**Scientific Operational Director: Dr E. Howard**

**Website:** <https://mft.nhs.uk/nwglh/>

**Referring Samples for Rare and Inherited Disease Investigations**

Dear Colleague,

Thank you for referring a sample for genetic testing to the North-West Genomic Laboratory Hub (NWGLH).

The full repertoire of rare disease tests on the NHSE Genomic Test Directory for Rare disease has now been launched in England. This Directory sets out the repertoire of tests available through the National Genomic Medicine Service. Funding for these tests will be met centrally however patients will need to meet defined eligibility criteria to access testing. A full listing of the available tests as well as eligibility criteria for rare and inherited disease is available at:

<https://www.england.nhs.uk/publication/national-genomic-test-directories/>

If you are a clinician based in the North West of England you can access this testing through your local laboratory hub. For Rare and inherited disease this hub is co-located on two sites for ease of access:

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| --- | --- |
| **Manchester Centre for Genomic Medicine**  6th Floor, St Mary’s Hospital,  Manchester  M13 9WL  Tel +44(0) 161 276 6122  Email: [mft.genomics@nhs.net](mailto:mft.genomics@nhs.net) | **Liverpool Centre for Genomic Medicine,**  Liverpool Women's NHS foundation trust,  Crown street,  Liverpool L8 7SS  Tel +44(0) 151 702 4228  Email: [dna.liverpool@nhs.net](mailto:dna.liverpool@nhs.net) |

In the North West of England requests for all rare and inherited test indications must be sent to the one of the two NWGLH laboratories. Samples will be processed and may be forwarded to a different specialist laboratory in the GLH network for testing. Referrals will be triaged and only tests that meet the approved criteria will be processed. When sending a sample please identify the disease indication code (available from the Directory above – ie “R” number, eg R125). Referral forms and specialist service proformas are available at our website <https://mft.nhs.uk/nwglh/>. Please submit Lithium Heparin samples for karyotype analysis. For all other genetic investigations EDTA samples are required.

**IMPORTANT**: From now on only tests listed on the NHSE England Directory will be available. Some historic tests have been removed and are no longer available. We strongly recommend that you check for test availability on the Directory before sending a sample. In particular please note that a number of the targeted tests previously available have been replaced by multigene panel tests. It is important to understand the nature of these new tests in order that informed consent can be obtained from your patient.

For clinicians not based in the North West of England you will access the same portfolio of tests via your local GLH. We can provide additional contact details on request.