Cystic Fibrosis Trust

Testing for cystic fibrosis carriers in families

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Contents

Introduction

A type of genetic testing called carrier testing can be used to find out if someone is a carrier of cystic fibrosis (CF). This testing is available for family members of someone who has CF and partners of known CF carriers.

This factsheet explains:

- what CF is
- what carrier testing is
- what it means to be a carrier
- the chances of being a carrier of CF
- 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 genetics specialist (expert on inherited conditions).

What is cystic fibrosis?

CF is an inherited condition caused by a variant (also known as a mutation) in the cystic fibrosis transmembrane regulator gene, or the 'CF gene'. This gene affects the balance of salt and water in cells. CF is one of the most common inherited conditions in the UK, affecting over 11,100 people.

Having CF can make breathing difficult and lead to a cough and repeated lung infections that are hard to get rid of. People with CF are often less able to absorb nutrients which can cause digestive problems. 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 a carrier?

Genes are our bodies' instructions for how to grow and function. Differences in the make-up of our genes are what make us unique. These differences are called 'variants' or 'mutations'. Most of the time, gene variants are harmless, but sometimes they can stop a gene from working properly and affect our health.

We all inherit two copies of every gene, one from each of our biological parents. A person with CF has inherited a variant in both copies of the CF gene. A carrier is a person who has inherited a variant in only one copy of the CF gene. Carriers do not have CF because they still have one copy of the CF gene working as it should. However, if their partner is also a carrier, there is a chance that their children will have CF. See page 5 for more information about how a child inherits CF.

Do carriers have symptoms?

Most carriers will not have any symptoms. Some research has found that carriers have a very small chance of having very mild symptoms, such as a higher risk of sinusitis or pancreatitis. Carriers do not need any CF treatment. If you have any concerns about your health, you can speak to your GP.

What is carrier testing?

Carrier testing is a type of genetic test to find out if you have a variant in one copy of the CF gene and are a carrier of CF. This testing is offered to relatives of someone who has CF or partners of known CF carriers.

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.

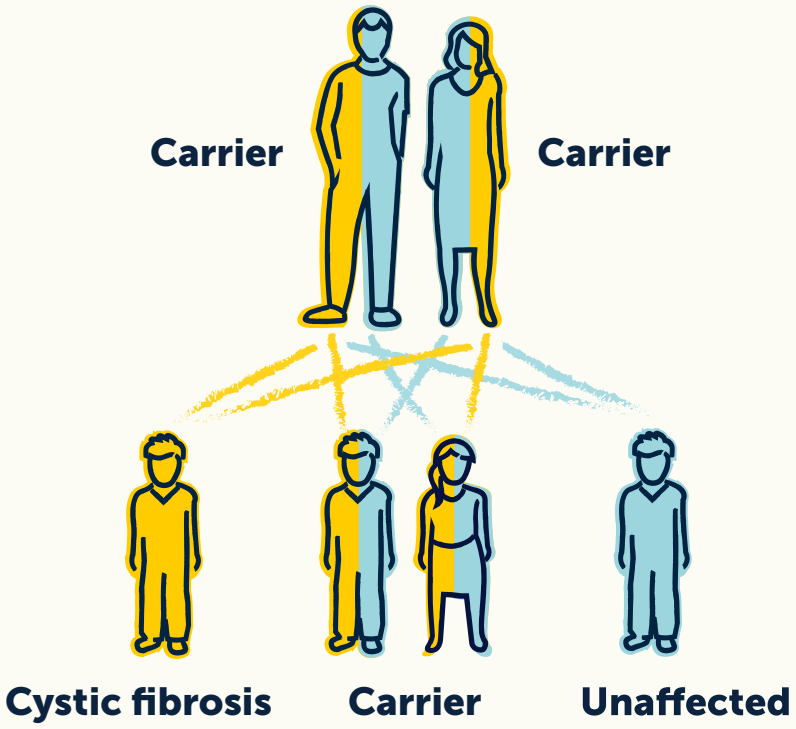
How does a child inherit CF?

A baby will be born with CF if they inherit two CF gene variants – one from each biological parent. This means that each biological parent of a child with CF is either a carrier or a person with CF themselves.

If both parents are carriers (so each has only one CF gene variant), their children have:

- a **1 in 4 (25%)** chance of inheriting two CF gene variants 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

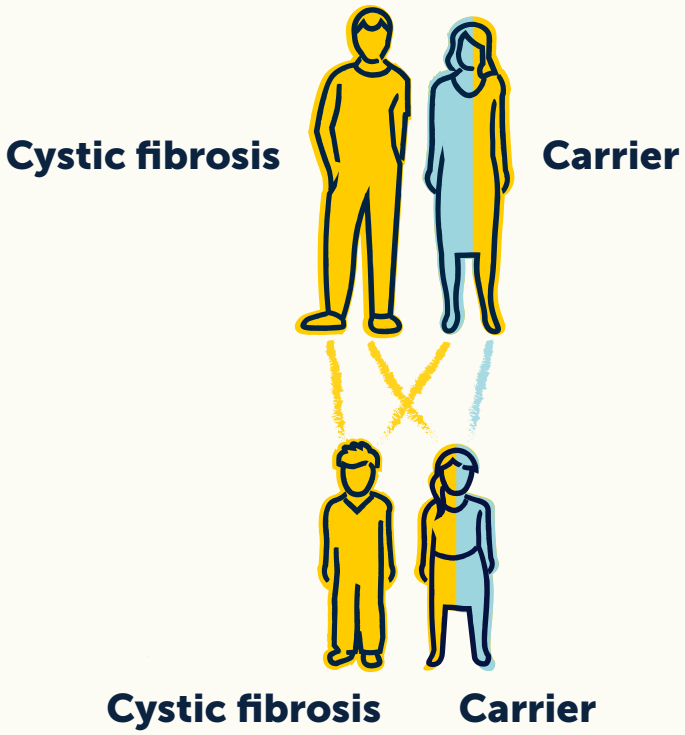
The chances are the same for each child the couple has.



If one parent has CF and one parent is a carrier, their children have:

- a **1 in 2 (50%)** chance of being a carrier, but not having CF
- a **1 in 2 (50%)** chance of inheriting two CF gene variants and having CF.

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



How common are CF gene variants in the UK?

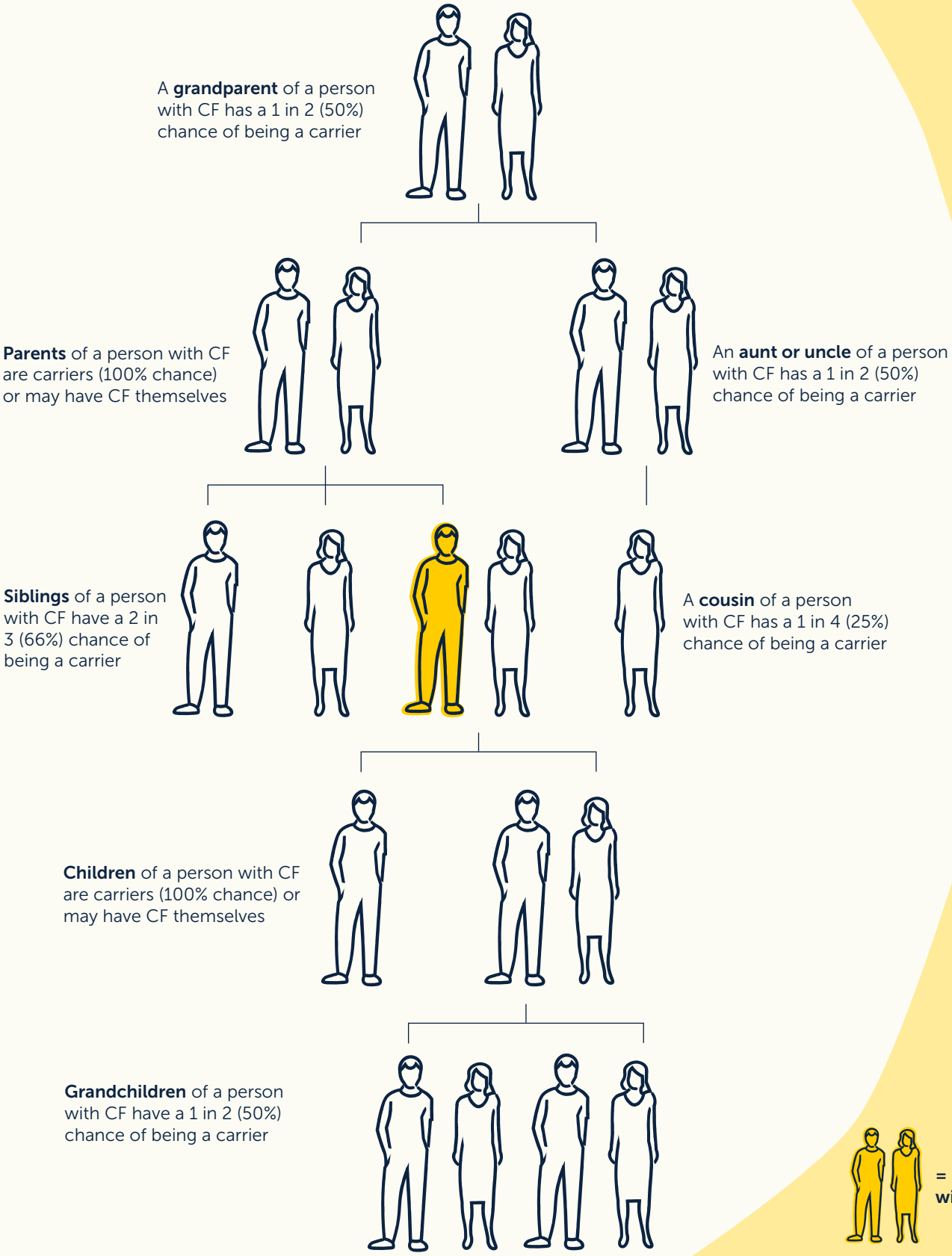
The CF gene was discovered in 1989. Since then, more than 2,000 different variants in the CF gene have been discovered. Some variants are more common than others. Some variants can also cause CF to be more severe than others.

In the UK, the majority of people with CF have at least one copy of the same CF gene variant. 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 2022 UK CF Registry report, 95% of the CF population in the UK are white, 3% are Asian, 0.3% are black and just under 1% are of mixed heritage. Some CF gene variants 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'm related to someone with CF. How likely is it that I am a carrier of CF?

Around 1 in 25 people in the UK are carriers of a CF gene variant– about 4% of the population. If you're related to someone with CF you have a higher chance of being a carrier.

The diagram below shows your chance of being a carrier, but the only way to know for sure if you're a carrier is by having a carrier test.



How likely is it that my child will have CF?

This depends on whether you are a carrier of CF or if you have CF.

- **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%). Carrier testing for your partner can help to better understand the chances.
- **If you are the biological parents of a person with CF but neither of you have CF,** the chance of you having another child with CF together is 1 in 4 (25%).
- **If you and your partner are carriers,** but neither of you have CF, the chance of you having a child with CF together is 1 in 4 (25%) (see diagram on page 5).
- **If you or your partner are related to someone with CF,** carrier testing can help to better understand your chances of having a child with CF.
- If you have CF and your partner is a carrier, there is a 1 in 2 (50%) chance you will have a child with CF (see diagram on page 6).

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 variant
- your partner has CF, is related to a person with CF, or is a carrier of a CF gene variant
- you and your partner are blood relatives (for example, first cousins) and are from an ethnic group where CF is more common (e.g. White Northern European).

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, we have written a letter to GPs to explain who is eligible for carrier testing which you can give to them. **Find this letter on our website** 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 variants your relative has. This test may also look at other common CF gene variants. If your relative's CF gene variants are not known, they will have tests first to find out what these are.

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 variants. **Standard carrier tests pick up about 85–95% of CF gene variants, so even if your test is negative there is still a small chance you may be a carrier of a rarer CF gene variant.** 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 of a CF gene variant through a blood test. If a blood test is not possible, a saliva sample can be used.

In a blood test, a small amount of blood will be collected. The genetic information from cells in your blood will be tested for CF gene variants.

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 variants.

When and how will I receive the test results?

Genetic testing can take longer than other types of health tests, often up to a few months. You can discuss with the person who ordered your test when you are likely to receive the results and how you would like to receive these.

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, the residual risk of being a carrier is low. Talk to the person that ordered your test if you want further information on this risk.**

Is urgent carrier 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 shorter timeframe. If you are pregnant, ask your GP or midwife to urgently refer you to your local clinical genetics department. This will allow your tests to be processed more urgently.

What do my test results mean?

Standard carrier tests pick up about 85–95% of CF gene variants. Getting a negative result from one of these tests means that the chance of you being a carrier is reduced by almost 10 times. It means you are not a carrier of the common CF variants that were tested for, but it does not mean you are definitely not a carrier of rarer CF variants.

Getting a positive result from a test for any CF gene variants 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 variants, the chances of you having a child with CF can be worked out more accurately. If one, or neither, of you are found to be a carrier, the chances of you having a child with CF is low. Tests for CF will likely not be offered during pregnancy, but you can talk to your doctor if you are worried. If both you and your partner are found to be carriers, you have a 1 in 4 (25%) chance of having a child with CF together (see diagram on page 5). A genetic counsellor will be able to tell you about any tests and support available to you. Ask your GP, or your midwife if you are pregnant, for a referral to clinical genetics.

Carrier testing and family planning

If you and your partner are carriers or have CF, you will be offered genetic counselling to discuss family planning options, including tests that may be available to you during pregnancy or before getting pregnant. If you are thinking of starting a family, having these discussions early on with a genetic counsellor can give you time to consider your options and, in some cases, undertake pre-pregnancy workup. A genetic counsellor will give you information to 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 villus sampling (CVS)**, where a tiny piece of the developing placenta is taken at 11–14 weeks.
- **Amniocentesis**, where a sample of the fluid in the womb is taken at 15-20 weeks. Amniocentesis can be done later in pregnancy if necessary.
- **Non-invasive prenatal diagnosis (NIPD)**, which involves taking a small blood sample from the mother after nine weeks of pregnancy onwards. This test is only available in certain circumstances, which your genetic counsellor will explain to you.

Your genetic counsellor can tell you what your options are, and about any risks that may come with these tests.

Pre-implantation genetic testing (PGT)

This is a test that is used with IVF, where eggs and sperm are fertilised to make an embryo in a laboratory. PGT is used to make sure that only an embryo that does not have CF is put into the womb. A couple must meet certain criteria to access NHS-funded PGT, which your genetic counsellor will explain to you.

For couples considering testing during pregnancy, it is important to get in touch with maternity services as soon as possible after finding out that you are pregnant to ensure a timely referral to clinical genetics. This will help to maximise the testing options available to you and give you time to consider these.

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 carrier testing affect my insurance?

If you or your partner are carriers of a CF gene variant, this should not affect your insurance (including life insurance, critical illness insurance or income protection insurance) and you do not have to declare that you are a carrier to insurers. This is because carriers of CF don't have the condition or any risk of ever developing CF.

The Code on Genetic Testing and Insurance provides more information about what an insurance company does and does not need to know about genetic testing. This code has been agreed by the UK Government and the Association of British Insurers.

The future of testing for CF

New CF gene variants are continually being discovered. This means that it should be possible to check for rarer variants in tests in the future and reduce false negatives.

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.

How to reach us:

- Call 0300 373 1000 or 020 3795 2184
- Email **helpline@cysticfibrosis.org.uk**
- Chat with us on Facebook, Twitter or Instagram
- Message us on WhatsApp on 07361 582053

Visit **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**.

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This factsheet was originally written with assistance from the late Dr Maurice Super, formerly of the Department of Clinical Genetics & CF Clinic, Royal Manchester Children's Hospital, and by Dr Martin Schwarz, Honorary Consultant Clinical Molecular Geneticist at Central Manchester & Manchester Children's Foundation Trust. This leaflet was updated in 2023/24 by Fiona Beecroft, Principal Genetic Counsellor, Birmingham Women's and Children's Hospital, and Anaar Sajoo, Genetic Counsellor, North West Thames Regional Genetics Service.

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Cystic Fibrosis Trust

Cystic Fibrosis Trust is the charity uniting people to stop cystic fibrosis. Our community will improve care, speak out, support each other and fund vital research as we race towards effective treatments for all.

We won't stop until everyone can live without the limits of cystic fibrosis.

cysticfibrosis.org.uk

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