Pre-implantation genetic diagnosis (PGD) is another option that might be available to some couples. This involves IVF (in vitro fertilisation), and testing embryos in a laboratory, before they are implanted into the female.

There is also a non-invasive testing option during pregnancy that might be available to some people. This is known as non-invasive prenatal diagnosis (NIPD), and can be discussed with a genetic counsellor.

Having a carrier test

A small blood sample is required for CF carrier testing. This will be checked for mutations in the genes that are involved in CF.

A standard carrier test looks for the 50 most common CF gene mutations that account for 9 out of 10 (90%) of the CF diagnoses in the northern European population.

If we know the gene mutation in your family, we can give you a definite answer of whether you carry the CF gene mutation specific to your family.

If we do not know the mutation in the family, you can have the above test. We will either identify a mutation or, if the result does not find any mutated genes, it is unlikely that you would be a carrier for CF. Note that it is not a **guarantee** that you are not a carrier.

Questions to think about

- What would being a CF carrier mean to me?
- Who would I tell if I found out I was a CF carrier?
- Would I consider testing in a pregnancy, or PGD, if available?
- Are there any other ways to avoid having an affected child?

Useful sources of information

Cystic Fibrosis Trust
Helpline 0300 373 1000
email enquiries@cftrust.org.uk
web www.cysticfibrosis.org.uk

Contact us

Clinical genetics department, 7th Floor, Borough Wing, Guy's Hospital, Great Maze Pond, London SE1 9RT
phone 020 7188 1364, **fax** 020 7188 1369
web www.guysandstthomas.nhs.uk/our-services/genetics

For more information on conditions, services, and treatments we offer, please visit **web** www.guysandstthomas.nhs.uk

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A list of sources is available on request

NHS
Guy's and St Thomas'
NHS Foundation Trust

Cystic fibrosis carrier testing

This leaflet explains more about tests to find out if you are a carrier of the cystic fibrosis gene.

showing we care

Cystic fibrosis (CF)

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

- repeated chest infections and coughing
- digestive problems
- diarrhoea and abnormal poo (stools)

What is a genetic condition?

A genetic condition is caused by a change in our genes. Our genes are the set of 'instructions' inside our bodies, which make each of us individual. There are thousands of different genes, and each gene has a functional role in the body. If a gene is altered, it can cause a genetic problem or disease. A fault in a gene is known as a mutation.

We have 2 copies of each gene. We get 1 copy from our mother and 1 from our father. When we have children, we pass on 1 copy of each of our genes to each child. This happens at random.

Recessive genetic confitions

CF is a recessive genetic condition. This means that people with CF have a mutation in **both** copies of their CF gene. People with only 1 mutated copy and one normal copy of the CF gene are healthy, and are known as CF carriers. Their normal CF gene keeps them healthy and compensates for the mutated copy of the gene.

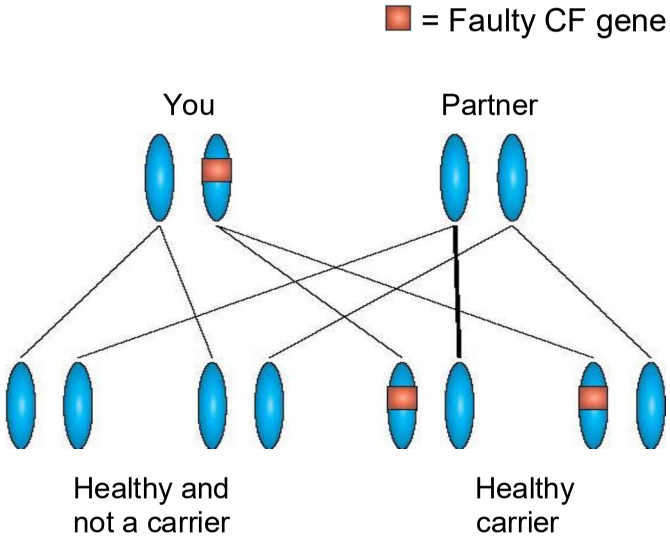
Carrier testing

Carrier testing can be done and will be discussed with you. This table gives carrier risk figures for various healthy family members.

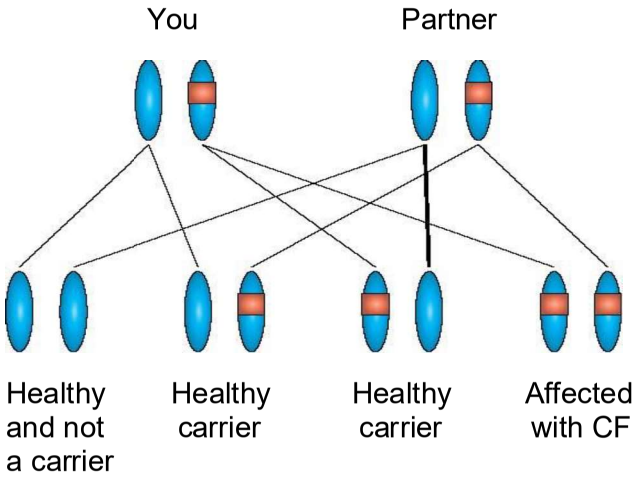
Relation to person affected with CF	Carrier risk
Parent	1 in 1 (100%)
Brother or sister	2 in 3 (66%)
Aunt or uncle	1 in 2 (50%)
Grandparent	1 in 2 (50%)
First cousin	1 in 4 (25%)

Your children

If your partner is not a carrier, it is 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 your partner is also a carrier, there will be a 1 in 4 (25%) chance that you will both pass on your mutated CF gene and have a child with cystic fibrosis.



There will be a 1 in 2 (50%) chance that **only one** of you will pass on a mutated CF gene. When this happens, the child will be a healthy carrier, like you.

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

These chances will be the same for each pregnancy.

Some couples who are both carriers want to know if a baby will be affected with CF. There are 2 types of test available during pregnancy, which can tell if the baby has CF. These tests can be discussed in more detail with a genetic counsellor. We will give you information about these tests.