


# Bitesize basics

## What is Cystic Fibrosis (CF) carrier testing?

Published 05 September 2024. Content is current at time of publication.  
Go to our [WEBSITE](#) for up-to-date genetic testing information.





This is one segment of an eight-part bitesize basics learning series for Primary Care, focusing on ordering Cystic fibrosis (CF) carrier testing:

1. How genetic testing is changing in Primary Care
2. What is CF carrier testing?
3. Should I order a CF carrier test?
4. Is ethnicity important in CF carrier testing?
5. Consenting for CF carrier testing
6. How to order a CF carrier test?
7. What do I do with a CF carrier test result?
8. Genetics quiz – CF carrier testing



# What is cystic fibrosis?

Cystic fibrosis (CF) is a genetic condition and is therefore inherited through families. Cystic fibrosis is caused by variants (also called “mutations”) in the *CFTR* gene.

We have two copies of the *CFTR* gene; we inherit one copy from each parent.

There are hundreds of variants that can prevent the *CFTR* gene from working correctly. Some variants are more common than others.

For more information about cystic fibrosis, see:

<https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/cystic-fibrosis/>

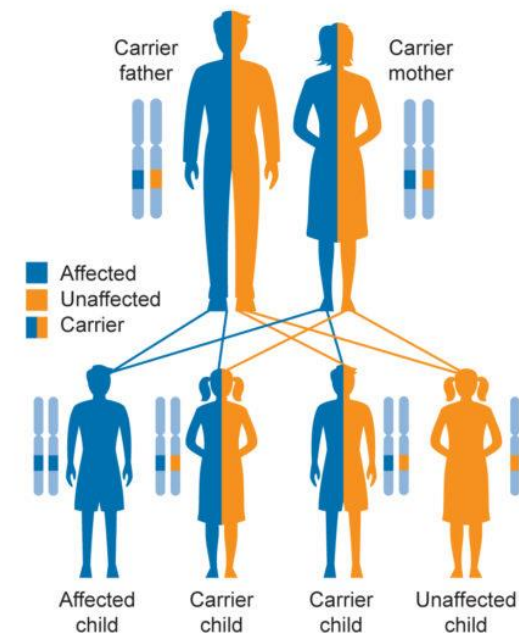
# Autosomal recessive inheritance

Cystic fibrosis is inherited through a family in an “**autosomal recessive**” pattern.

If an individual has a genetic variant in one of their two copies of the *CFTR* gene, then the second copy compensates, and they are known as an **unaffected carrier**.

If an individual has no working copies of the *CFTR* gene, they would be **affected** with cystic fibrosis.

Autosomal Recessive Inheritance





# What is a CF carrier test?

A CF carrier test is a genetic test that looks to see if an individual is a carrier of CF.

This test is offered to individuals who are **not affected** with CF but are at risk of being carriers.

This test looks for the 50 most common variants in the *CFTR* gene that cause CF in the Northern European population.

Information correct at time of publication. The number of common variants tested may change over time.



# Why have a CF carrier test?

Having a CF carrier test helps to clarify the chance that an individual is a CF carrier.

The test gives information to individuals and couples about the chance of having a child with CF and can guide family planning options.



# Contacts and information

To contact the North Thames Genomic Medicine Service, email: [nt-gmsa@gosh.nhs.uk](mailto:nt-gmsa@gosh.nhs.uk)

To contact the North Thames Genomic Medicine laboratory hub, email: [gos-tr.norththamesgenomics@nhs.net](mailto:gos-tr.norththamesgenomics@nhs.net)

Visit our website: <https://norththamesgenomics.nhs.uk>

For a more comprehensive understanding of the information provided in this bitesize resource, please download our *CF Carrier Testing Toolkit*:

<https://norththamesgenomics.nhs.uk/wp-content/uploads/2024/05/20240905-NTGMS-Toolkit-Cystic-Fibrosis-CF-Carrier-Testing-in-Primary-Care.ppt>

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