North Thames
Genomic Medicine Service

Bitesize basics – What do I do with a Cystic Fibrosis carrier test result?

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

CF carrier test results

There are three possible outcomes for a CF carrier test:

Your patient is a carrier of CF
A variant is found in one of your patient's two copies of the *CFTR* gene.

Your patient is not a confirmed carrier of CF
None of the 50 common variants are found in either of your patient's two copies of the *CFTR* gene.

Caution! This result reduces the chance that your patient is a carrier of CF, but it does not rule it out completely.

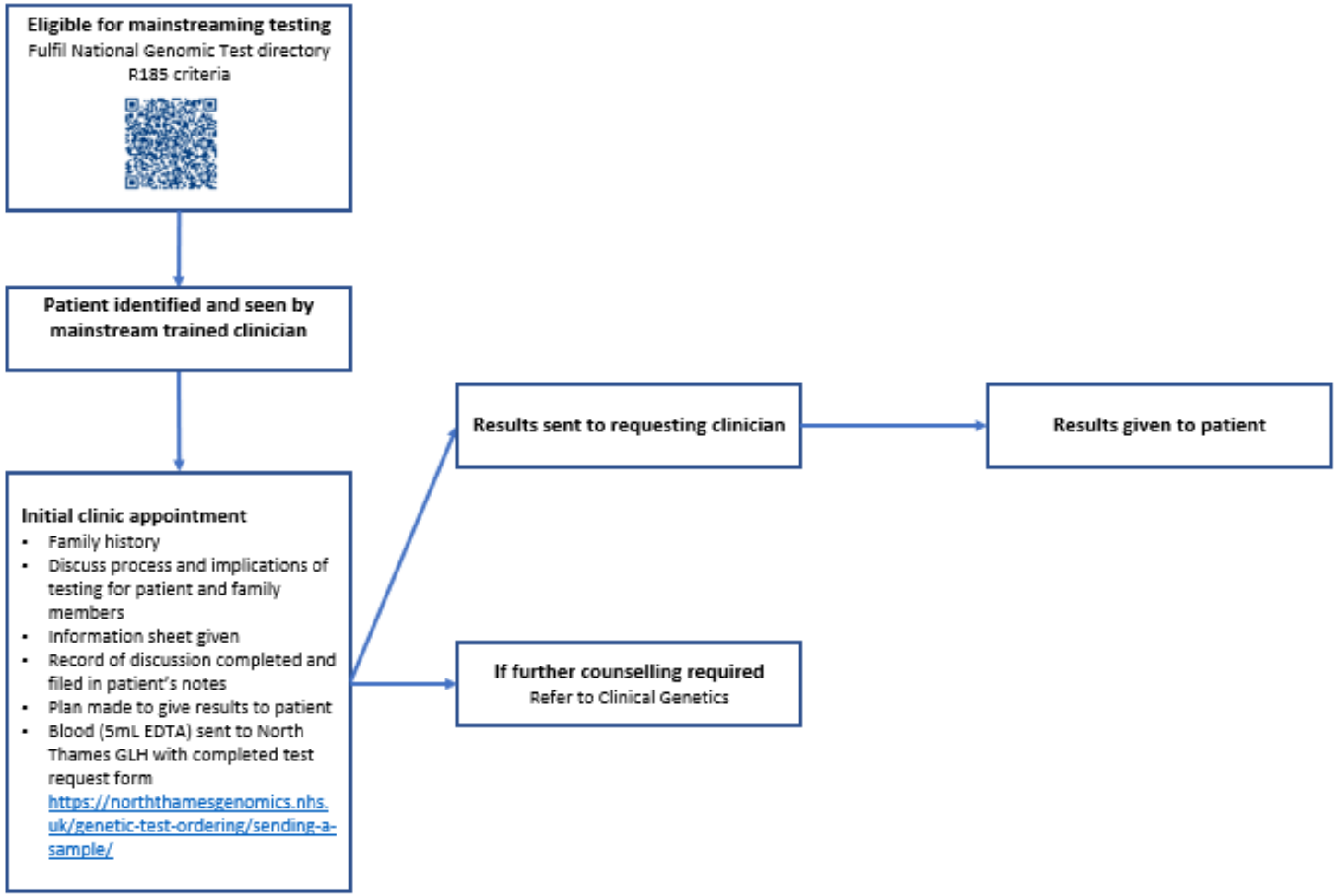
Your patient is found to have more than one *CFTR* variant
Rarely, a patient is found to have more than one *CFTR* variant.

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

NHS
North Thames
Genomic Medicine Service

CF carrier test result: what next?

- ❑ Read the genetic test report all the way through.
- ❑ During the consent discussion, you will have agreed with the patient how they wish to receive their result.
- ❑ Inform the patient of their result.



NHS
North Thames
Genomic Medicine Service

CF carrier test report: a confirmed carrier

The “result summary” section provides the take-home message about your patient’s result. The patient’s CF carrier status is given.

The “result” section describes the variant found in your patient.

Joe BLOGGS' relative is a heterozygous carrier of a *CFTR* pathogenic variant (report reference, report dated: 00/00/0000. The referral card stated him to be of XXXX ethnic origin.

Result Summary
Cystic fibrosis carrier status confirmed.

Result

JOE BLOGGS is heterozygous for the c.1521_1523del p.(Phe508del) cystic fibrosis pathogenic variant assumed to also be carried by his relative, but does not have any of the further 49 cystic fibrosis pathogenic variants tested for. This confirms his carrier status.

Further Testing

Carrier testing can be offered to adult relatives of JOE BLOGGS. It is recommended that any future partner of JOE BLOGGS also be considered for carrier testing. Please refer to Clinical Genetics so that counselling can be given.

Reported by:	Date:
Authorised by:	Date:

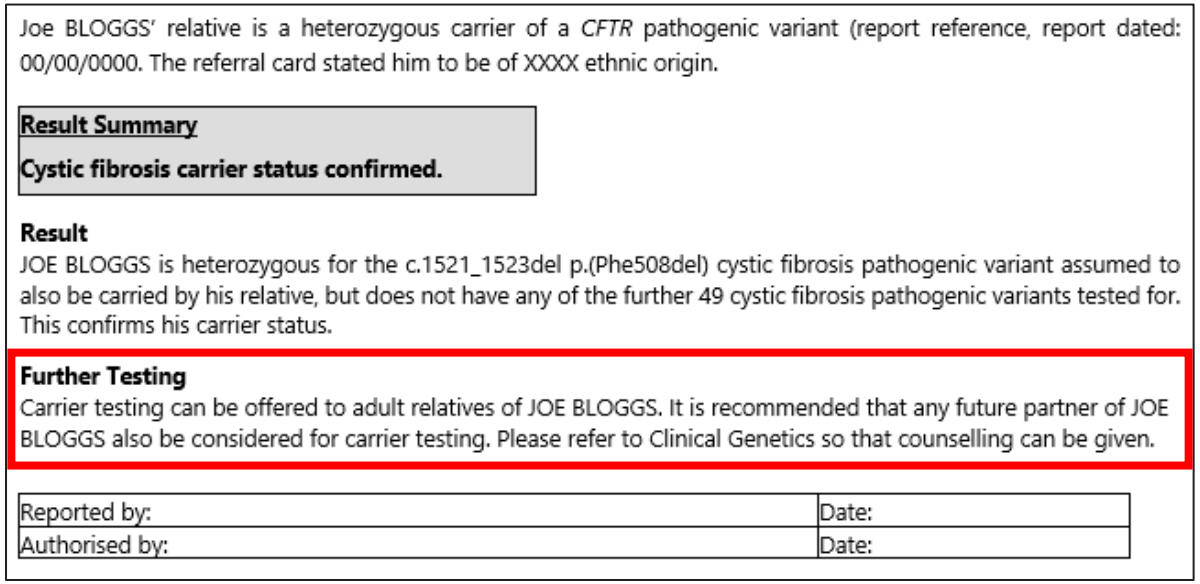
Information is correct at time of publishing. The number of common variants tested may change over time.

NHS
North Thames
Genomic Medicine Service

CF carrier test report: a confirmed carrier

The “further testing” section describes what the result means for a patient and their relatives.

- Their partner can access CF carrier testing.
- Their future children can access CF carrier testing.
- Their relatives may also be CF carriers. The relatives can access genetic testing.



Joe BLOGGS' relative is a heterozygous carrier of a *CFTR* pathogenic variant (report reference, report dated: 00/00/0000. The referral card stated him to be of XXXX ethnic origin.

Result Summary

Cystic fibrosis carrier status confirmed.

Result

JOE BLOGGS is heterozygous for the c.1521_1523del p.(Phe508del) cystic fibrosis pathogenic variant assumed to also be carried by his relative, but does not have any of the further 49 cystic fibrosis pathogenic variants tested for. This confirms his carrier status.

Further Testing

Carrier testing can be offered to adult relatives of JOE BLOGGS. It is recommended that any future partner of JOE BLOGGS also be considered for carrier testing. Please refer to Clinical Genetics so that counselling can be given.

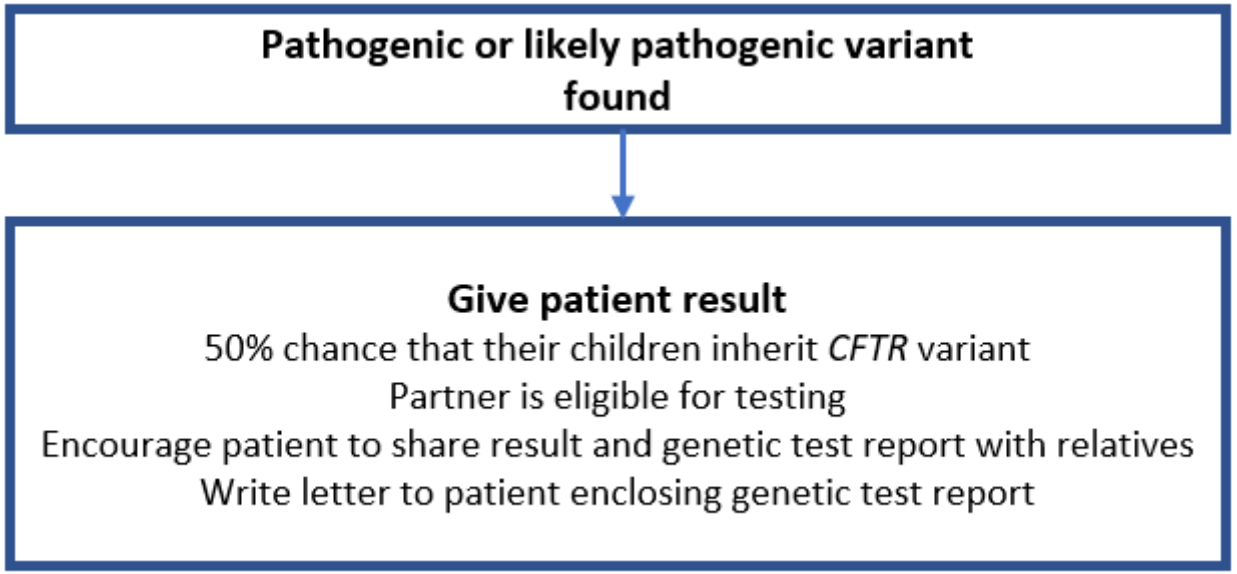
Reported by:	Date:
Authorised by:	Date:

If their partner has had CF carrier testing, a summary of their results and the implications for future children will be provided here.

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

What next: a confirmed carrier

A variant is found in one of your patient's two copies of the *CFTR* gene.



CF carrier test report: not a confirmed carrier

The “result summary” section provides the take-home message about your patient’s result.

The “result” section describes your patient’s remaining risk of being a CF carrier.

Joe BLOGGS father is a heterozygous carrier of the CFTR pathogenic variant c.1521_1523del p.(Phe508del) (report reference XXX, reported 00/00/000). The referral card stated him to be of White British ethnic origin.

Result Summary

Cystic fibrosis carrier risk reduced from 1/2 to 1/351.

Result

Joe BLOGGS does not carry any of the 50 cystic fibrosis pathogenic variants tested which includes the c.1521_1523del p.(Phe508del) pathogenic variant carried by his father. This reduces his carrier risk from 1/2 to 1/351 using UK population data.

Recommended action

No further testing will be carried out.

Reported by:	Date:
Authorised by:	Date:

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

What next: not a confirmed carrier

"What if the risk of my patient being a carrier hasn't been reduced significantly?"

For example:

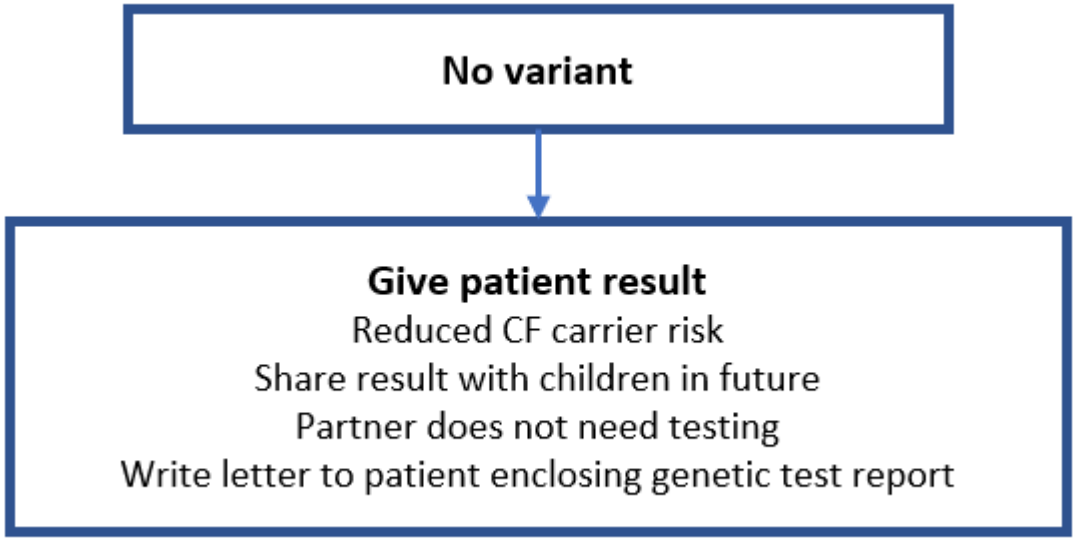
Result Summary
Cystic fibrosis carrier risk reduced from 1/2 to 1/8

This usually happens when a patient has a family history of CF, but the laboratory was not made aware of the *CFTR* variant(s) in this patient's family when this test was requested.

The risk is not significantly reduced because the laboratory cannot guarantee that the test looked for the specific *CFTR* variant(s) in your patient's family. You can contact the laboratory to provide this information for an updated genetic test report.

What next: not a confirmed carrier

None of the 50 common variants were found in either of your patient's two copies of the *CFTR* gene.



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

CF carrier test report: more than one *CFTR* variant

Rarely, a patient is found to carry two or more variants in the *CFTR* gene. There are two possibilities with this result:

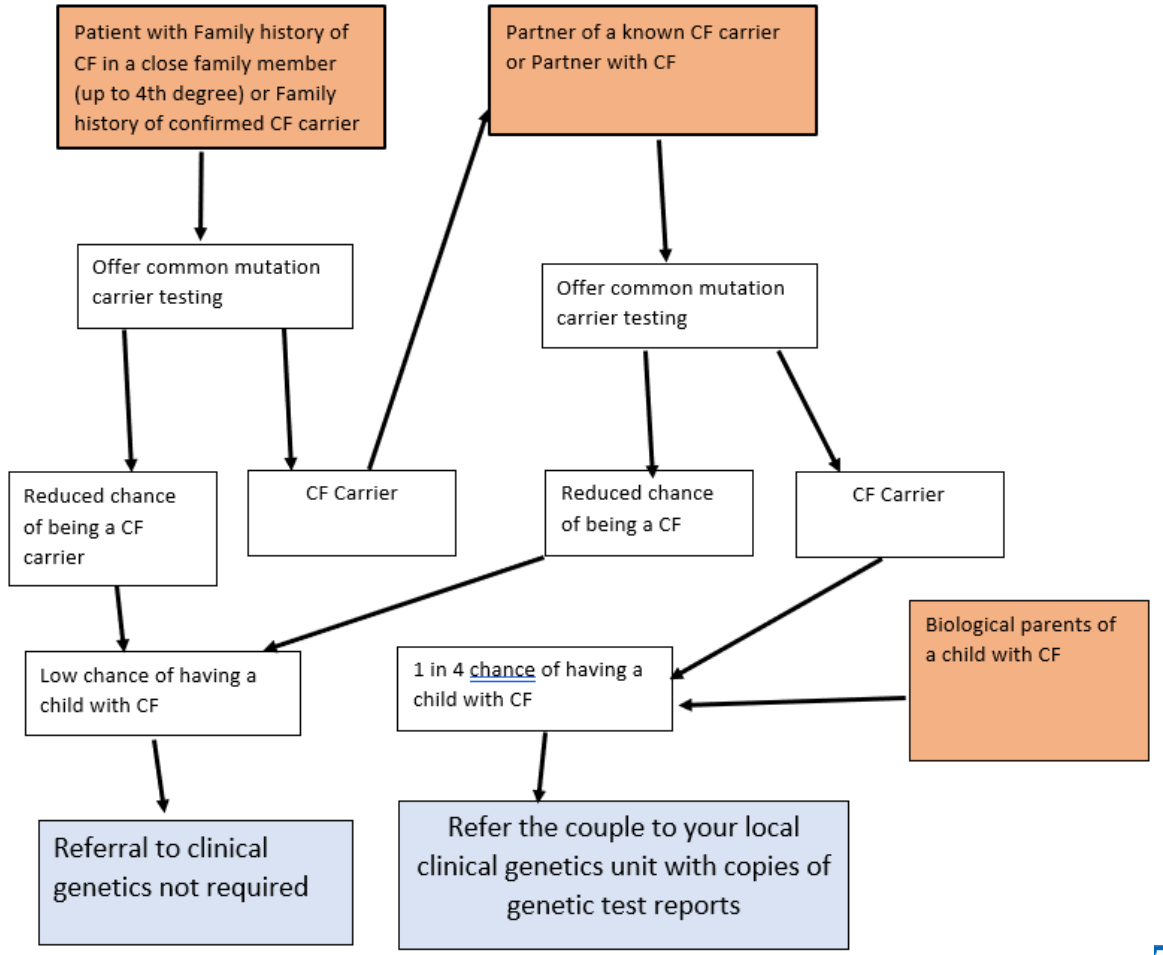
- If both variants are in the same copy of the gene, this patient is still only a CF carrier.
- If the variants are present in each copy of your patient's *CFTR* gene, this patient may have a mild form of CF.

If a patient is found to carry more than one *CFTR* variant, refer to Clinical Genetics for interpretation of this result. Further testing may be offered if appropriate.

When to refer to Clinical Genetics

See flow chart plus:

- Your patient is eligible for CF carrier testing but needs further support with deciding whether to have the test.
- As a confirmed carrier they have questions that you cannot answer.
- As a confirmed carrier they are struggling to cope with their result.
- Your patient has had two or more *CFTR* variants identified by the test.



NHS
North Thames
Genomic Medicine Service

What to include in a Clinical Genetics referral

Send enough information to show that the patient needs to be seen by Clinical Genetics.

This may include:

- The reason for referral
- Genetic test report of your patient (if done)
- A description of the reason that they were eligible for testing (if done)

Regional Clinical Genetics Service

To make a referral, GPs should use the electronic referral system (eRS). All other specialists can email the team using a secure nhs.net email

A patient's regional Genetics service is determined by GP postcode (listed below).

North West Thames

Web: www.lnwh.nhs.uk/genetics
Referrals: LNWH-tr.geneticreferrals@nhs.net
Clinical Queries: LNWH-tr.nwtrgsclinicalgenetics@nhs.net

N: 2,3,11,12,20 - **NORTH LONDON**
NW: 2,4,5,6,7,8,9,10,11
EN: 4,5,6,7,8,10,11
W: 1 – 14 - **WEST LONDON**
SW: 1,3,5,6,7,10 - **SOUTH WEST LONDON**
SW1P
SW1V

St Albans	AL	**BEDFORDSHIRE, HERTFORDSHIRE & MDDX**
Harrow	HA	
Hertfordshire	HP	
Luton	LU	
Bedfordshire	MK	40-45
Stevenage	SG	
Twickenham	TW	3, 4, 5, 6, 7, 8, 13 and 14
Uxbridge	UB	[UB9 to Oxford]
Watford	WD	

North East Thames

Web: https://www.gosh.nhs.uk/wards-and-departments/departments
Referrals: Through e-RS
Clinical Queries: gos-tr.clinicalgenetics@nhs.net

EN (Enfield): 1,2,3,9
N (London): 1,4,5,6,7,8,9,10,13,14,15,16,17,18, 19,21,22
E (London 1 –
18) EC (London
1- 4)
WC (London 1& 2)
NW (London 1, 3, 5 & 6)
RM (Romford)
IG (Ilford)
SS (Southend, Basildon and Thurrock)
CM (Chelmsford, Epping and Harlow)
CO (Colchester)
All referrals from Essex

NHS
North Thames
Genomic Medicine Service

What should a patient expect in a Clinical Genetics appointment?

In an appointment where a couple are both CF carriers, the Genetics clinician would discuss:

- CF inheritance
- Reproductive options depending upon each couple's individual circumstances and the particular *CFTR* variants that they each carry
- How the patients can discuss their results with other at-risk relatives, including giving supporting letters about the CF results for relatives
- Psychosocial impact of the results

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.

NHS
North Thames
Genomic Medicine Service