North Thames
Genomic Medicine Service

Bitesize basics
What is Cystic Fibrosis (CF) carrier testing?

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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.

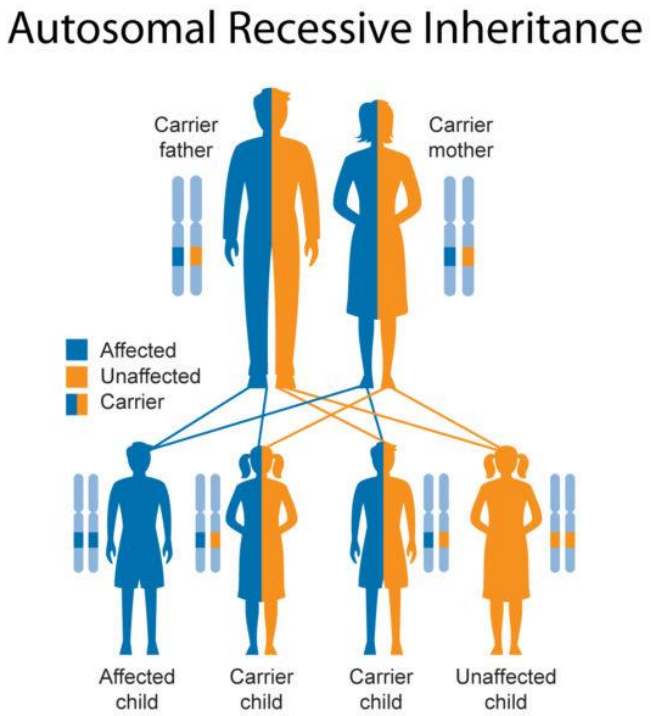
For more information about cystic fibrosis, see:
https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/cystic-fibrosis/

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.



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

To contact the North Thames Genomic Medicine laboratory hub, email: 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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