



NORTH WEST GENOMIC LABORATORY HUB

General Genomic Testing – Helpful Resources

There is lots of useful information on our website: <https://mft.nhs.uk/nwglh/>
Contact us: mft.genomics@nhs.net

Genomic testing is delivered through a single national network of Genomic Laboratory Hubs (GLHs) across England, each is responsible for coordinating testing within their geographical region. The North West Genomics Laboratory Hub (NWGLH) is led by Manchester University NHS Foundation Trust with laboratory sites at Manchester and Liverpool and includes other delivery partners across the region.

We provide core rare disease and cancer testing for the North West region, including whole genome sequencing (WGS), and specialist tests for neurology, hearing loss, cardiac, metabolic, immunology, haematology, inherited cancer and eye disorders as a designated national testing provider. For other specialist tests, samples may be forwarded onto other GLHs, but this will not affect the referral process or the return of results.

Finding the right test

The **National Test Directories** for rare diseases and cancer have set out the clinical conditions or indications where there is clear evidence for the value of genomic testing for patients. The test directories are updated each year to ensure access to the latest testing.

A supplementary **eligibility criteria** document describes which patients should be considered for rare disease testing under each indication, and the clinical specialties who can request the test.

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

It is important that **clinical utility** is also considered.

Test Directory frequently asked questions guide:

<https://www.england.nhs.uk/publication/national-genomic-test-directory-supporting-material/>

For Whole Genome Sequencing requests consider submitting family trio samples where possible.

The **NHS Genomic Medicine Service Panels Resource** provides a search facility to view the gene content of panels and search for individual genes:

<https://nhsgms-panelapp.genomicsengland.co.uk/>

Informed Consent

Informed consent should be taken for a genetic test. Where the person lacks capacity, best interests may need to be considered.

Suggested standard consent discussion:

- DNA change may be identified related to an individual's health concerns, and results may have implications for other family members
- No genetic changes found - cause could still be genetic, but no causal changes identified on test
- Finding of uncertain significance - potential effect cannot be confirmed with current knowledge
- Unexpected findings - sometimes unexpected information is found that may have health implications for the patient or family members
- Sample storage - a small amount of DNA or cells are used for the test. The rest is stored and may be used for family testing or quality control purposes.

Consent should be recorded in the medical records. A standard form may be used:

<https://mft.nhs.uk/nwglh/documents/consent/>

For WGS, a record of discussion must be completed and a copy submitted with the sample for testing:

<https://mft.nhs.uk/app/uploads/2021/05/1-The-Record-of-Discussion-form-for-WGS.pdf>

Test ordering

- Find the form at the NW GLH website: <https://mft.nhs.uk/nwglh/documents/test-request-forms/>
- **Please include full clinical details, testing eligibility & clinical utility, R or M code and contact details of requestor - testing may be delayed if documentation is incomplete**
- Referrals can be made electronically from some linked Trusts
- Collect the correct sample type – refer to guidance notes on specific referral forms or our website
- Please indicate any infection risk associated with submitted samples.
- Send the labelled sample and completed form to the NWGLH via the usual laboratory transport systems
- **Unlabelled samples cannot be accepted**

Educational Resources



GeNotes

Genomic notes for clinicians; educational resource for healthcare professionals, designed to be used in a clinical setting

<https://www.genomicseducation.hee.nhs.uk/about-us/genotes-genomic-notes-for-clinicians/>



GeneReviews

International point-of-care resource that provides clinically relevant and medically actionable information for inherited conditions

<https://www.ncbi.nlm.nih.gov/books/NBK1116/>



NHS
North West

Genomic Medicine Service Alliance

NW GMSA is one in a network of regional alliances launched in England by the NHS Genomic Medicine Service (GMS) to oversee and co-ordinate the embedding of genomics into routine healthcare across England.

<https://www.nw-gmsa.nhs.uk>



Genomics
England

Online resource to learn about genomics

<https://www.genomicsengland.co.uk/genomic-medicine/understanding-genomics>