Genomics

NHS Genomic Medicine Service

The role of NHS England is to enable the NHS to harness the power of genomic technology and science to improve the health of our population and deliver on the commitments in the NHS Long Term Plan. These commitments will be delivered on by the NHS Genomic Medicine Service for England.

Workforce development

To help achieve this NHSE are delivering three workforce development programmes covering nursing and midwifery, pharmacy and medical practitioners, with the support of Health Education England's Genomics Education Programme.

Genomics Clinical Reference Group

The Genomics Clinical Reference Group (CRG) has been convened to support implementation of the NHS Genomic Medicine Service (GMS). This group is made up of professional, patient, and public representation. They will:

- Advise on clinical policy and strategy for genomics, including implementation of NHS Long Term Plan commitments and future development of the NHS GMS;
- Oversee a clear and transparent process for annual review of the National Genomic Test Directory (supported by three test evaluation working groups covering rare and inherited disease, cancer and pharmacogenomics, see below).

National Genomic Test Directory

The National Genomic Test Directory specifies which genomic tests are commissioned by the NHS in England, the technology by which they are available, and the patients who will be eligible to access to a test.

The following specific tests are available to dermatology:

Genomic testing with multiple genes

For genomic testing where multiple genes are tested in parallel, NHS England have deployed the PanelApp tool to ensure that panels have an agreed set of curated genes and that this content is supported by scientific evidence. Using this tool, you can search using the national test directory code or the name of the panel (e.g. Primary immunodeficiency).

Testing Location

As the 7 Genomic Laboratory Hubs will operate as a national network, the laboratory that will carry out a specific test may be different than the current arrangements. However, in most cases, the laboratory that you send your samples to will not change. If testing is carried out elsewhere then your local GLH will forward the sample on.

Tests site:

- Central and South Genomic Laboratory Hub led by Birmingham Women's and Children NHS Foundation Trust
- East Genomic Laboratory Hub led by Cambridge University Hospitals NHS Foundation Trust
- North West Genomic Laboratory Hub led by Manchester University NHS Foundation Trust
- North Thames Genomic Laboratory Hub led by Great Ormond Street Hospital for Children NHS Foundation Trust
- South East Genomic Laboratory Hub led by Guy's and St Thomas' NHS Foundation Trust
- South West Genomic Laboratory Hub led by North Bristol NHS Trust
- North East and Yorkshire Genomic Laboratory Hub led by The Newcastle upon Tyne Hospitals NHS Foundation Trust

For a comprehensive list of WGS indications (with prospective activation start dates under active review) please see:
Rare Disease WGS Clinical Indications

Please note that eligibility criteria apply to WGS testing, these are available at https://www.england.nhs.uk/publication/national-genomic-test-directories/. For some clinical indications the WGS test is dependent on carrying out a prior non-WGS genomic investigation.

Patient Choice Consent Framework

Referring clinicians should be familiar with and have undertaken induction on the Patient Choice Consent Framework prior to ordering a WGS test.

WGS Rare Disease – Documents

New Test Order and Record of Discussion forms are available that can be completed electronically or printed and completed by hand. Download these forms onto your computer and open in Adobe software to enable electronic editing.
Test Order Form – Complete ONE form per family

Record of Discussion Form – Complete ONE form per individual
⇒ WGS Record of Discussion (RoD) Form

The patient choice conversation can be performed remotely, and the form submitted without a patient signature – please tick the "Remote consent" check box in the Healthcare Professional use only section on p3 of the form to indicate this.

Send completed Test Order Form and Record of Discussion Forms to mft.nwglhdnalab@nhs.net
Include "WGS Rare Disease" in the subject heading.
Samples for WGS and their Transport

Samples for DNA extraction for WGS should be sent to the Manchester site laboratory accompanied by the NW GLH WGS Test Request Form (Rare Disease Referral form).
Blood samples should be sent in EDTA.

If DNA is already stored at the Manchester or Liverpool site laboratory for an individual, please indicate this and the location on the test request form. If DNA is stored at another GLH please request for an aliquot of DNA to be transferred. Please indicate that the DNA is required for WGS (a minimum 2ug of DNA is required for WGS).

Patient Information
Patient Information for Rare Disease
Patient Information for Rare Disease – Easy Read
Patient Information for WGS Research

Questions?
We have prepared an information sheet covering Frequently Asked Questions (FAQs) this can be viewed from the link below.
Rare Disease WGS – FAQs

Education and training Resources

NWGLH provides a comprehensive Genomics catalogue of useful resources which contributes to the education and training of staff members working towards professional registration and/or continuous professional development (CPD)

https://mft.nhs.uk/nwglh/nwglh-education-and-training-resources/