

Testing for cystic fibrosis carriers in families

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Introduction

Genetic testing is available for family members of someone who has cystic fibrosis (CF), to find out if they are carriers of CF mutations.

This factsheet explains:

- what CF is,
- what carrier testing screening is and how it works,
- what it means to be a carrier,
- the odds of being a carrier of the CF gene,
- who can have carrier testing,
- where you can go for testing and further information.

This is a complicated topic. If you have any questions please discuss it with your GP, or CF team if you have access to one. They can refer you to a genetic specialist (expert on inherited conditions).

What is cystic fibrosis?

CF is an inherited condition caused by a fault, or 'mutation', in a gene that affects the balance of salt and water in cells. CF is one of the most common inherited conditions in the UK, affecting over 10,800 people.

Having CF can make breathing difficult and lead to a cough and repeated lung infections that are difficult to get rid of. People with CF are often less able to absorb nutrients which can cause digestive problems and difficulty putting on and maintaining weight. CF often affects other parts of the body, like the liver, pancreas and bones.

CF can also affect fertility. Most men will not be able to have a child without fertility treatments. Women with CF are often able to become pregnant, though there are some factors that might impact fertility, and becoming pregnant can have an impact on their health.

There is currently no cure for CF, but there is a huge amount of research being done to develop new treatments and to understand the condition better. Daily treatments that people with CF may take to control their symptoms include:

- chest physiotherapy, exercise, inhalers and mucus thinners to clear the lungs
- inhaled antibiotics to treat or prevent lung infections
- a special diet, digestive enzyme supplements and vitamin supplements to help with digestion
- new medicines called modulators that treat the underlying cause of the disease but do not cure the condition (not everyone with CF is able to benefit from modulators)

What is carrier testing?

Carrier testing is the name for the process of testing relatives and partners of someone with CF to see if they are a carrier of a faulty CF gene.

This is sometimes called 'cascade screening'. The name 'cascade' is used because whenever a carrier is found, their relatives will then be offered testing, and so on.

What is a carrier? Are carriers healthy?

A person with only one copy of a CF gene mutation is called a carrier. A carrier does not have CF, but if their partner is also a carrier there's a chance that their children will inherit CF. See the section '**How does a child inherit CF?**' below for more information.

Some research has found that carriers have a very small chance of having mild symptoms of CF, but most carriers will not have any symptoms. Carriers do not need any CF treatment. If you have any concerns about your health, you can speak to your GP.

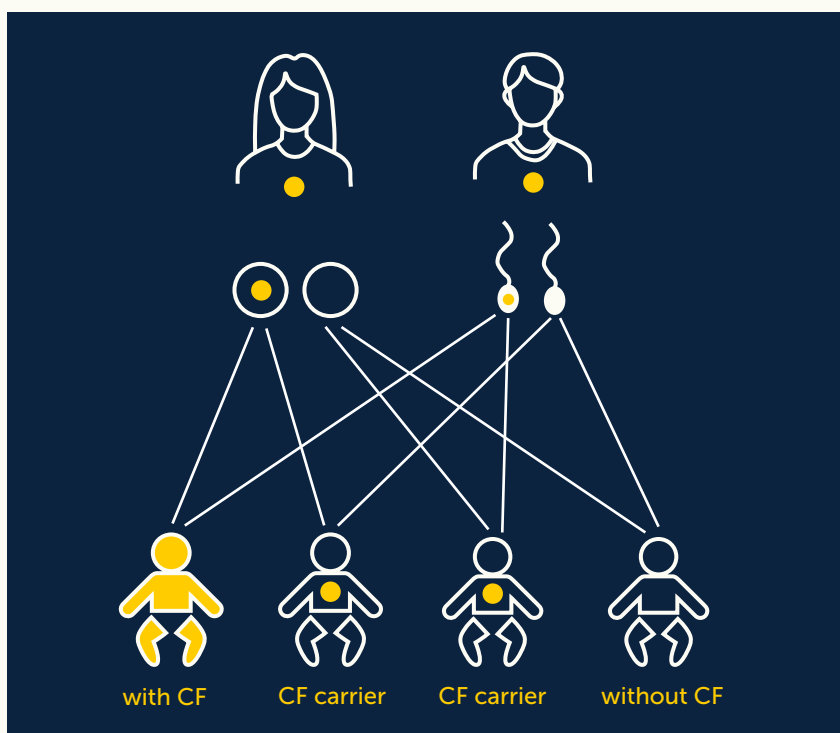
How does a child inherit CF?

For a baby to be born with CF they need to inherit two CF gene mutations – one from each parent. The parents of a child with CF must have at least one copy of a CF gene mutation, being either a carrier or a person with CF themselves.

If both parents are carriers, with only one copy of a CF gene mutation, their children have:

- a **1 in 4 (25%)** chance of inheriting two copies of a CF gene mutation and having CF
- a **1 in 2 (50%)** chance of being a carrier, like their parents, but not having CF
- a **1 in 4 (25%)** chance of not being a carrier or having CF

The chances are the same for each child the couple has.



CF gene mutations in populations

The CF gene was discovered in 1989. Since then, more than 2,000 different mutations of the CF gene have been discovered. Some mutations are more common than others. Some mutations can also cause CF to be more severe than others.

In the UK, the majority of people with CF have at least one copy of the same CF gene mutation. It is called F508del. The majority of CF carriers have one copy of F508del too.

CF is more common in people of white backgrounds, but it is seen in other ethnic groups. According to the 2020 UK CF Registry report, 93% of the CF population in the UK are white, 3% are Asian, 0.2% are Black and just over 1% are of mixed heritage. Some CF gene mutations are found more often in some ethnic groups than others. This can affect whether they are picked up in standard CF carrier tests in the UK. Speak to your GP if you are worried about this.

I have not had a carrier test. How likely is it that I am a carrier of CF?

Around 1 in 25 people in the UK are carriers of a faulty gene that causes CF – about 4% of the population. Relatives of someone with CF have a higher chance of being carriers.

If you are related to someone with CF and you **have not** been carrier tested, the table below shows your chance of being a carrier. It also tells you the chance of having a child with CF if your partner has also not been tested.

Your relationship to someone with CF	Your chance of being a carrier	Chance that you will have a child with CF
I am a biological parent of someone with CF	100%	If you and your partner are the biological parents of your child with CF, the chance of having another child with CF is 1 in 4 (25%)
		If your partner is not the biological parent of your child with CF, there is a 1 in 100 (1%) chance of you having a child with CF
I am a child of someone with CF	100% (if you do not have CF yourself)	1 in 100 (1%)
I am a sibling of someone with CF	2 in 3 (66%)	1 in 150 (0.7%)
I am an aunt or uncle of someone with CF	1 in 2 (50%)	1 in 200 (0.5%)
I am a grandparent of someone with CF	1 in 2 (50%)	1 in 200 (0.5%)
I am a first cousin of someone with CF	1 in 4 (25%)	1 in 400 (0.25%)
I am a second cousin of someone with CF	1 in 8 (12.5%)	1 in 800 (0.125%)

If you have CF and you do not know whether your partner is a carrier, the chance that you will have a child with CF is **1 in 50 (2%)**.

Who can have a carrier test?

Carrier testing is available as a free NHS service if:

- you are related to someone who has CF, or someone who is a carrier of a CF gene mutation
- your partner has CF or is related to a person with CF, or is a carrier of a CF gene mutation
- you are related to someone who is a carrier of a CF gene mutation

If you have difficulty getting a carrier test through your GP, you can show them this information or contact the Cystic Fibrosis Trust Helpline (details at the end of this factsheet).

If you are not in the list above and want to have a carrier test, it may be difficult to get one on the NHS but private testing is available. Speak to your GP to see if this is a good option for you.

How can I find out if my partner and I are carriers?

Speak to your GP if you would like to have carrier testing. If you have difficulty getting a carrier test through your GP, you can show them this information or contact the Cystic Fibrosis Trust Helpline (details at the end of this factsheet).

If you are a relative of someone with CF, your test will look at whether you're a carrier of the CF gene mutations your relative has. If your relative's CF gene mutations are not known, they will have tests first to find out what these are.

Some CF gene mutations are too rare to be picked up with these tests. If this is the case, carrier testing can be a little more complicated as more detailed genetic testing will need to be done. The parents of the person with CF will have their entire CF gene tested for the mutations they are carrying. Once these are found, other relatives can be tested for these mutations too.

If you are not related to someone with CF, but your partner is, or has CF themselves, your test will only look for the more common mutations. **Standard carrier tests pick up about 90–95% of CF gene mutations, so even if your test is negative there is still a small chance you may be a carrier of a rarer CF gene mutation.** If you later find out you are a carrier, this first negative test result is called a 'false-negative'.

How will the tests be done?

You can find out if you are a carrier for CF gene mutations with saliva or blood tests.

In a saliva test, you'll be asked to spit into a tube to collect cells that line the cheek. Genetic information in these cells will be tested for CF gene mutations.

In a blood test, a small amount of blood will be collected. In a similar way to the saliva test, the genetic information from cells in your blood will be tested for CF gene mutations.

When and how will I receive the test results?

You should be told your test results within about 10 working days. You'll usually receive your results in a letter. How you receive your test results may be different depending on where you live in the UK. If you need the results in another format, let your GP know.

The results will tell you whether you have a positive test and are a carrier, or a negative test and are not a carrier. **If your test result is negative, you should be told the chances that you could still be a carrier of a rare CF gene mutation.**

If your test is positive and you are a carrier, you will usually be invited to talk to a genetic counsellor. This can be by telephone or in person. The genetic counsellor can tell you what your result means, and about any tests and support that is available to you.


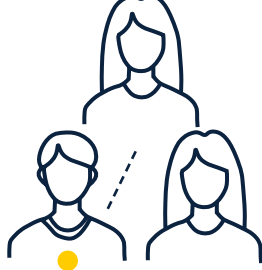
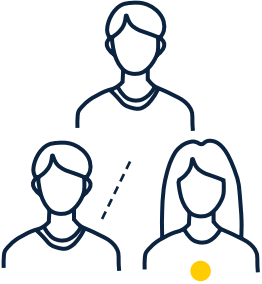

What do my test results mean?

Standard carrier tests pick up about 90–95% of CF gene mutations. Getting a negative result from one of these tests means that the chance of you being a carrier is reduced by 10 times. It means you are not a carrier of the common CF mutations that were tested for, but it does not mean you are definitely not a carrier of rarer CF mutations.

Getting a positive result from a test for any CF gene mutations means you are definitely a carrier, even if you don't have a family history of CF.

After you and your partner have a test for common CF gene mutations, the chances of you having a child with CF can be worked out more accurately.

The diagram on page 8 explains what you and your partner's chances are of having a child with CF depending on your test results and possible options for you.

 <p>'My partner and I are both carriers'</p>	<p>You have a 1 in 4 chance of having a child with CF together (see diagram on page 5).</p>	<p>A genetic counsellor will be able to tell you about any tests and the support available to you. See 'What if my partner and I are both carriers of a CF gene mutation?'</p>
 <p>'One of us is related to someone with CF and is a carrier, but the other is not a carrier'</p>	<p>You have less than a 1 in 1,000 chance of having a child with CF together.</p>	<p>A genetic counsellor will be able to tell you about any tests and support available to you. As the chance of you having a child with CF is low, tests for CF will likely not be offered during your pregnancy, but you can talk to your doctor if you are worried.</p>
 <p>'One of us is related to someone with CF but is not a carrier, and the other is a carrier'</p>	<p>You have less than a 1 in 2,000 chance of having a child with CF.</p> <p>If the relative is a full sibling of someone with CF, the chance lowers to zero.</p>	<p>The relative of a person with CF is not a carrier of the CF gene mutations they have, or any common or rare mutations that were tested for. You can request a retest to double check. A genetic counsellor will be able to tell you and your partner about the support available to you both since your partner is a carrier. As the chance of you having a child with CF is low, tests for CF will likely not be offered during your pregnancy, but you can talk to your doctor if you are worried.</p>
 <p>'My partner and I are both not carriers'</p>	<p>You have less than a 1 in 250,000 chance of having a child with CF.</p>	<p>As the chance of you having a child with CF is very low, tests for CF will likely not be offered during your pregnancy, but you can talk to your doctor if you are worried.</p>

Since 2007, all newborn babies are tested for CF and this picks up most cases. If your child is unwell when they are born, they should be tested for CF more urgently.

What if my partner and I are both carriers of a CF gene mutation?

If you and your partner are carriers, you will be offered genetic counselling. A genetic counsellor will give you information that will help you make decisions that are right for both of you. Some things they may talk about include:

- **Antenatal tests**

These are tests that are done during pregnancy to see if the baby has CF. They include chorionic biopsy (where a piece of the developing placenta is taken at 10–12 weeks) and amniocentesis (where a sample of the fluid in the womb is taken). Your genetic counsellor can tell you what your options are, and about any risks that may come with these tests.

- **Pre-implantation genetic diagnosis (PGD)**

This is a test that is used with IVF, where eggs and sperm are fertilised to make an embryo in a laboratory. PGD is used to make sure that only an embryo that does not have CF is put into the womb.

Is urgent testing available?

Yes. If you are pregnant and are concerned about whether you're a carrier, you and your partner can be tested and know the results within a few days. Speak to your GP for more information.

Home testing

There are home testing kits for inherited conditions including CF, but these are not recommended by Cystic Fibrosis Trust. Testing should be done with the support of a genetic counsellor. Home genetic testing kits cannot give you this support and the results are also less reliable.

Will it affect my insurance?

If you or your partner are carriers of a CF gene mutation, this should not affect your insurance. You do not have to declare this to insurers. This is because carriers of CF don't have the condition or any risk of ever developing CF.

The future of testing for CF

We continue to discover new CF gene mutations. This means that we can check for rarer mutations in tests and false negatives can be avoided more often.

Work is being done to develop tests that detect CF during pregnancy using a sample of the mother's blood. This is less invasive than the current tests currently available during pregnancy.

Further information

Find more information resources about living with cystic fibrosis at cysticfibrosis.org.uk/information.

Our Helpline is open 10am – 4pm Monday to Friday. It's available to anyone looking for information or support with any part of cystic fibrosis, a listening ear, or just to talk things through.

You can contact our friendly team by:

- phoning **0300 373 1000** or **020 3795 2184**

If you are worried about the cost of the call please let us know and we'll call you back.

- emailing [**helpline@cysticfibrosis.org.uk**](mailto:helpline@cysticfibrosis.org.uk)
- reaching out on all our social media channels

Visit [**cysticfibrosis.org.uk/helpline**](https://cysticfibrosis.org.uk/helpline) for more information.

We welcome your feedback on our resources.

You can also ask for this resource in large print or as a text file. Email [**infoteam@cysticfibrosis.org.uk**](mailto:infoteam@cysticfibrosis.org.uk).

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Updated in 2021 by the Cystic Fibrosis Trust and reviewed by members of the Clinical Advisory Group.

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The information in this resource does not replace any advice from your doctor or CF team. It is important that you seek your team's advice whenever you want to change your treatment.

Cystic Fibrosis Trust

Cystic Fibrosis Trust is the charity uniting people to stop cystic fibrosis (CF). We fund vital research, improve care, speak out and race towards effective treatments for all. Cystic Fibrosis Trust is here to make sure everyone with cystic fibrosis can live without limits.

Since 1964, we've supported people with cystic fibrosis to live longer, healthier lives – and we won't stop until everyone can live without limits imposed by CF.

cysticfibrosis.org.uk

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