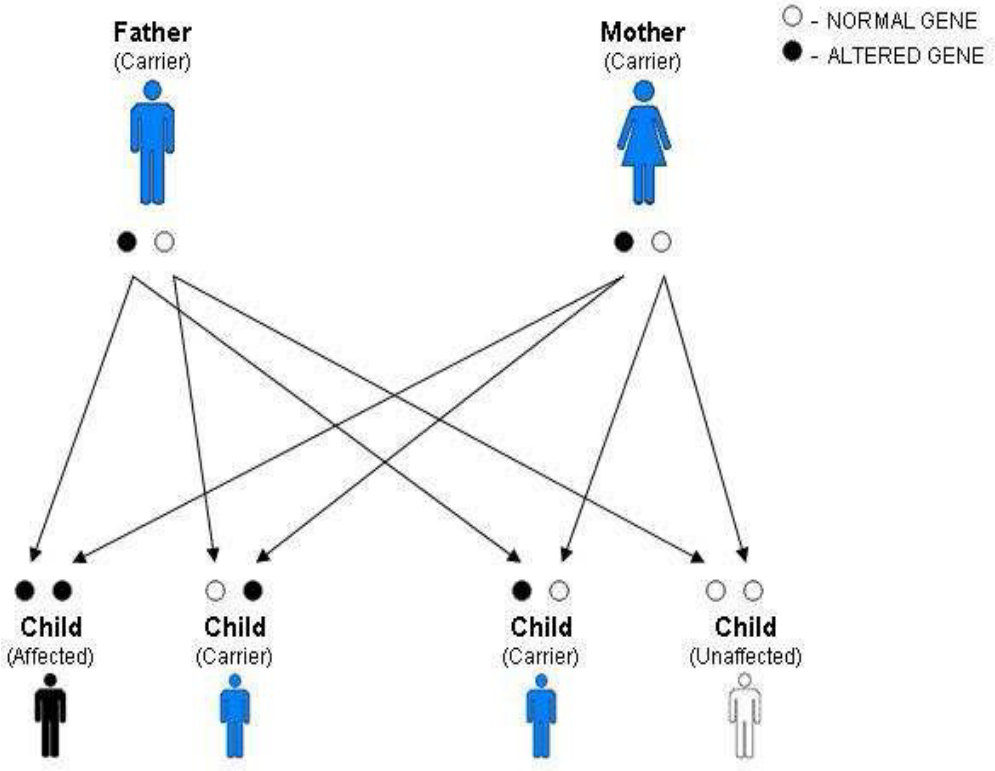
CF is passed on in what is known as an autosomal recessive way.

In any pregnancy, where both parents are CF carriers, there is a:

- 1 in 4 (or 25%) chance that the pregnancy will inherit two copies of the CF gene that both have variants in them and the child will have CF.
- 2 in 4 (or 50%) chance the pregnancy will have a variant in one copy of their CF gene. The child will also be a carrier of CF. This has no implications for their own health
- 1 in 4 (or 25%) chance that the pregnancy will have inherited two working copies of the gene. The child is not a carrier and will not develop CF.

If both parents have had a genetic test and only one of them has been found to be a carrier, then the chance that their baby will be a carrier of CF is 1 in 2 (or 50%). The chance that they will be affected by CF is very small.

The diagram below might be helpful.




## Cystic fibrosis carrier testing

It is possible to have a blood test to find out if you are a CF carrier. If necessary, CF carrier testing can be arranged on an urgent basis.

The usual test you will be offered looks for the most common variants in the CF gene. In all, this covers about 90% of the variants that cause CF.

If someone in your family has been diagnosed with CF or is known to be a carrier, then the test should be able to tell you if you have any of the CF variants that have been identified in your family.

## The results of CF carrier testing

The results of CF carrier testing are usually reported within four weeks and the person who requests the testing will arrange to get the results back to you.

The results may show that you are a CF carrier. In this case, your partner should be offered testing if you are planning to have children. If you are **both** found to be carriers, you should be referred to a genetic service for advice.

Alternatively, the test may not identify any of the common variants in your CF genes. In this circumstance, your chance of being a carrier will be significantly reduced. A residual risk that you are a carrier will be given.

If you and your partner have both had testing and are not found to be carriers, a residual risk will be given of you having a child with CF. The overall chance of you having a child with CF will be extremely low and no further testing would be recommended.

## Where can I find more information?

**The Cystic Fibrosis Trust**
The UK's leading charity for people affected by cystic fibrosis is the **Cystic Fibrosis Trust.** Their website contains a range of useful information, an online forum and news items about ongoing research into cystic fibrosis:

www.cysticfibrosis.org.uk

The charity also operates a helpline: telephone 0300 373 1000 or email helpline@cysticfibrosis.org.uk, which is available from 9am and 5pm, Monday to Friday.

## Local contacts

**Your local genetics service:**

South East of Scotland Clinical Genetic Services
Molecular Medicine Centre (MMC)
Western General Hospital
Crewe Road South
Edinburgh
EH4 2XU

Telephone: 0131 537 1116