NHS
Central & South
Genomic Laboratory Hub

# Genomic Medicine Service

## Background

An **NHS Genomic Medicine Service (GMS)** is being established in England with the aim of providing consistent and equitable access to genomic testing.

All existing genetic testing in England was incorporated into the GMS. The GMS will provide genetic testing for patients with rare and inherited disease and for patients with cancer.
The GMS is being delivered through a national testing network, consolidating and building on existing laboratory services. Regional Genetics Laboratories in England have been grouped into seven **Genomic Laboratory Hubs** (GLHs) each responsible for coordinating services for a particular part of the country.

The **Central and South GLH** includes:

- The Genomics Laboratory Hub:
    - West Midlands Regional Genetics Laboratory at Birmingham Women's and Children's Hospital NHS FT
- Local Genomics Laboratories:
    - Wessex Regional Genetics Laboratory at Salisbury NHS FT
    - Wessex Molecular Pathology Laboratory at University Hospitals Southampton NHS FT
    - Oxford Genetics Laboratories at Oxford University Hospitals NHS FT
    - Molecular Pathology Diagnostic Service at University Hospitals Birmingham NHS FT

## The National Genomic Test Directory (TD)

A central feature of the GMS is the **National Genomic Test Directory** (TD), which lists the types of genomic tests – including karyotyping, microarray testing, tests for single genes and molecular markers, gene panel tests, whole exome sequencing (WES) and whole genome sequencing (WGS) – that will be available through the NHS in England.

The TD **specifies which genomic tests are** funded by NHSE as part of the GMS. It also details the technology by which they are delivered. An **eligibility criteria** document supplements the TD, setting out clinical indications for each test and the clinical specialties who are expected to request a given test. (The clinical eligibility criteria for cancer referrals are still in development.) All tests that are listed within the TD are funded via the NHSE Specialist Commissioning provision for England**.**

**Tests and indications not specified within the Test Directory will not be centrally funded.**

The TD can be found here:
https://www.england.nhs.uk/publication/national-genomic-test-directories/

There is a submission and approval process for the addition of new indications and tests to be included in the Test Directory that is managed by NHSEI via the Genomics Clinical Reference Group (CRG). This will ensure that the tests and technologies available are supported by the most up to-date scientific, clinical and health economic evidence.

NHS
Central & South
Genomic Laboratory Hub

The GLHs are currently transitioning to full implementation of the Test Directory. This will be completed by **April 2021** for rare disease with some further work required during 2021/22 for cancer tests. The 7 GLH laboratories will operate as a network to provide the full TD to all eligible patients.

**What this means for you**

- The GMS will provide a nationally funded laboratory service for "rare disease" and cancer patients in England, ensuring equitable access for clinicians and patients across England.
- **It will be important for all service users to become familiar with the Test Directory. The TD uniquely identifies rare disease tests by "R numbers". Cancer test indications are uniquely identified by "M numbers". Proper use of these codes will ensure that the GLH laboratories can accurately undertake the test requested by the clinical team.** It is essential that clinicians provide comprehensive demographic and clinical information to avoid unnecessary processing delays.
- The majority of previously available genetic tests are still found in the TD, however many single gene tests are no longer available. If you cannot find the test you require please contact the laboratory via the emails provided below, providing the name/details of the test you requested previously so that we can help you to identify the most appropriate 'R' or "M" code.
- An interactive web based version of the Test Directory is under development and we will inform you of the launch date as soon as this is available.
- Wherever possible, service users should use **their Local Genomic Referral Form** to order non-whole genome sequencing tests from the TD. These can be located on each laboratory's website (listed below). A single GLH wide referral form is currently being designed.
- Clinical teams from your local clinical genetics service will have been organising and running training and education sessions for clinicians who need to engage with the GMS. If you feel that further sessions are required in your area or for your clinical team you should let us know via the contact details below.
- A small number of tests will now be provided by **Whole Genome Sequencing (WGS)** (such as R29 Intellectual disability and R27 Congenital malformation and dysmorphism syndromes). WGS requires specific Test Order Forms and a completed "Record of Discussion" form. If you require access to these please contact your local partner laboratory. Again, you should also have had access to WGS request training sessions run by members of your local Genomic Medicine clinical team.

Clinicians in your local clinical genetics unit can also be contacted if you require help with the new GMS. The main email contact for the GLH is via **bwc.centralsouthglh@nhs.net**

**Contact details for all GLH consortium Laboratories**

| | | |
|---|---|---|
| West Midlands Regional Genetics Laboratory | website | bwc.centralsouthglh@nhs.net |
| Wessex Regional Genetics Laboratory | website | shc-tr.WRGLdutyscientist@nhs.net |
| Wessex Molecular Pathology Laboratory | website | uhs.molecularpathology@nhs.net |
| Oxford Genetics Laboratories | website | orh-tr.dutyscientist.oxfordgen@nhs.net<br>oxford.molecularhaem@nhs.net |
| Molecular Pathology Diagnostic Service | website | MPDS.Enquiries@nhs.net |