CYSTIC FIBROSIS AND GENETIC TESTING

THIS LEAFLET IS WRITTEN FOR PEOPLE CONSIDERING GENETIC 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 a genetic condition. This means it is caused by a change 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 change in both copies of their CF gene and therefore have no working copies of the gene.

If a person has a change 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.

HOW IS CF INHERITED?

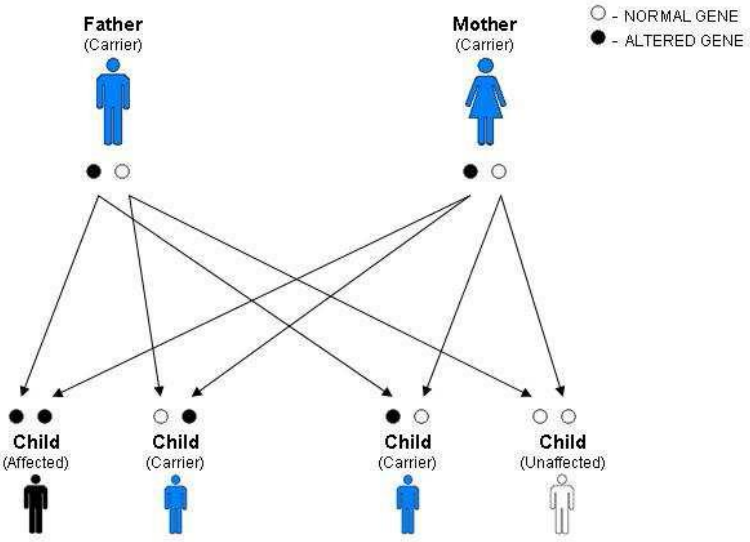
CF is passed on in what is known as an autosomal recessive way.

For someone to be affected by CF, both of their parents must be carriers of a changed gene.

In any pregnancy, where both parents carry a CF gene change, there is a:

- 1 in 4 (or 25%) chance that the pregnancy will have two copies of the changed CF gene and the child will have CF
- 2 in 4 (or 50%) chance the pregnancy will have a change in one copy of their CF gene. The child will be a carrier of CF. This has no implications for their own health
- 1 in 4 (or 25%) chances 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 TESTING

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

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

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

THE RESULTS OF CF TESTING

The results of CF carrier testing are usually ready within four weeks and the person who arranges the testing will make arrangements to get the results back to you.

The results may show that you are definitely a carrier for a CF gene change or it may be that the test does not identify any gene changes in your CF genes. In this circumstance, your chance of being a carrier will be significantly reduced.

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 (**https://www.cysticfibrosis.org.uk**) contains a range of useful information, an online forum and news items about ongoing research into cystic fibrosis.

The charity also operates a helpline: **0300 373 1000**, which is available from 9am and 5pm, Monday to Friday.

LOCAL CONTACTS

Your local genetics service:

SE of Scotland Clinical Genetic Services
MMC, Western General Hospital,
Crewe Road South,
Edinburgh, EH4 2XU.

Telephone: 0131 537 1116