


Bitesize basics – How to order Cystic Fibrosis carrier test

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Go to our [WEBSITE](#) for up-to-date genetic testing information.

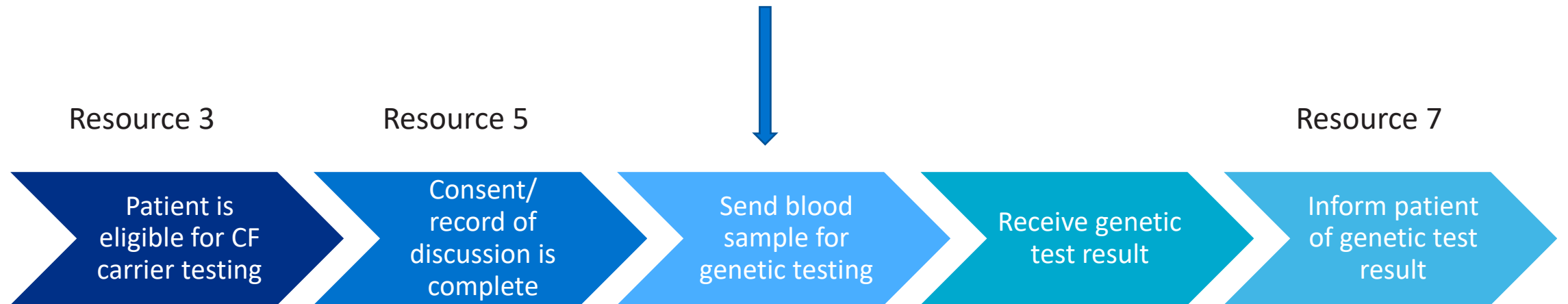




This is one segment of an eight-part bitesize basics learning series for Primary Care, focusing on ordering Cystic fibrosis (CF) carrier testing:

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 pathway





Arranging a CF carrier test

You will need to send both of the following:

1. A 2-5ml blood sample in an EDTA bottle

AND

2. A completed “test request form” (see next slide).

Test request form

A Molecular Genetic Testing request form can be found on the North Thames GMS website:

https://norththamesgenomics.nhs.uk/wp-content/uploads/2023/08/Genetic-Test-Request-Form_v5.pdf

The clinical indication code for CF carrier testing is **R185**.

For guidance on how to fill out this form download our *CF Carrier Testing Toolkit*, available on our website.

The form is titled 'GENETIC TEST REQUEST FORM' and is issued by the North Thames Genomic Laboratory Hub, Rare & Inherited Disease Genomic Laboratory, Level 5 Barclay House, 37 Queen Square, London WC1N 3BH. It includes logos for UKAS, NHS, and the Great Ormond Street Hospital for Children NHS Foundation Trust. A warning states: 'Please note that forms received with missing patient identifiers or no referring clinician/facility may not be tested.'

GENETIC TEST REQUEST FORM

Referring Clinician Details

Lab Ref (lab use only) | Date Received (lab use only)

Referring Clinician: (Full name required)

Contact Number:

NHS.net email: (mandatory)

Department:

Hospital: (Full hosp. name & address required)

Submitter ID (Outreach):

Referring Consultant: (if different from referring clinician)

Referring Consultant Email:

Referring Clinician: I have discussed genomic testing with this patient and have retained a record of discussion (see page 2). Consent is not required for DNA storage.

Patient Details - use four patient identifiers

First name: | Surname:

DOB: | Sex Assigned at birth:

NHS Number: (mandatory) | Hospital No/Your Ref:

Ethnicity: | GOSH Family ID:

Patient Address:

Postcode:

NHS Patient (England) | NHS Patient (Wales, Scotland, N.I.)* | Private/International Patient*

*Billing Address (if organisation to be invoiced): | Purchase Order No.:

*Patient Email Address (if Self Funding):

Specimen Details

Specimen Details	If high risk please specify:	Sample Type	Date / Time Collected	Collected By
High Risk Specimen?	Yes <input type="checkbox"/> No <input type="checkbox"/>			

⁹Clinical Indication Code: **R** | Urgent Routine

Reason for referral: (please give clinical details & details of previous genetic investigations in the family, if known)

⁹ For NHS England referrals, please refer to the National Genomic Test Directory for available tests and eligibility criteria - <https://www.england.nhs.uk/publication/national-genomic-test-directories/>

Molecular Genetic Testing (EDTA, except NIPD, see below)

DNA storage ONLY	Microarray (EDTA only)	Karyotype (Lithium Heparin)
Diagnostic test	If requesting urgent microarray (e.g. pregnancy, infants <3 months) please send a Lithium Heparin as well	To exclude Turner Syndrome (Short Stature/Amenorrhoea ONLY)
Carrier test	Cytogenetic follow up (EDTA & Lithium Heparin)	To exclude Ring 20 (Epilepsy)
Predictive test	Please give the name & GOSH MRN of index patient above or include copy of index patient report	Azoospermia/Male Infertility/IVF
NIPD (PAXgene or Streck cell stabilising tube)	Rapid testing for infants (Lithium Heparin & EDTA)	Premature Ovarian Failure/IVF
Please provide relevant family history above	13/18 <input type="checkbox"/> 21 <input type="checkbox"/> Aneuploidy (please specify)	Sample requested by lab
	Presence of SRY (chromosomal sex)	Chromosome Breakage (not Fragile X) (Lithium Heparin)
		Fanconi Anaemia
		Bloom Syndrome
		Other—contact the lab



Sending a sample to the laboratory

The sample should be sent to the regional genomic laboratory hub, address as below:

North Thames GLH, Rare & Inherited Disease Genomic Laboratory
Specimen Reception
Level 5, Barclay House
37 Queen Square
London
WC1N 3BH

Samples sent by Royal Mail or courier must comply with PI 650 for category B substances.

Samples can be shipped at room temperature.

For more details about sample requirements and sending your sample, please refer to the test request form or contact the North Thames GLH.

Tel: 0207 829 8870 Email: gos-tr.norththamesgenomics@nhs.net



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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