North Thames
Genomic Medicine Service

Bitesize basics – How to order Cystic Fibrosis (CF) carrier test

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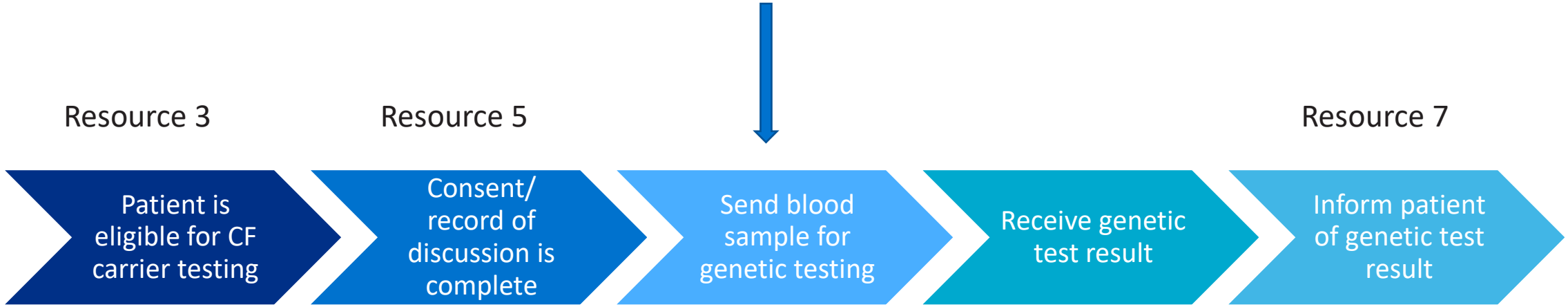
Go to our **WEBSITE** for up-to-date genetic testing information.

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 pathway

Resource 3 | Resource 5 | Resource 7



To do this, you will need:

1. A viable sample
2. A complete test request form

See next slides…

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Sample requirements

There are **TWO** sample requirements to order a CF carrier test:

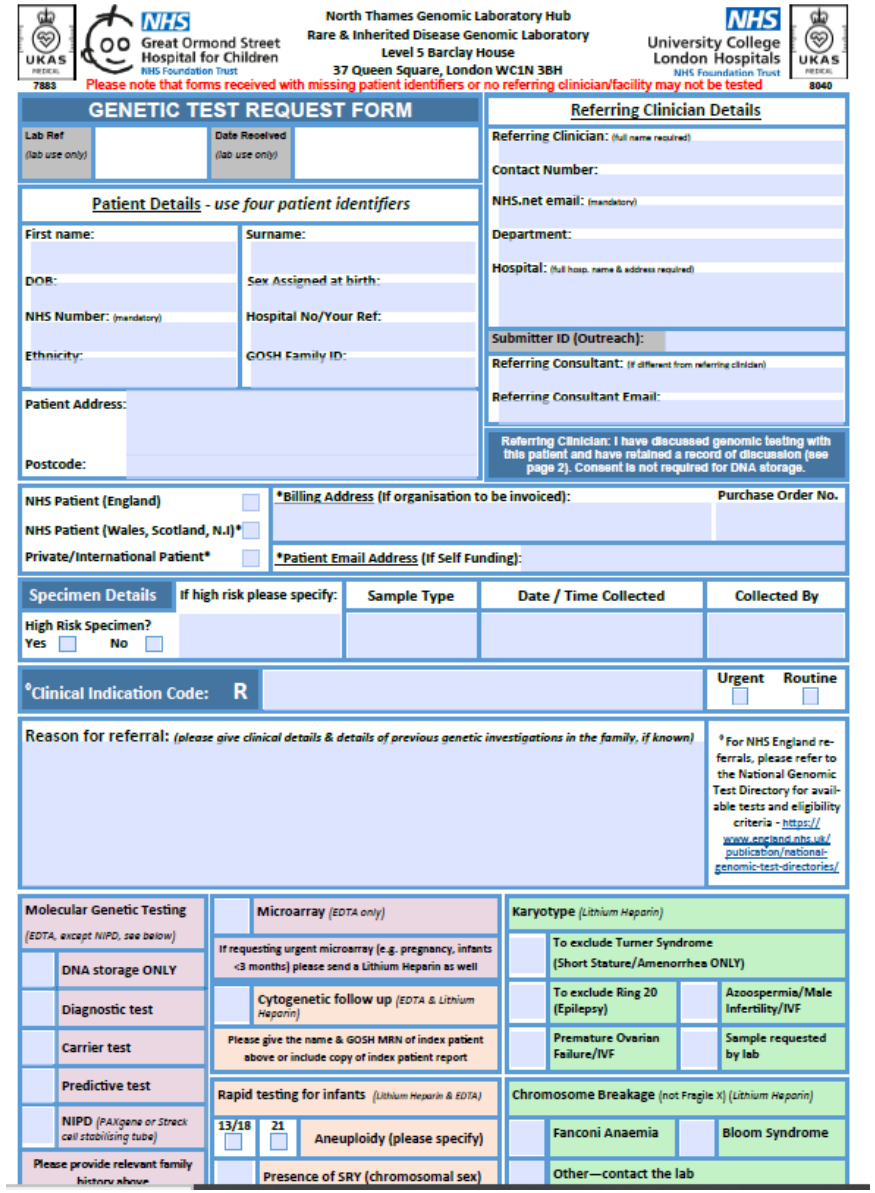
1. A 2-5ml blood sample in an EDTA bottle,
2. An accompanying completed “test request form”.

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

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



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

Great Ormond Street Hospital for Children NHS Foundation Trust
University College London Hospitals NHS Foundation Trust

Please note that forms received with missing patient identifiers or no referring clinician/facility may not be tested

GENETIC TEST REQUEST FORM

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

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:

Referring Clinician Details

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.

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 | If high risk please specify: | Sample Type | Date / Time Collected | Collected By
High Risk Specimen? Yes No

*Clinical Indication Code: R
Urgent Routine

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

*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
Diagnostic test
Carrier test
Predictive test
NIPD (PAXgene or Streck cell stabilising tube)
Please provide relevant family history above

Microarray (EDTA only)
If requesting urgent microarray (e.g. pregnancy, infants <3 months) please send a Lithium Heparin as well
Cytogenetic follow up (EDTA & Lithium Heparin)
Please give the name & GOSH MRN of index patient above or include copy of index patient report

Rapid testing for infants (Lithium Heparin & EDTA)
13/18 21 Aneuploidy (please specify)
Presence of SRY (chromosomal sex)

Karyotype (Lithium Heparin)
To exclude Turner Syndrome (Short Stature/Amenorrhea ONLY)
To exclude Ring 20 (Epilepsy)
Azoospermia/Male Infertility/IVF
Premature Ovarian Failure/IVF
Sample requested by lab

Chromosome Breakage (not Fragile X) (Lithium Heparin)
Fanconi Anaemia
Bloom Syndrome
Other—contact the lab

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Sending a sample

The sample should be sent to the regional genomic laboratory hub – see test request form for postal address.

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

Samples can be shipped at room temperature.

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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