



**The Leeds
Teaching Hospitals**
NHS Trust

Yorkshire Regional Genetics Service

Cystic Fibrosis Carrier Testing

Information for patients



Yorkshire Regional
Genetics Service

What is Cystic Fibrosis (CF)?

CF is a genetic condition affecting around 1 in 2,500 people. CF affects a number of organs in the body (especially the lungs and pancreas) by clogging them with thick, sticky mucus. The symptoms of CF can include:

- Repeated chest infections and coughing
- Digestive problems
- Diarrhoea and abnormal stools

What is a genetic condition?

A genetic condition is caused by an alteration in our genes. Genes are the set of instructions inside our bodies which makes each of us an individual. There are thousands of different genes, each gene has a role in the body. If a gene is altered, it can cause a genetic problem or disease. We have two copies of each gene. One copy comes from our mother and the other comes from our father. When we have children, we pass on one copy of each of our genes.

CF is a recessive genetic condition. This means that people with CF have an alteration in both copies of their CF gene. Individuals with only one altered copy are known as carriers. Their unaltered CF gene keeps them healthy and compensates for the altered copy of the gene.

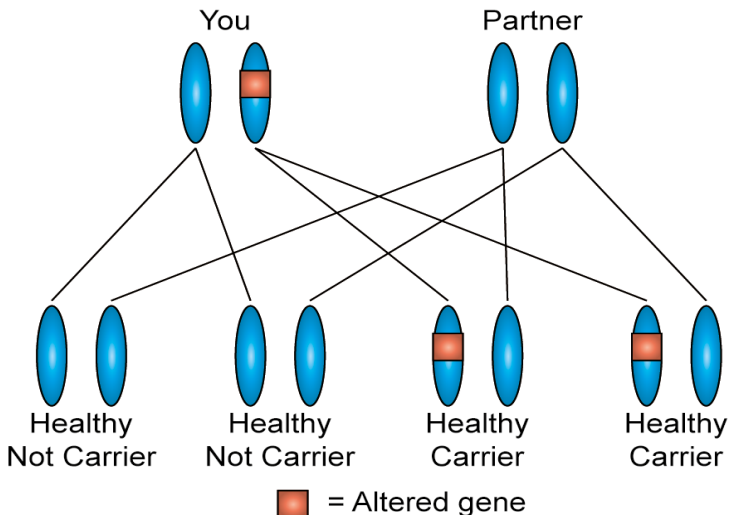
Might I be a carrier?

Carrier testing can be done and will be discussed with you. The table below shows the chance of being a carrier for various healthy relatives.

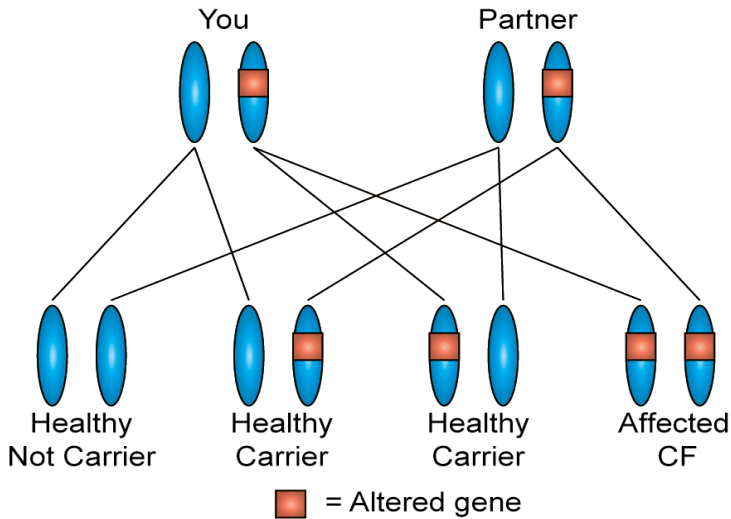
Relation to person affected by CF	Carrier Risk
Parents	1 (100%)
Brother or Sister	2 in 3
Aunt or Uncle	1 in 2
Grandparent	1 in 2
First Cousin	1 in 4

Will my children have CF if I am a carrier?

If your partner is not a carrier, you will not have a child with CF but there will be a 1 in 2 (50%) chance that your child will be a healthy carrier.



If, however, your partner is also a carrier there is a possibility of having a child with CF. There will be a 1 in 4 (25%) chance that you will both pass on your altered copy of the gene and have a child with Cystic Fibrosis.



There will be a 2 in 4 (50%) chance that only one of you will pass on an altered copy of the gene. When this happens, the child is a healthy carrier of CF.

There will also be a 1 in 4 (25%) chance that you both pass on your normal copies and have a child who is not a carrier.

These chances will be the same in each pregnancy.

Some couples who are both carriers want to know if a baby will be affected by CF. There are two types of test available during pregnancy which can tell whether the baby is affected with CF. These tests can be discussed with you in more detail by a genetic counsellor.

What does a carrier test involve?

When you come in for your appointment, a small amount of blood will be taken. This will be sent to our laboratory where they will look for any alterations in the gene that is involved in CF.

A carrier test is able to look for 90% of alterations. If you have an unusual alteration in your family, the laboratory may not be able to find it. Therefore, if no alteration is found this does not mean you are definitely not a carrier. It will, however, greatly reduce your chance of being a carrier.

Questions to consider

- What would being a carrier mean to me?
- Who would I tell if I found out I was a carrier?
- Would I consider tests in a pregnancy?
- Are there any other ways to prevent having an affected child?

For more information:

If you need more information, please contact your local Genetics Department. If you live in the Yorkshire region, please contact:

Department of Clinical Genetics
Level 3
Chapel Allerton Hospital
Chapeltown Road
Leeds
LS7 4SA

Tel: **(0113) 3924432**

Further information and support is available from:

Cystic Fibrosis Trust
11 London Road
Bromley
BR1 1BY

Telephone: **0300 373 1000** CF Trust Helpline

Telephone: **020 8464 7211** General Enquiries

email: **enquiries@cysticfibrosis.org.uk**



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