North Thames
Genomic Medicine Service

Genetics Quiz:
Cystic Fibrosis carrier testing in Primary Care

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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

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Case study quiz

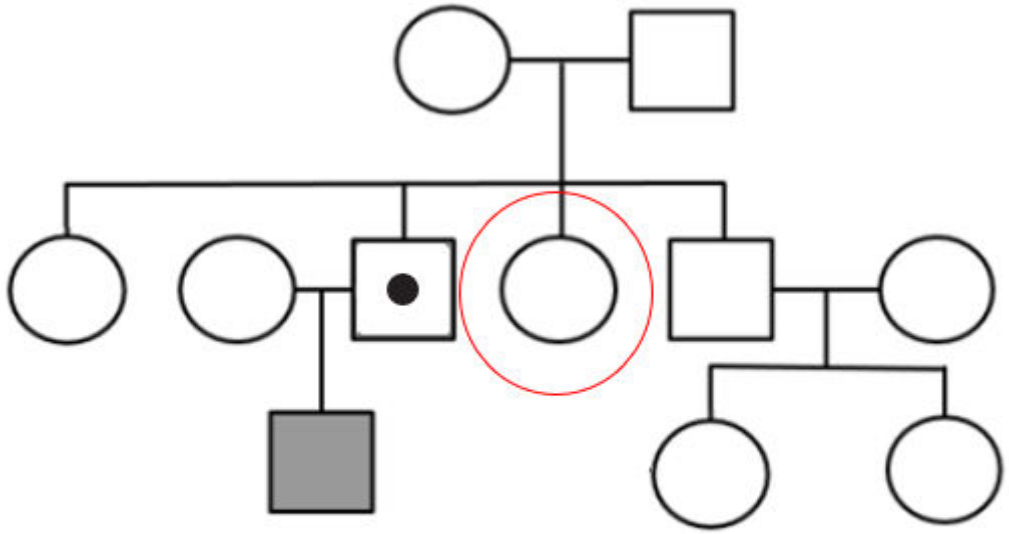
The following four case studies are designed to help test your understanding of offering cystic fibrosis (CF) carrier testing to your patients, in a primary care setting.

Each case will consist of three slides: a description of the scenario, the question and the answer.

Case study 1

A 30yo female patient (circled in red) reports that she has a family history of cystic fibrosis.

You take a family history and draw this information in the form of a pedigree (right). Her brother's son is affected with CF. She reports that her brother has had genetic testing and has been found to be a carrier of CF.



Key:

= male
= female
= affected
= confirmed carrier

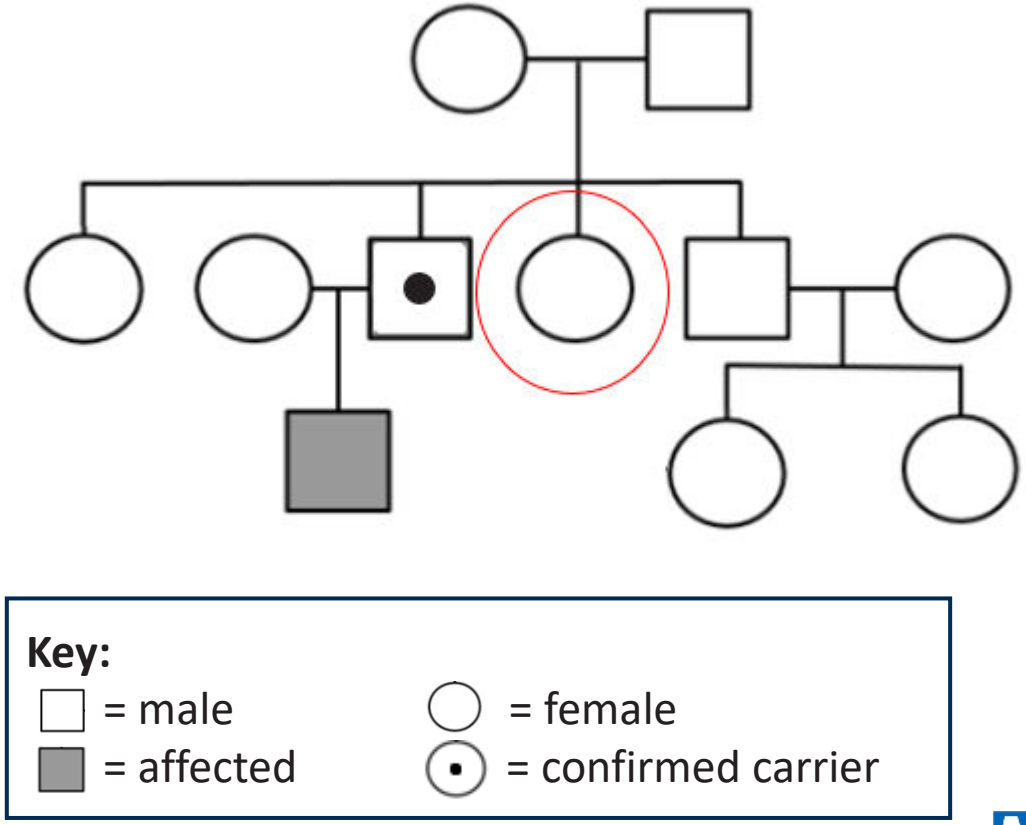
Case study 1

A 30yo female patient reports that she has a family history of cystic fibrosis.

Q1: What is the circled individual's chance of being a carrier of CF??

100%
50%
25%
12.5%
Population risk

Q2: Is the circled individual eligible for genetic testing?



Case study 1

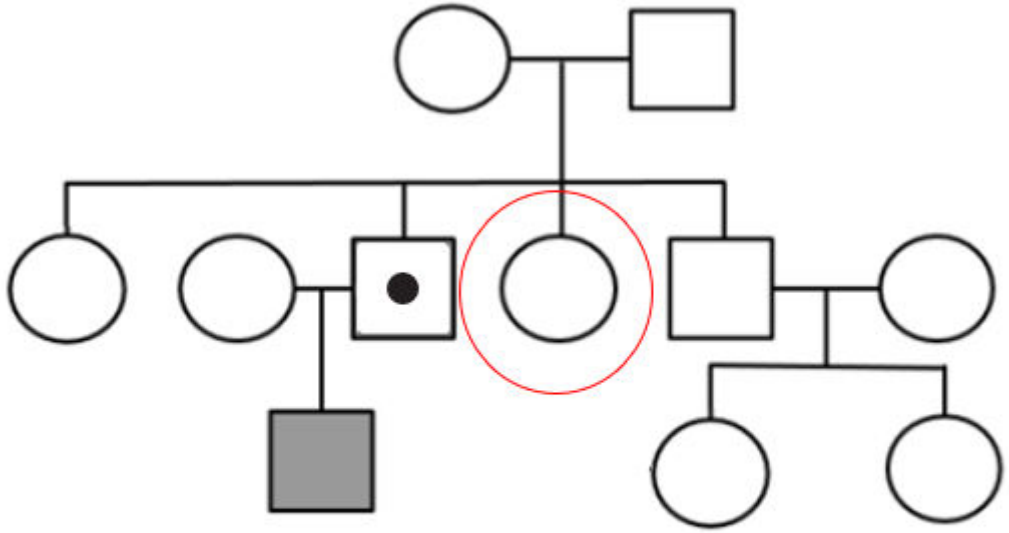
A 30yo female patient reports that she has a family history of cystic fibrosis (CF).

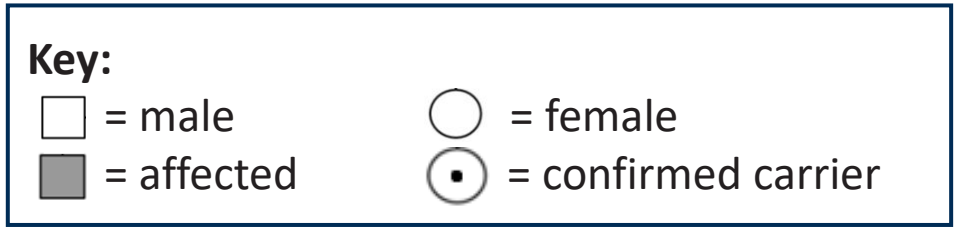
Q1: What is the circled individual's chance of being a carrier of CF??

A: 50%

Q2: Is the circled individual eligible for genetic testing?

A: Yes. Any individual with a relative (up to fourth degree) who is a CF carrier is eligible for testing.

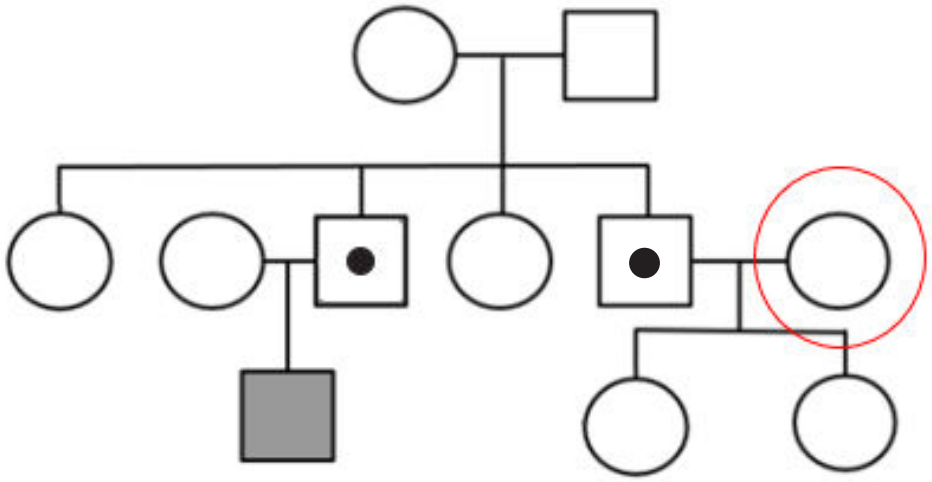


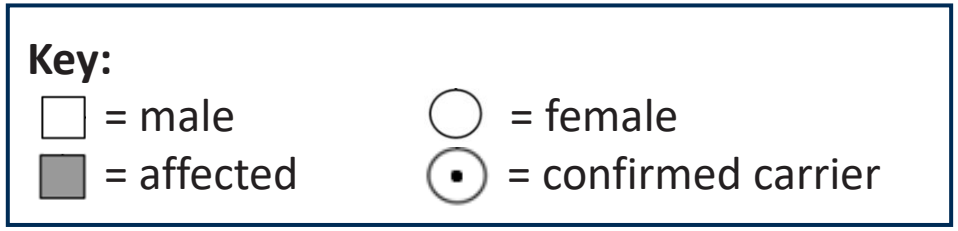


Case study 2

A 35yo female patient (circled) reports that her partner is a carrier of cystic fibrosis (CF).

You take a family history and draw this information in the form of a pedigree (right). You confirm that your patient does not have any family history of CF themselves. Your patient's ethnicity is White British.





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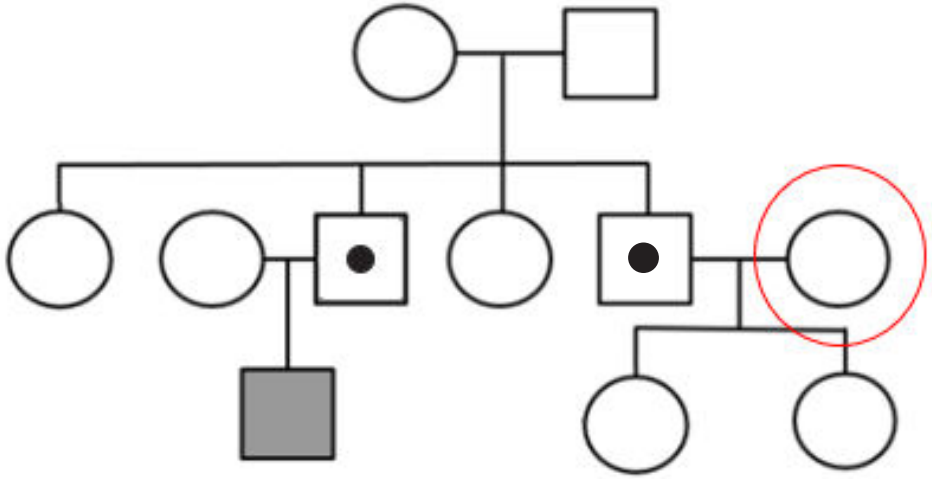
Case study 2

A 35yo female patient reports that her partner is a carrier of cystic fibrosis (CF).

Q1: Is your patient eligible for genetic testing?

Q2: What is the chance that your patient is a carrier?



Key:

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- = female
- = affected
- = confirmed carrier

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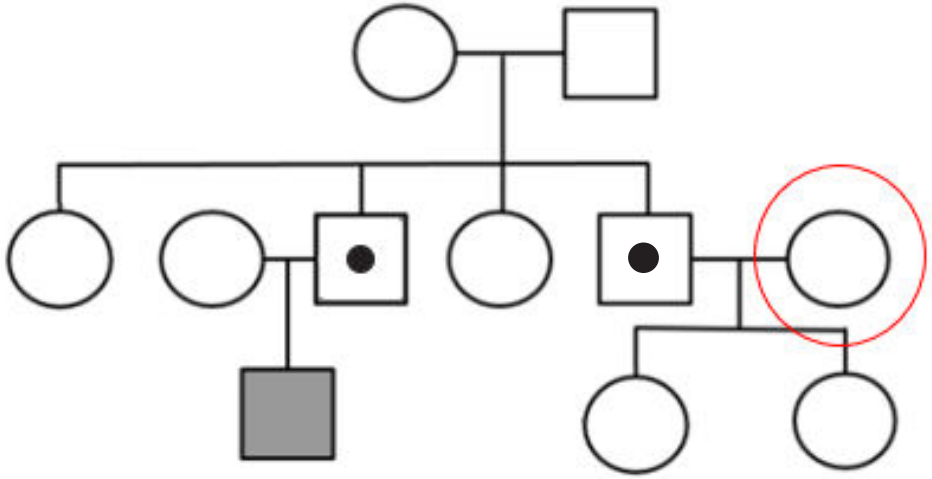
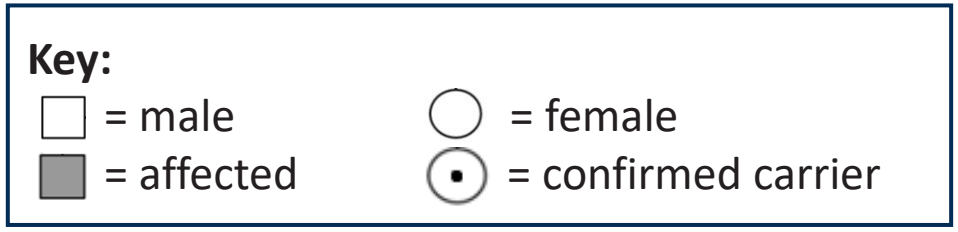
Case study 2

A 35yo female patient reports that her partner is a carrier of cystic fibrosis (CF).

Q1: Is your patient eligible for genetic testing?
A: Yes, as she meets criteria for R185 CF carrier testing.

Q2: What is the chance that your patient is a CF carrier?
A: ~1 in 25. The chance that your patient is a CF carrier is based on their ethnicity, as they do not have any family history of the condition.





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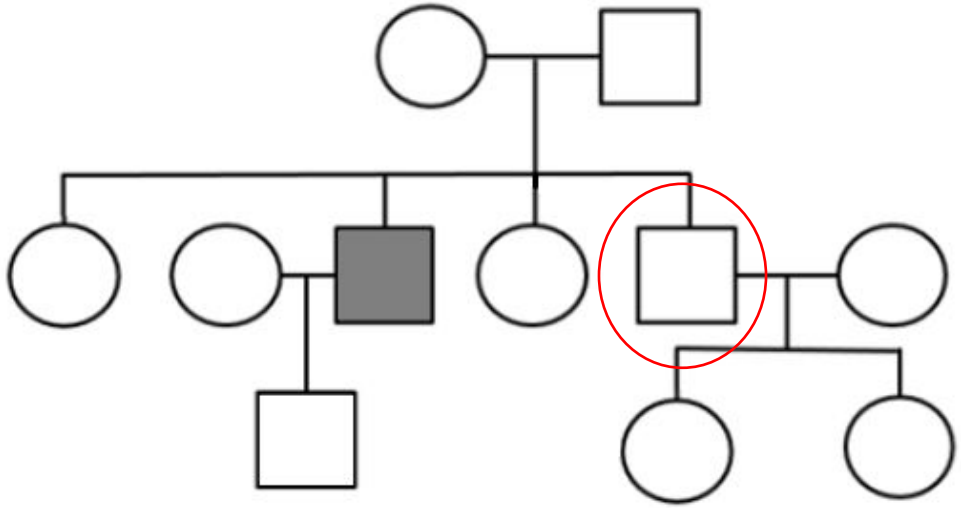
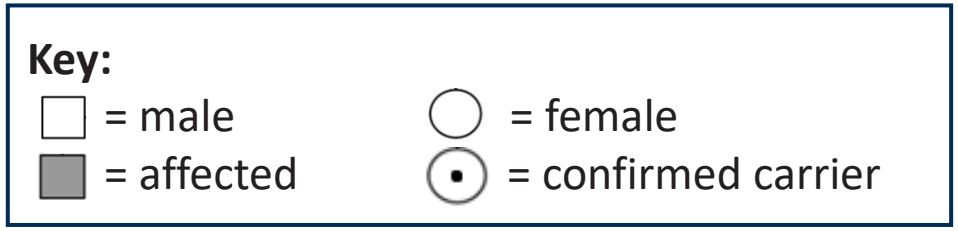
Case study 3

A 25yo male patient (circled) reports that he has a family history of cystic fibrosis.

You take a family history and draw this information in the form of a pedigree (right). His brother is affected with CF. He reports that his brother had genetic testing to confirm the diagnosis of CF.





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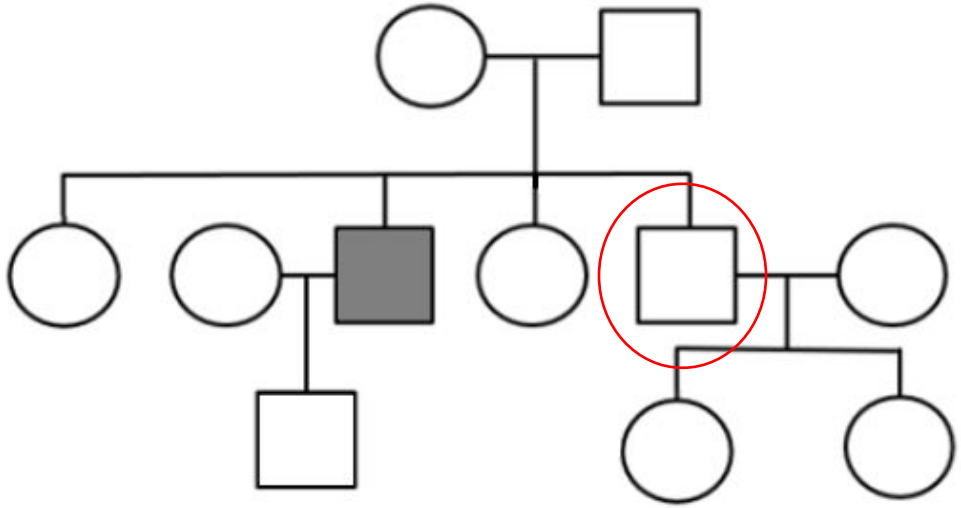
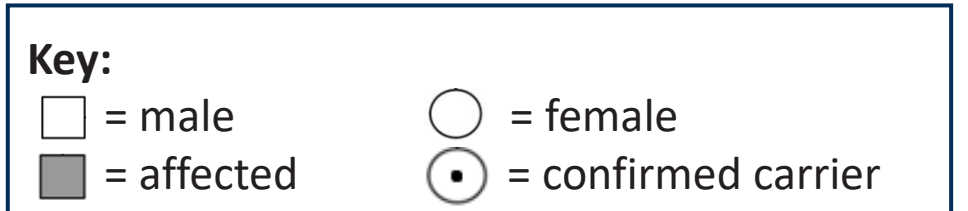
Case study 3

A 25yo male patient reports that he has a family history of cystic fibrosis.

Q: What is the circled individual's chance of being a carrier of CF??

100%
50%
~66%
25%
Population risk





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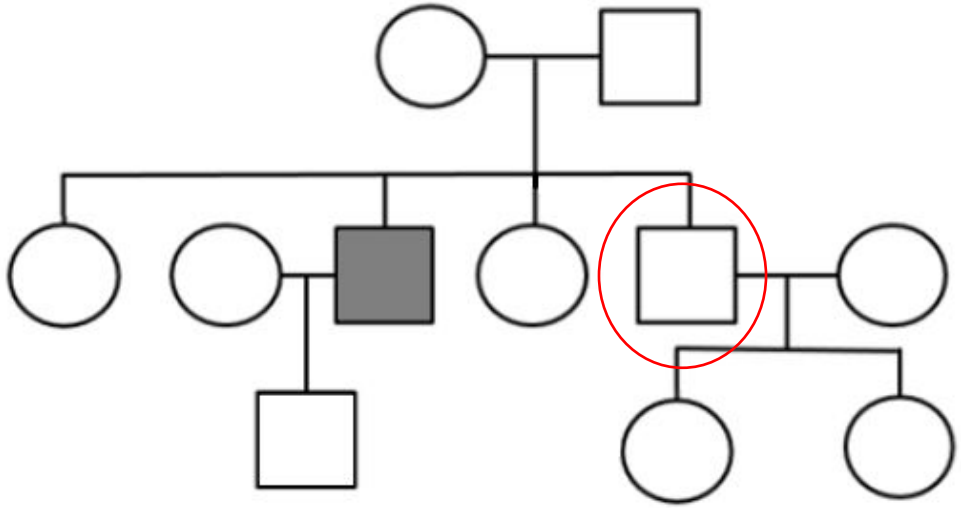
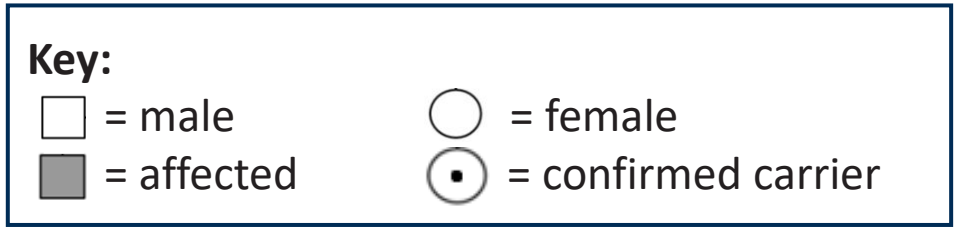
Case study 3

A 25yo male patient reports that he has a family history of cystic fibrosis.

Q: What is the circled individual's chance of being a carrier of CF??

A: ~66%.





Case study 4

You offer CF carrier testing to your patient. You have already assessed that they are eligible for testing. They have a number of questions about the test.

The following examples are to help you think about how you might answer some common patient questions.

Case study 4

You offer CF carrier testing to your patient. You have already assessed that they are eligible for testing. They have a number of questions about the test.

Q1: “What does a CF carrier test look for?”

Q2: “If my test is negative, does this mean that there is no chance of my partner and I having a child in the future with CF?”

Case study 4

You offer CF carrier testing to your patient. You have already assessed that they are eligible for testing. They have a number of questions about the test.

Q1: "What does a CF carrier test look for?"
A: The test looks for 50 variants in the *CFTR* gene. In the Northern European population, these are the most common variants that cause CF.

Q2: "If my test is negative, does this mean that there is no chance of my partner and I having a child in the future with CF?"
A: No. A negative result will reduce your chance of being a CF carrier and it will reduce the chance of you having a child affected with CF.

Common pitfalls

Q: What would an unexpected finding be for CF carrier testing?

Q: If a patient's test does not find a *CFTR* variant, why might their carrier risk only be slightly reduced?

Q: Why would we be concerned about revealing intervening relative's results?

Curious about the answers? You can find them in the *CF Carrier Testing Toolkit*, along with other information to support genetic testing in Primary Care. Visit our website - https://norththamesgenomics.nhs.uk

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/

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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