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 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 practitioners in primary care.

The funding of genetic tests is regionally commissioned, where the cost of testing is covered by the regional Genomics Laboratory Hub.

National Test Directory

Up-to-date eligibility criteria and 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. The relevant eligibility criteria for patients in primary care is 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)

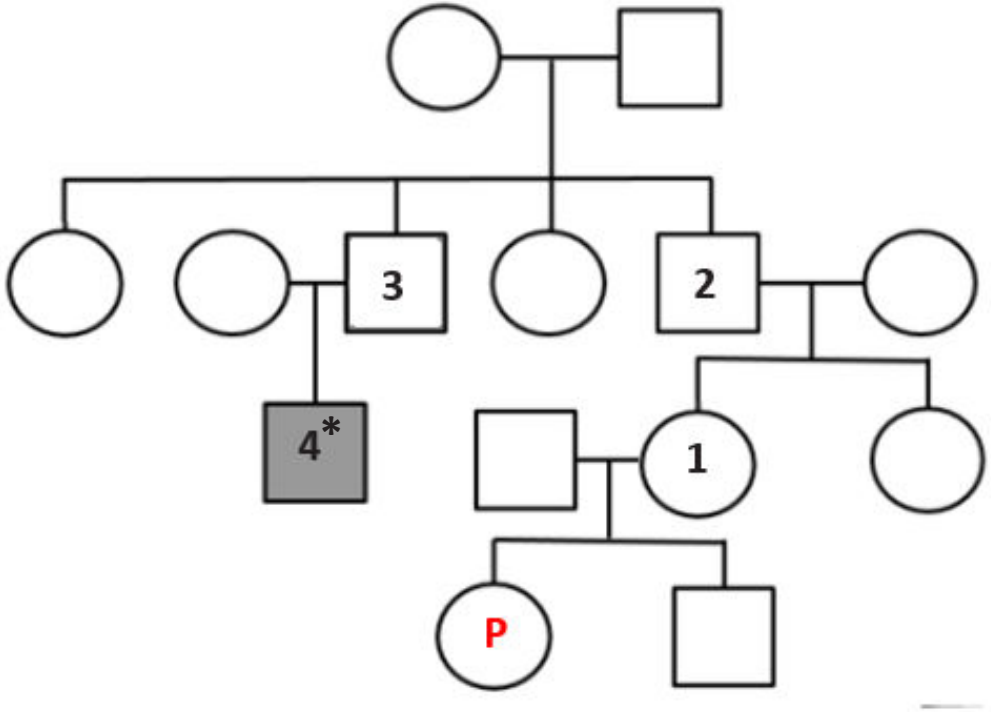
Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

Information is correct as of 27 May 2024. Refer to the National Genomic Test Directory for the most current list of available tests, as this information is subject to change.

Testing criterion: family history

A patient (e.g., "**P**") 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.



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Why is knowing the familial variant important?

If a CF carrier test does not find any of the 50 common *CFTR* mutations, this only *reduces* a patients' risk of being a carrier.

For individuals with a family history of CF, there may be a rare mutation in the family that this test does not look for.

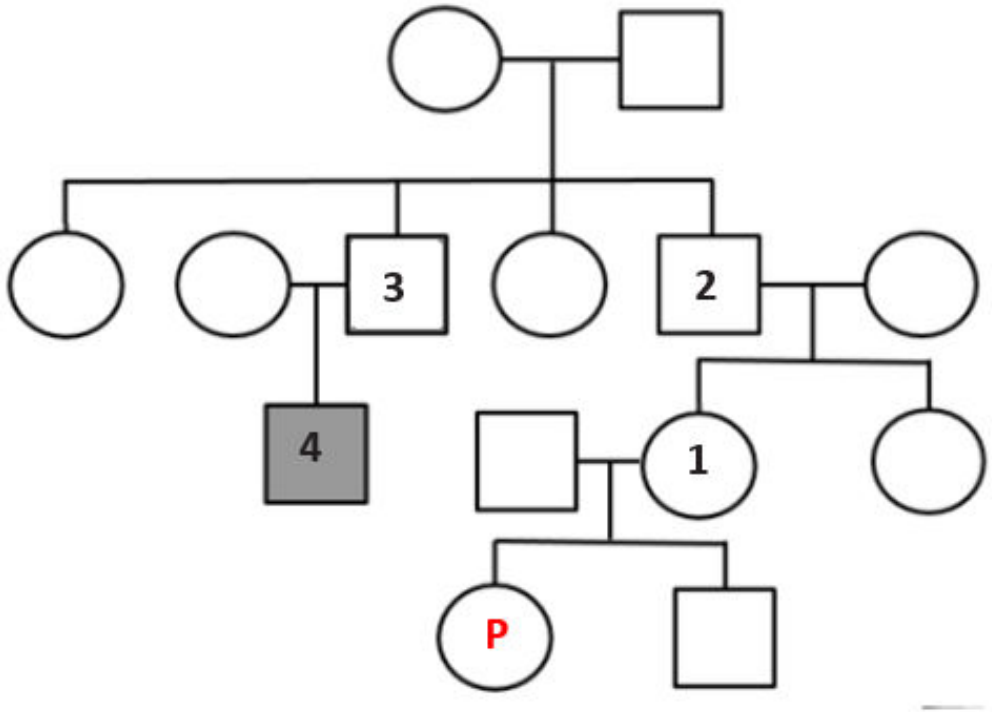
If your patient has a family history of CF, include details of the affected (or confirmed carrier) family member e.g. name and DOB. Please also include details of the CF variant(s) carried by that individual, if this information is known by your patient.

Without variant information, we might miss the familial variant with this test.

Who is the best relative to test?

By offering patient “**P**” a CF carrier test, we inadvertently reveal “intervening relatives” 1 to 3 are also CF carriers.

In this scenario, we should test sequentially: test “3”, if a carrier, test “2”, if a carrier, test “1” etc.



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“Intervening relatives” discussion

The conversation with patient “**P**” could cover:

- Explaining the concept of revealing intervening relative’s results,
- Are the intervening relatives aware they are eligible?
- Have the intervening relatives refused testing?
- Could the patient discuss this with the intervening relatives before having testing?
- If any of relatives 1 to 3 are not carriers, this could show that patient P is not at any increased chance of being a carrier.

Testing criterion: partner of a known carrier

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

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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.

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

Contact us, email: nt-gmsa@gosh.nhs.uk

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

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

Coming soon! Our *CF Carrier Testing* module video.

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