Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Cystic Fibrosis Carrier Testing

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.

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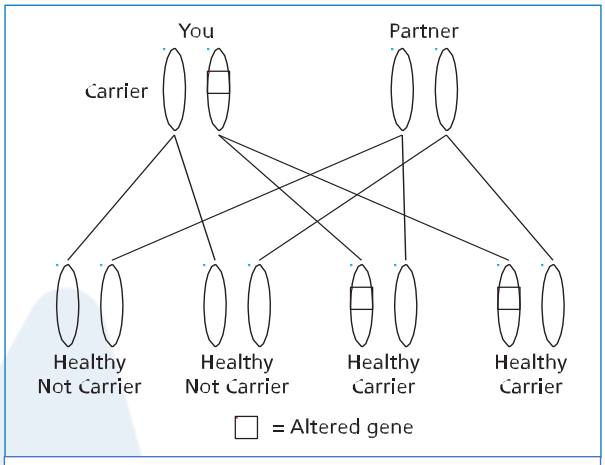
What chance is there that I am a carrier?

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

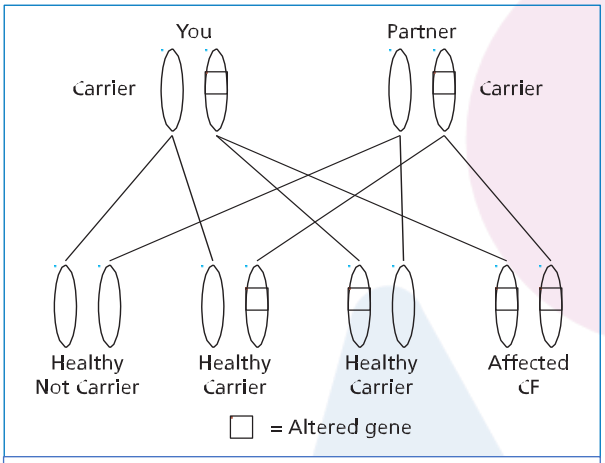
Relation to person affected by CF	Risk
Parents	100%
Brother or Sister	2 in 3
Aunt of Uncle	1 in 2
Grandparent	1 in 2
First Cousin	1 in 4
Second Cousin	1 in 8

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.



One partner carrier



Both partner's carriers

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.

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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.

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 that 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?

For more information

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

Manchester Centre for Genomic Medicine
Sixth Floor,
Saint Mary's Hospital,
Oxford Road Manchester, M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

Department staffed Monday - Friday, 9.00 am to 5.00 pm.

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For support and advice, please contact:

Cystic Fibrosis Trust
11, London Road,
Bromley,
BR1 1BY

Telephone: 0845 859 1000 CF Trust Helpline
Telephone: 020 8464 7200 CF Trust
Facsimile: 020 8313 0472
E-mail: enquiries@cftrust.org.uk
Website: http://www.cftrust.org.uk

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally designed and wrote this leaflet.

Seen in clinic by (doctor): _____

And (Genetic Counsellor): _____

Telephone number: _____

Family reference number: _____

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