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; and
- diarrhoea and abnormal stools.

Recently, new treatments have been developed, that are already improving the symptoms for many individuals with CF. These treatments have the potential to improve long-term outcomes significantly. You may wish to visit the Cystic Fibrosis Trust website (**www.cysticfibrosis.org.uk**) to learn more about these.

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 most of our genes. 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?

If you are related to someone who has been diagnosed with CF, or someone who knows they are a carrier of CF, there is a greater chance that you could be a carrier. You can usually request carrier testing though your GP.

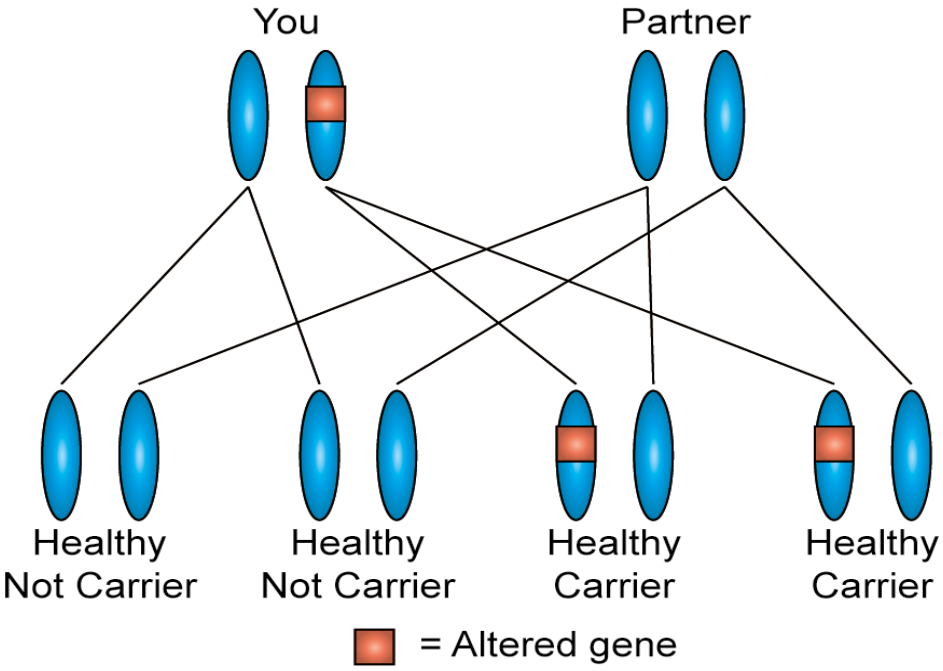
You can also request carrier testing if your partner is a carrier of CF or if your partner has a diagnosis of CF. This is because, if you are a carrier too, there is a chance you could have a child with CF.

If you need carrier testing because you or your partner are pregnant and one of you has a family history of cystic fibrosis, please ask your GP or midwife to refer you to Clinical Genetics. The table below shows the usual chance of being a carrier for various healthy relatives.

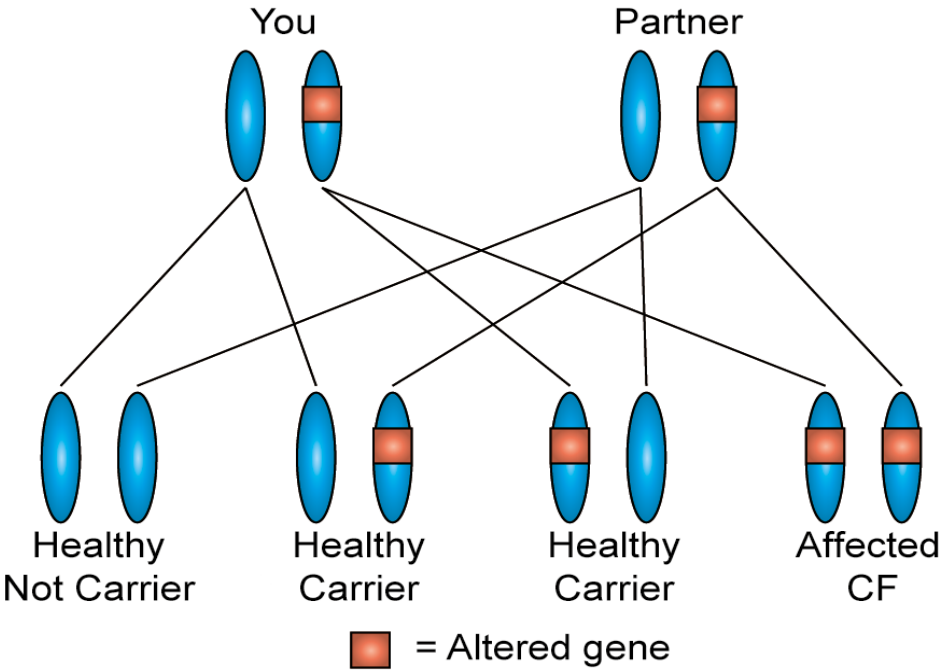
Relation to person affected by CF	Usual chance of being a carrier
Parents	1 (100%)
Brother or Sister	2 in 3
Aunt or Uncle	1 in 2
Grandparent	1 in 2
First Cousin	1 in 4

Will my children have CF if I am a carrier?

If your partner is not a carrier, it is very unlikely that you will 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 working copies of the gene and have a child who is not a carrier.

These chances will be the same in each pregnancy.

Many people who are carriers of CF choose to have a natural pregnancy and not pursue any genetic testing before their baby is born, in the knowledge that CF testing can be carried out at birth; however, some couples who know they are both carriers of a CF gene alteration choose to pursue reproductive genetic testing. There are several options that may be available including:

1. **Pre-implantation Genetic Testing (PGT):**

   A special form of IVF in which embryos undergo genetic testing before being transferred into the womb. Only embryos that have not inherited CF would be transferred.

2. **Prenatal Diagnosis:**

   Once someone is pregnant, they have a genetic test to find out if the pregnancy has inherited CF.

If you and your partner are both carriers of a CF gene alteration and want to know more about these reproductive options, please ask your GP or midwife to refer you to Clinical Genetics.

What does a carrier test involve?

A small amount of blood will be taken. This will be sent to the Genetics laboratory, where they will look for any alterations in the gene that is involved in CF. The lab will usually need details of any relative(s) who have CF to enable them to be able to interpret your results accurately.

A carrier test is able to look for approximately 90% of the cystic fibrosis gene alterations found in people of Northern European ancestry. This figure will vary for other populations. If someone has a rare alteration, this may not be picked up by the test; 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.

Some alterations in the CF gene do not cause the usual symptoms of cystic fibrosis but may cause mild symptoms or affect fertility; therefore, carrier testing can occasionally reveal possible health implications for the individual being tested. If this occurs, you would be referred to the Genetic Clinic to discuss this.

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
One Aldgate
2nd Floor
London EC3N 1RE

Tel: **0300 373 1000** or **020 3795 2184** **(CF Trust Helpline)**
Tel: **020 3795 1555** **(General Enquiries)**

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

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

What did you think of your care?

Scan the QR code or visit **bit.ly/nhsleedsfft**

Your views matter

Developed by: Dr Alison Kraus, Consultant

Reviewed by: William Beckett, Consultant Lead Genetic Counsellor (June 2023)

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