North Thames
Genomic Medicine Service

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

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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 test results

There are three possible outcomes to a CF carrier test:

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

Your patient is not confirmed carrier of CF.
None of the 50 common variants have been 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 does not rule it out entirely.

Your patient has an unexpected finding
A rare result when your patient is found to carry two variants in the *CFTR* gene.

Information is correct as of 27 May 2024. The number of common variants tested may change over time.

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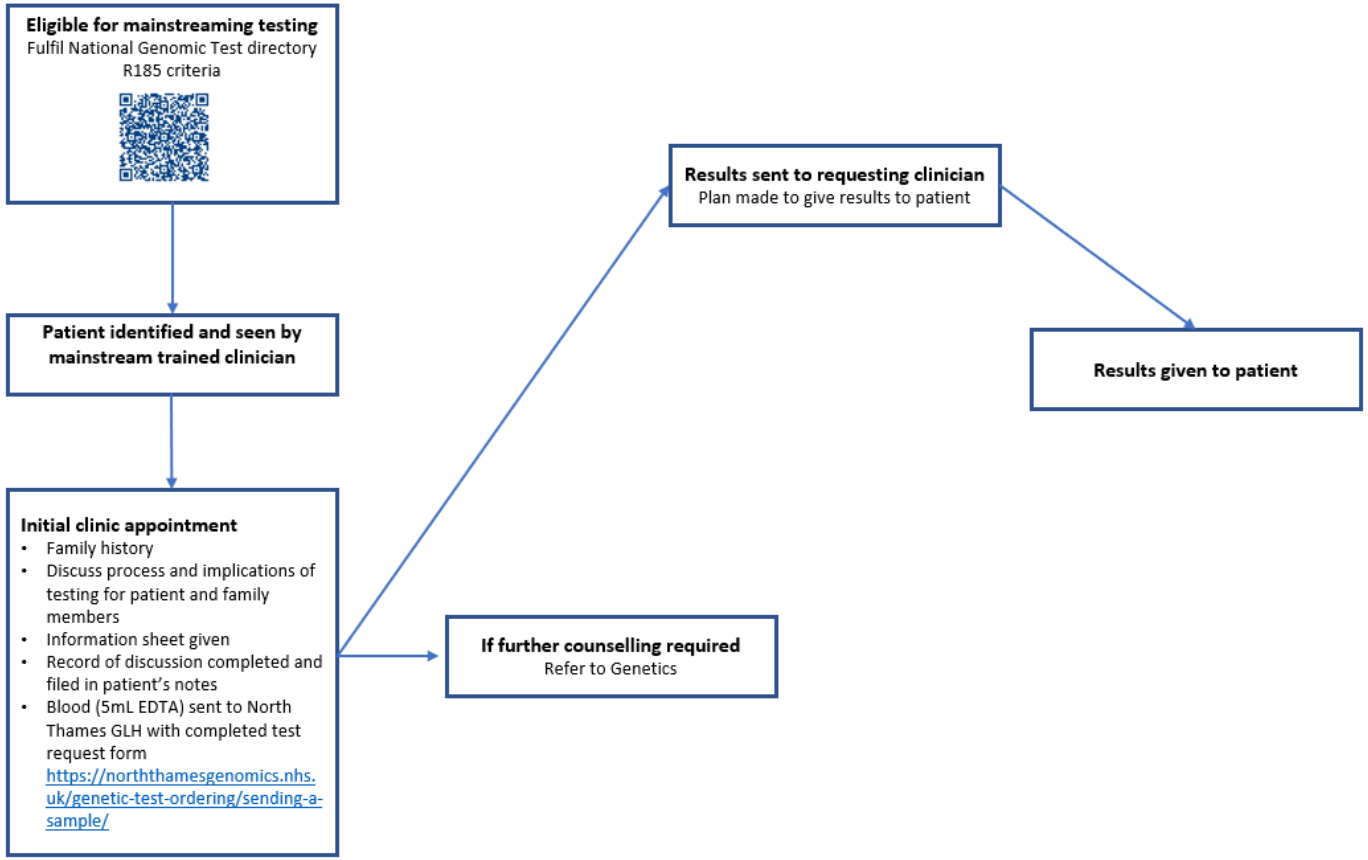
CF carrier test result: what next?

- ❑ Read the genetic test report all the way through.
- ❑ During the consent discussion, you'll agree with the patient how they'll receive their result.
- ❑ Inform the patient



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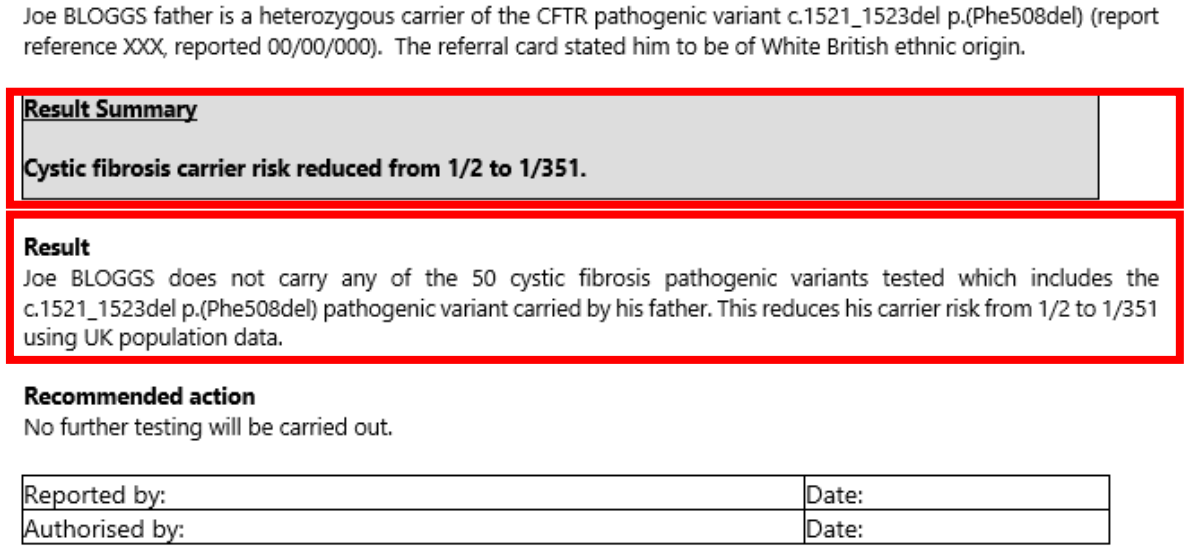
CF carrier test report: not confirmed carrier

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

The patient’s remaining risk of being a CF carrier is stated.

The “result” section gives a little more detail to the result.



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:

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What next: not confirmed carrier

“What if the risk of my patient being a carrier hasn’t been reduced significantly?”

For example:

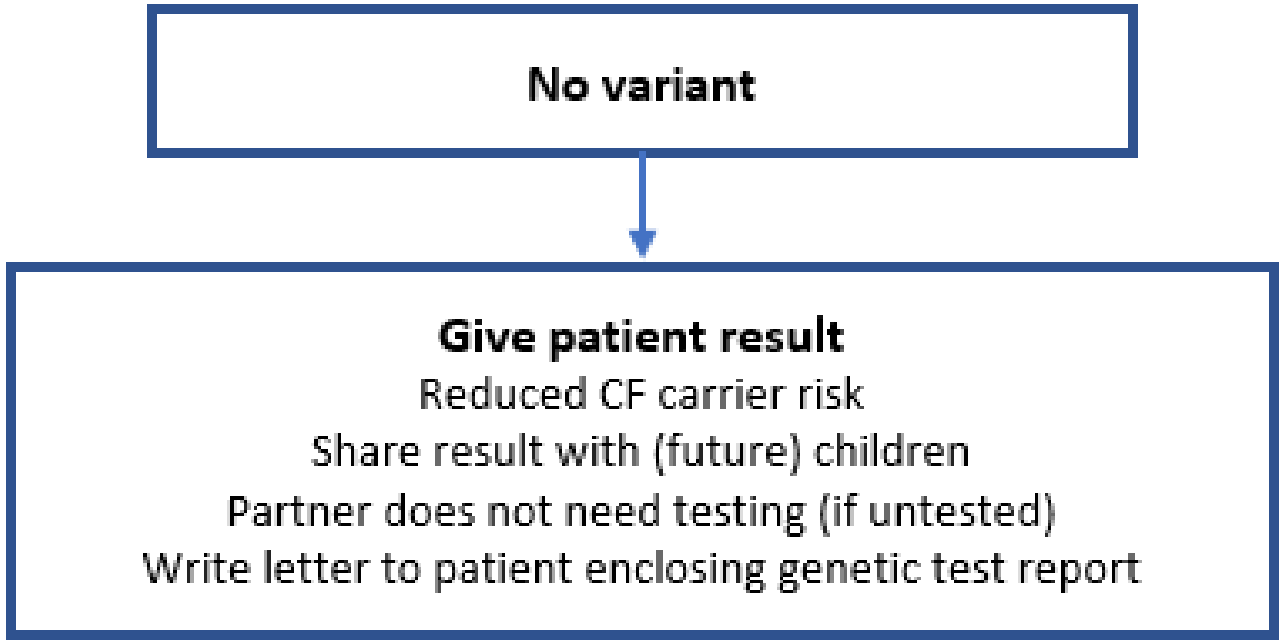
Result Summary
Cystic fibrosis carrier risk reduced from 1/2 to 1/8

This usually happens when your patient has a family history of CF and the laboratory was not given information about the variants in this patients family.

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

What next: not confirmed carrier

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



Information is correct as of 27 May 2024. The number of common variants tested may change over time.

CF carrier test report: confirmed carrier

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

The patient’s CF carrier status is given.

The “result” section gives a little more detail to the result. It describes the exact 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 Caucasian 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.

Implications of result

The risk of JOE BLOGGS and their partner *** having a child affected with cystic fibrosis is therefore reduced to less than that of the general population OR 1 in XXX. The situation is uninformative for prenatal diagnosis, which is therefore not indicated.

Further Testing

Carrier testing can be offered to the 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.

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CF carrier test report: confirmed carrier

The “implications of result” section describes what this result means for a patient.

- Their family members are at risk of also being carriers. The family members can also access genetic testing.
- Their partner can access CF carrier testing.
- Their future children can access CF carrier testing.

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

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Result Summary

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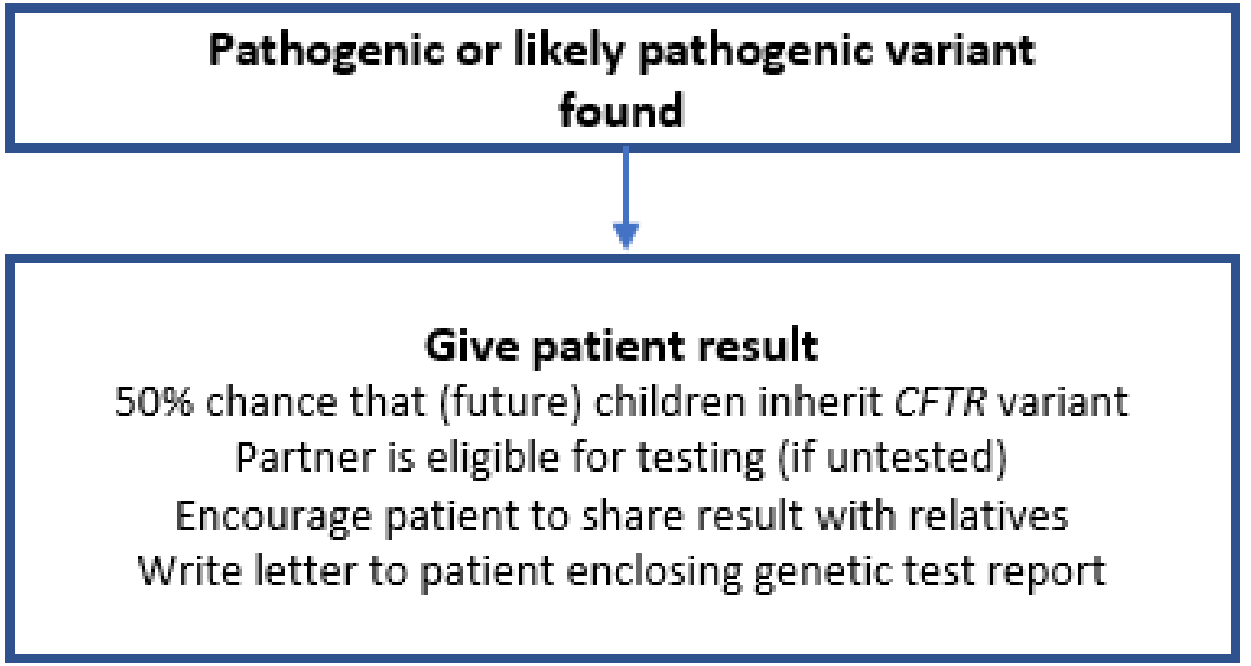
What next: confirmed carrier

- We have found a variant in one of your patient's two copies of the *CFTR* gene.



CF carrier test report: unexpected findings

A patient is found to carry two variants in the *CFTR* gene. There are two possibilities with this result:

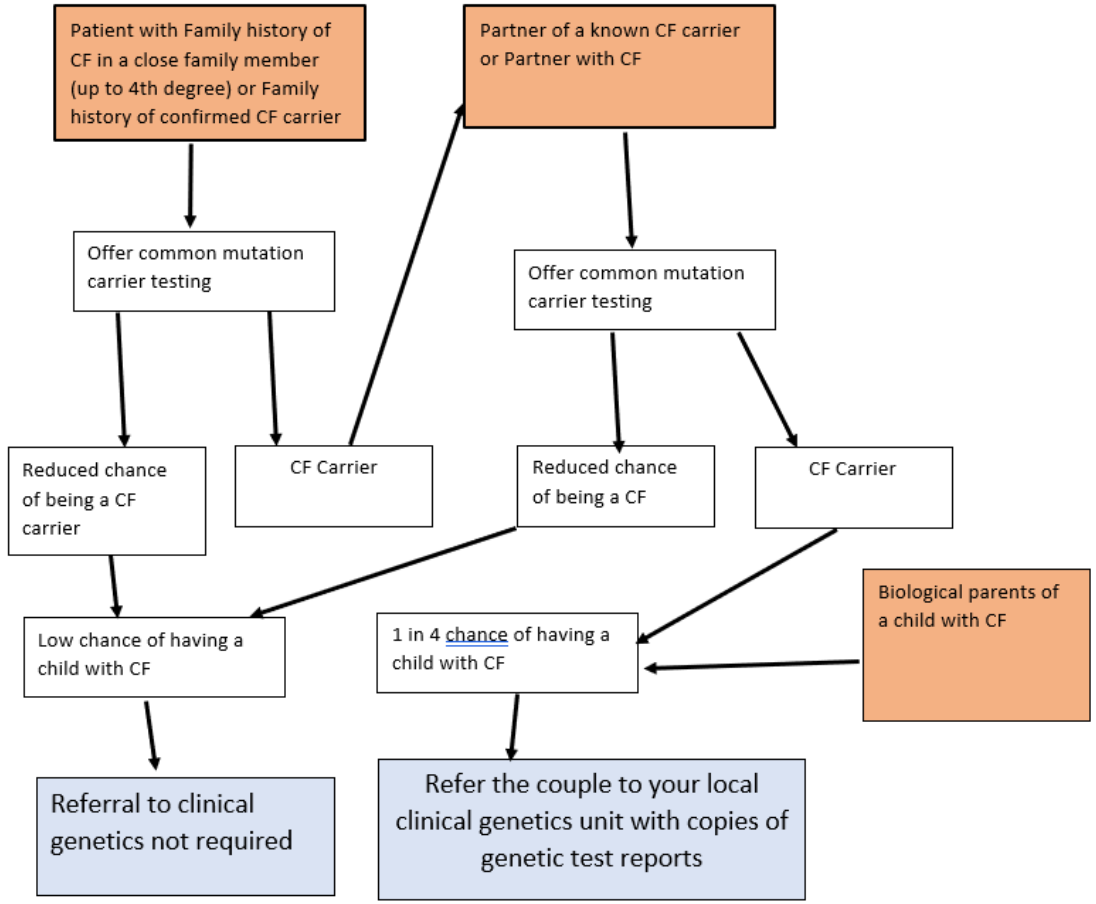
- If both variants are in the same copy of the gene, this patient is still only a CF carrier.
- If the variants are present in each of your patient's two copies of the CFTR gene, this patient may have a mild form of CF.

For an unexpected finding always refer your patient to Clinical Genetics for interpretation of this result. Further testing may be offered if appropriate.

When to refer to Clinical Genetics

See flow chart or:

- 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 variants identified in the CF carrier test.



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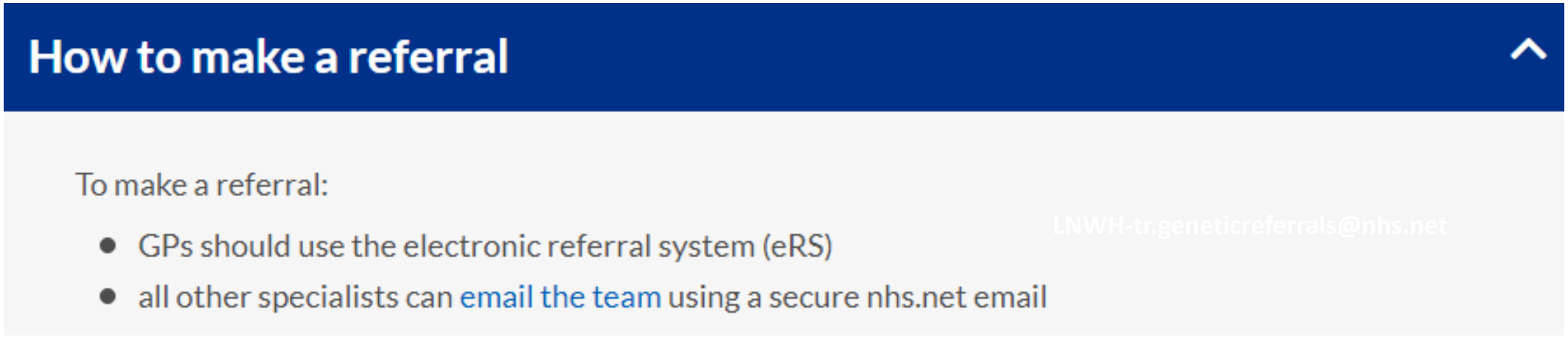
What to include in a Clinical Genetics referral

Include sufficient evidence demonstrating that your patient is appropriate for a Clinical Genetics referral.

This may include:

- Genetic test reports of your patients,
- A description of the reason they were eligible for testing,
- The reason that you are referring.

Regional Genetics Service



A patient's regional Genetics service is determined by GP postcode:

North West Thames	North East Thames
Web: www.lnwh.nhs.uk/genetics **Referrals:** LNWH-tr.geneticreferrals@nhs.net **Clinical Queries:** LNWH-tr.nwtrgsclinicalgenetics@nhs.net	**Web**: https://www.gosh.nhs.uk/wards-and-departments/departments **Referrals:** Through e-RS **Clinical Queries:** gos-tr.clinicalgenetics@nhs.net

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What should patients expect from an appointment in Clinical Genetics?

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

- CF inheritance,
- Reproductive options that will vary based on each couple's individual circumstances and the particular variants that they each carry,
- Encouraging patient to disseminate information to other at-risk relatives, including supporting letters,
- Psychosocial impact of the results.

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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