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|   **9865 9322** | **North West Genomic Laboratory Hub (Liverpool)**Manchester Centre for Genomic MedicineLiverpool Women’s Hospital, Crown Street, Liverpool, L8 7SSTelephone: 0151 702 4228Email: dna.liverpool@nhs.netWebsite: https://mft.nhs.uk/nwglh/Scientific Operational Director: Dr. E Howard |  |

## REQUEST FOR GENETIC CYSTIC FIBROSIS (CF) CARRIER TESTING (*CFTR* GENE) – V3

(Form to be used by General Practitioners requesting cystic fibrosis carrier testing)

The North West NHS Genomic Laboratory Hub is able to offer cystic fibrosis (CF) carrier testing direct to general practitioners in either of the following two scenarios:

* Individuals with a family history of CF and therefore at an increased risk of being a carrier of CF (i.e. they have a family member who is affected with, or is a carrier of, CF). Please note we cannot accept carrier testing requests for individuals under 16 years of age, as they cannot give informed consent.
* Individuals with a partner who is either affected with, or is a confirmed carrier of CF, and as a couple they wish to know their risk of having a child with CF.

If your patient has clinical symptoms suggestive of CF or a *CFTR*-related disorder, please refer them to the appropriate clinical speciality.

Please note that funding for this service is currently met by contracting arrangements established prior to the Health and Social Care Act 2012. Therefore, the costs are covered by the former Greater Manchester, Cumbria, Lancashire, Cheshire & Merseyside commissioning arrangements. For referrals outside this geographical area, please contact the laboratory for billing arrangements.

**GUIDANCE NOTES**

**Please complete sections 1 – 4 for all referrals and either section 5 or 6**

**Sample Information:**

* Sample type required: 4ml venous blood sample in an EDTA tube. Store sample at 4˚C if required.
* Samples in Lithium Heparin or Serum tubes are unsuitable for testing.
* In accordance with the Health & Safety at Work Act and the COSHH Regulations, the laboratory must be informed of any infection risk associated with submitted samples. The sender has the responsibility for minimising the risk to laboratory staff by giving sufficient information to enable the laboratory to take appropriate safety precautions when testing a specimen. If the sample is high risk, please state the nature of the risk on the referral form.
* The sample container should be sealed in a biohazard bag in case of a leakage. To prevent contamination of referral form and paperwork this should not be sealed with the sample. All packaging should conform to UN650 standards (as applied to UN3373 – Biological Samples, Category B).

**FORWARD THE COMPLETED REFERRAL FORM AND EDTA BLOOD SAMPLE TO SAMPLE RECEPTION AT THE LIVERPOOL LABORATORY SITE (full address above).**

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| **1. PATIENT DETAILS *(affix a printed label if available)*** |  | **2. REFERRING CLINICIAN** |
| Surname: |  | GP Name: |
| Forename: |  | Surgery Address: |
| DoB: | NHS No: |  |  |
| Sex: | Hospital No: |  | Telephone No: |
| Address: |  | Email: |
|  |  | Consent Statement.It is the referring clinician’s responsibility to ensure the patient/carer knows the purpose of the test and that DNA may be stored |
| Postcode: |  | GP Signature: |  |

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| **3. PATIENT’S ETHNICITY/COUNTRY OF ORIGIN:** |  |  |
| This information is important as the detection rate of the test varies between different ethnicities and populations, and it critical for calculating residual carrier risks. Please be specific, i.e. white British rather than Caucasian, Polish rather than Eastern European. |

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| **4. SAMPLE INFORMATION:** | Date Taken | High Infection Risk:(See guidance notes) |  |
| 4ml venous blood sample in an **EDTA tube.** Store sample at 4OC if required. Lithium Heparin or Serum tubes are unsuitable for testing. |

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| **5.** **PATIENT HAS A FAMILY HISTORY OF CF**  |
| There are over 2000 different pathogenic variants / mutations in the *CFTR* gene. Our standard panel covers 94% of those for patients of white British origin. Providing information on the family member and their pathogenic variant / mutation will allow us to initiate the most relevant test. |
| Name of relative(s): |  |
| DoB of relative(s): |  |
| Status of relative (affected or carrier): |  |
| Relationship to the patient (i.e. mother, paternal uncle etc.), please draw a pedigree if appropriate |
| Details of the familial *CFTR* pathogenic variant (if known):(If the relative was tested in the North West NHS Genomic Laboratory Hub, we may be able to access this information) |

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| **6. PARTNER OF A** **PATIENT WHO IS AFFECTED WITH, OR A CARRIER OF, CF** |
| Providing details of the patient’s partner will allow us to provide a pregnancy risk specific to this couple. |
| Name of partner: |  |
| DoB of partner: |  |
| Status of relative (affected or carrier): |  |
| Details of the partner’s *CFTR* variant (if known):(If the relative was tested in the North West NHS Genomic Laboratory Hub, we may be able to access this information) |