North Thames
Genomic Medicine Service

# Bitesize basics - Should I order a Cystic Fibrosis (CF) carrier test?

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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

# CF carrier testing and primary care

NHS Cystic fibrosis (CF) carrier testing can now be ordered by general practice.

The funding of genetic tests is nationally commissioned. General practice will not be charged for ordering this test.

# National Test Directory

Up-to-date eligibility criteria and information about who can order genetic tests in England are given in the NHS National Genomic Test Directory.

NHS England » National genomic test directory
https://www.england.nhs.uk/publication/national-genomic-test-directories/

The National Genomic Test Directory code for CF carrier testing is **R185**.

# Who is eligible for CF carrier testing?

Patients must be over the age of 16 years and able to give informed consent. The eligibility criteria most relevant for patients in primary care are highlighted by the blue box:

## R185 Cystic fibrosis carrier testing

### Testing Criteria

1. Prospective egg or sperm donor
2. Family history of CF in close relative (up to 4th degree, i.e. in 1st cousin's child or closer relative), or in a similar close relative of partner
3. Partner of a known CF carrier
4. Close consanguineous couple (1st cousins), AND from an ethnic group with a high carrier frequency
5. Both parents of a fetus with echogenic bowel (where both parents are available)
6. Both parents of a fetus with dilated bowel (where both parents are available)

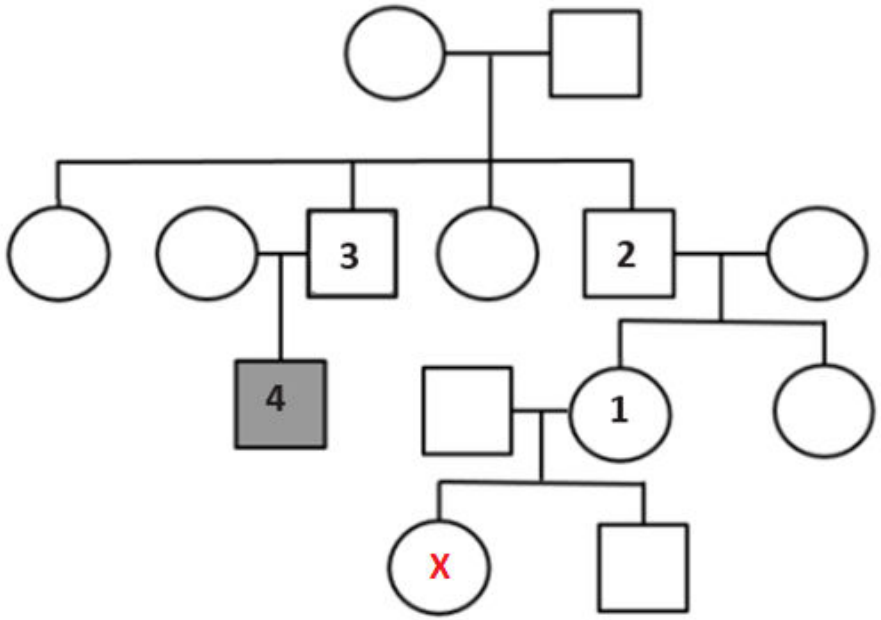
Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

Information correct at the time of publication. Please refer to the National Genomic Test Directory for up to date criteria as this is liable to change.

# Testing criterion: family history

A patient (e.g. X) with a family history of CF is eligible for CF carrier testing, if they have a relative affected by CF who is at least a fourth degree relative.

They are also eligible if they have a relative who is a CF carrier, who is at least a fourth degree relative.




**Key:**

= male
= female
= affected
= confirmed carrier

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# Why is it key to know the familial variant?

The CF carrier test only looks for 50 common *CFTR* variants. If a CF carrier test does not identify any of these variants, this only *reduces* a patient's chance of being a CF carrier.

If we do not know the familial variant, the standard CF carrier test may miss the familial variant. This is because if the relative has a **rare** variant (not one of the 50 common *CFTR* variants) the test will not identify it. This could mean that the patient is falsely reassured about their carrier status; or that their chance of being a carrier is not reduced as much as it could be.

Therefore, if your patient has a family history of CF, it is vital to include details of the affected (or confirmed carrier) family member e.g. name, DOB and where they had genetic testing. Please also include details of the CF variant(s) carried by that individual, if this is known by your patient.

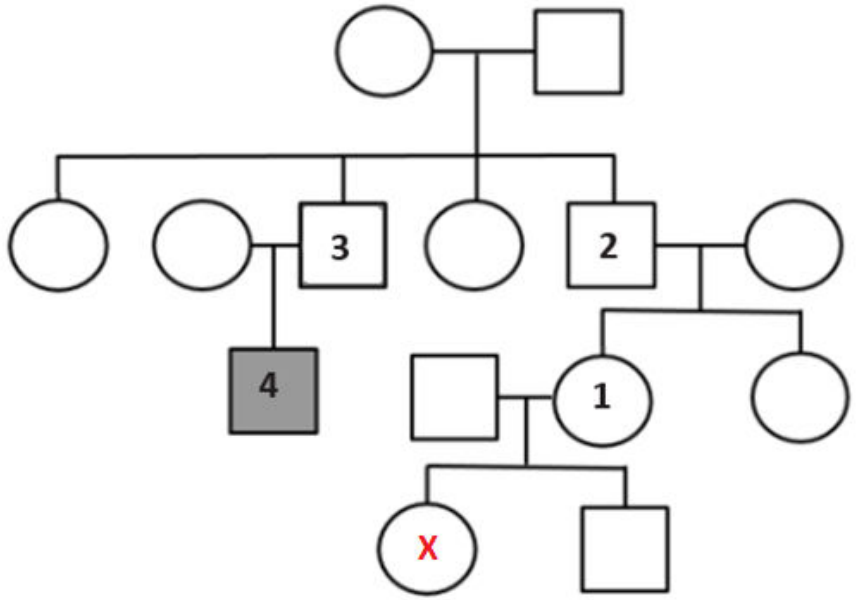
# Who is the best relative to test?

By offering patient X a CF carrier test, we could unintentionally reveal that relatives 1, 2 and 3 are also CF carriers.

In this scenario, we should ideally test relatives sequentially as follows:

- Test 3.
- If 3 is a carrier, test 2.
- If 2 is a carrier, test 1 etc.

It is not always possible to follow this process e.g. in urgent situations, or if testing is declined.

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# Discussing the reasons for sequential testing with your patient

The conversation with patient X could cover:

- Explaining the concept of revealing intervening relatives' results.
- Are the intervening relatives aware they are eligible for genetic testing?
- Have the intervening relatives refused genetic testing?
- Could the patient discuss this with the intervening relatives before having testing?
- If any of relatives 1, 2 or 3 are not carriers, this would mean that patient X does not have an increased chance of being a carrier of the familial variant.

# Testing criterion: partner of a known carrier

Any patient who has a partner who is known to be a carrier of CF or who is affected with CF is eligible for CF carrier testing.

Information correct at the time of publication. Please refer to the National Genomic Test Directory for up-to-date criteria as this is liable to change.

# Testing criterion: close consanguineous couple

Any close consanguineous couple (i.e. first cousins) are eligible for CF carrier testing, if they are both from an ethnic group where there is a high carrier frequency or if there is a known familial variant.

For more information about the impact of ethnicity and CF carrier testing, please see resource 4 in this series.



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# 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

Primary author: Ailidh Watson, Genetic Counsellor, GOSH.

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