

A carrier test is able to look for 90% of mutations. If you have an unusual mutation in your family the laboratory may not be able to find it. Therefore, if no mutation is found this does not mean you definitely are 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?

For more information:

If you need more advice about any aspect of Cystic Fibrosis, you are welcome to contact:

Clinical Genetics Department

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 Fax: 01224 559390
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035 Fax: 01382 645731
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)
Tel: 0131 651 1012 Fax: 0131 651 1013
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 201 0808 Fax: 0141 201 0361
(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

Seen in clinic by.....

Support Services
Cystic Fibrosis Trust
11 London Road
Bromley
BR1 1BY



Tel 0845 859 1000 CF Trust Helpline
Tel 020 8464 7200 CF Trust
Fax 020 8313 0472
e-mail: enquiries@cftrust.org.uk
Web Site: <http://www.cftrust.org.uk>

Cystic Fibrosis Carrier Testing



Information for Patients

This leaflet was originally designed by Guy's & St. Thomas' Clinical Genetics. Updated by Genetic Interest Group Scotland. Last updated March 2003.

What is Cystic Fibrosis (CF)?

CF is a genetic condition affecting around 1 in 2500 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
- Poor weight gain

What is a genetic condition?

A genetic condition is one that is caused by a change in our genes. Genes are the unique set of instructions inside our bodies which makes each of us an individual. There are many thousands of different genes, each carrying a different instruction. If a gene is altered, it can cause a genetic condition or disease. This gene alteration is sometimes known as a *mutation*.

We have two copies of each gene. One copy is inherited from each of our parents. When we have children, we pass on only one copy of each of our genes.

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

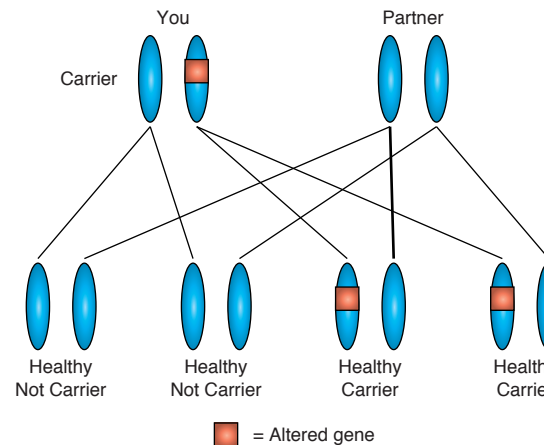
What chance is there that I am a carrier?

The table below gives risk figures for various healthy relations, assuming a partner without a family history of CF.

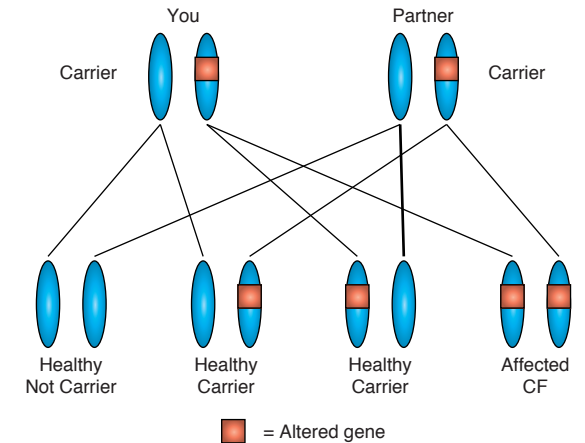
Relation to person affected by CF	Risk
Parents	1 (100%)
Brother or Sister	2 in 3
Aunt or Uncle	1 in 2
Grandparent	1 in 2
First Cousin	1 in 4
Second Cousin	1 in 8

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 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.

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. There are also leaflets available which tell you more about these tests.

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 changes (mutations) in the gene that is involved in CF.