The genomics facilitator's toolkit

NTGLH Module ID	Module Title	Module Format
NTGHL_001	NHS Genomic Medicine Service	Handbook
NTGHL_002	Ordering from the National Test Directory	Handbook
NTGHL_003	Whole Genome Sequencing consent	Handbook
NTGHL_004	Whole Genome Sequencing sample requirements	Handbook
NTGHL_005	Clinical genetic testing methods	Powerpoint with narration
NTGHL_006	Clinical testing DNA sequence variant interpretation	Powerpoint with narration
NTGHL_007	Whole Genome Sequencing results	Powerpoint with narration
NTGHL_008	Introduction to genomics	Powerpoint with narration
NTGHL_009	Test cases in cancer	Handbook

NHS
North Thames
Genomic Laboratory Hub

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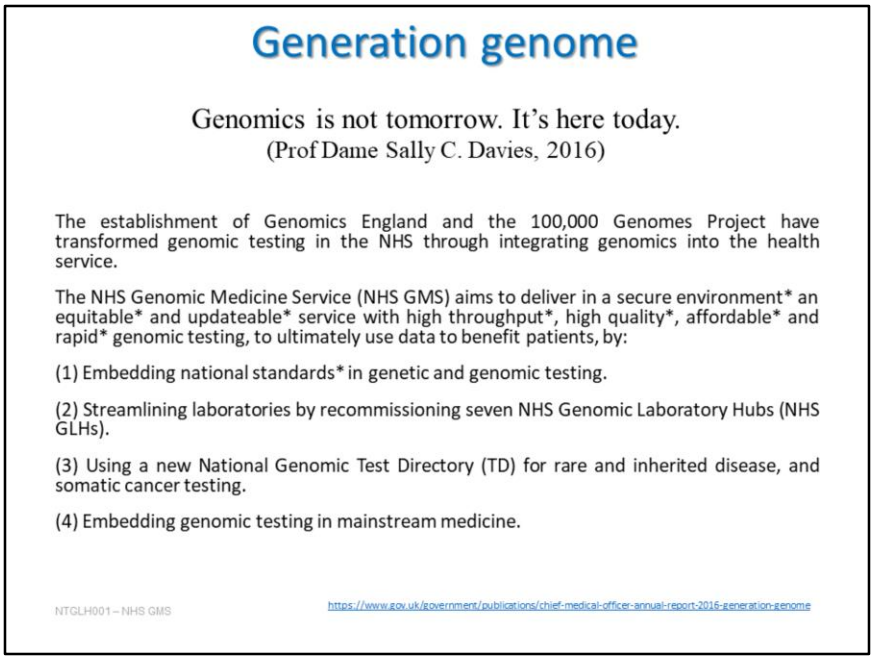
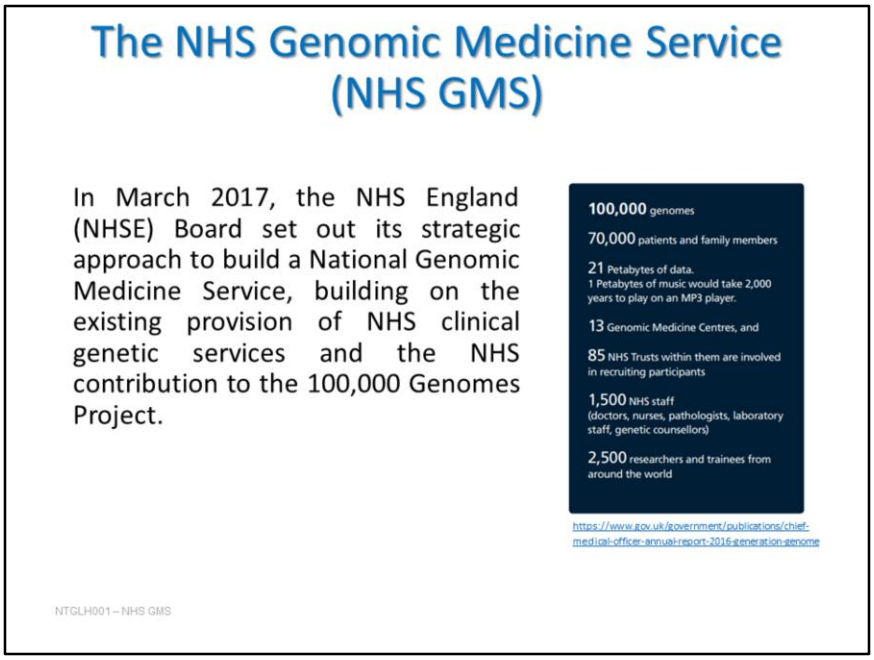
Contents



In 2016 Prof Dame Sally C. Davies wrote the Annual report of the Chief Medical Officer: generation genome. This report looked at how genomics can improve health and prevent ill-health. The report made recommendations to address gaps in infrastructure, public engagement, organisation of research and provision of services, to widen access to genomic services. See, https://www.gov.uk/government/publications/chief-medical-officer-annual-report-2016-generation-genome

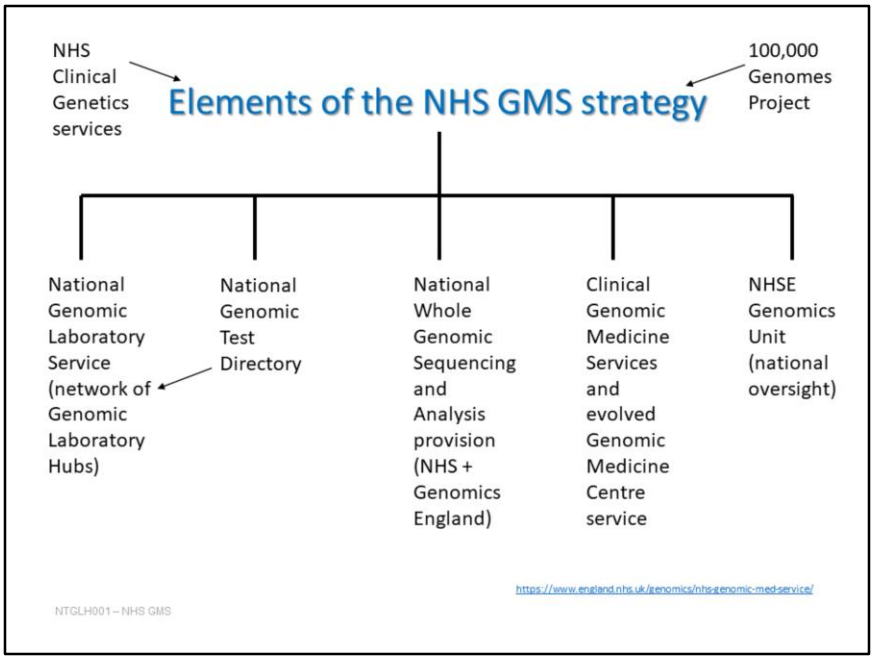
The National Genomic Test Directory can be accessed from, https://www.england.nhs.uk/publication/national-genomic-test-directories/

*All attributes are made possible by moving away from cottage industry-delivered testing, to the creation of Genomic Laboratory Hubs, see https://www.england.nhs.uk/genomics/genomic-laboratory-hubs/



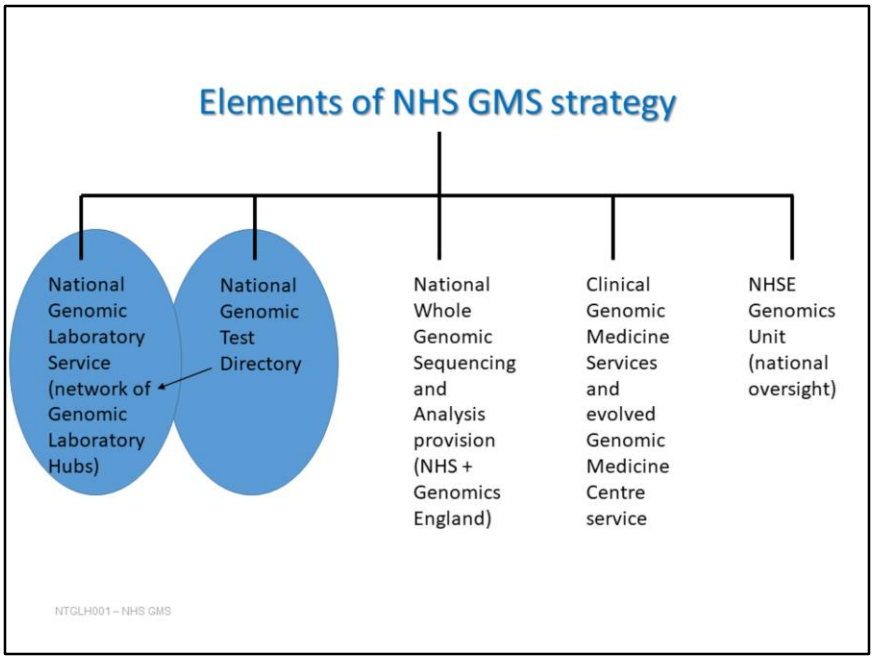
The 100,000 Genomes Project launched in 2012, https://www.england.nhs.uk/genomics/100000-genomes-project/. For more information on genomic medicine and an overview of the 100,000 Genomes Project see, http://researchbriefings.files.parliament.uk/documents/POST-PN-0504/POST-PN-0504.pdf for a downloadable POSTnote.

Through the establishment of Genomics England and the 100,000 Genomes Project, infrastructure – reengineering of the clinical pipeline, development of standards and databases alongside a platform for bioinformatics and genome interpretation – was laid to integrate genomics into the NHS.



The NHS Genomic Medicine Service – https://www.england.nhs.uk/genomics/nhs-genomic-med-service/ – is built on the provision of NHS Clinical Genetics services and the NHS contribution to the 100,000 Genomes Project. The NHS Genomic Medicine Service comprises of five key elements:

1. A national Genomic Laboratory Service through a network of Genomic Laboratory Hubs
2. A new National Genomic Test Directory to underpin the genomic laboratory network
3. A national Whole Genomic Sequencing provision and supporting informatics infrastructure developed in partnership with Genomics England
4. Clinical genomic medicine services and an evolved Genomic Medicine Centre service, https://www.genomicsengland.co.uk/about-genomics-england/the-100000-genomes-project/genomic-medicine-centres
5. A national co-ordinating and oversight function within NHSE; formation of the NHSE Genomics Unit.



The remainder of this module concentrates on the first two key elements of the national NHS GMS:

1. A national Genomic Laboratory Service through a network of Genomic Laboratory Hubs
2. A new National Genomic Test Directory to underpin the genomic laboratory network.

Genomic Laboratory Hubs (GLHs)

- Seven GLHs are users of the NHS GMS systems
- They operate to common national standards, guidelines and protocols
- They deliver genetic and genomic testing from a single national testing directory - national equity
- Testing will be split:

(1) Core - all GLHs

(2) Specialist - GLHs who are National Specialist Test Providers.

NTGLH001 – NHS GMS

https://www.england.nhs.uk/genomics/genomic-laboratory-hubs/

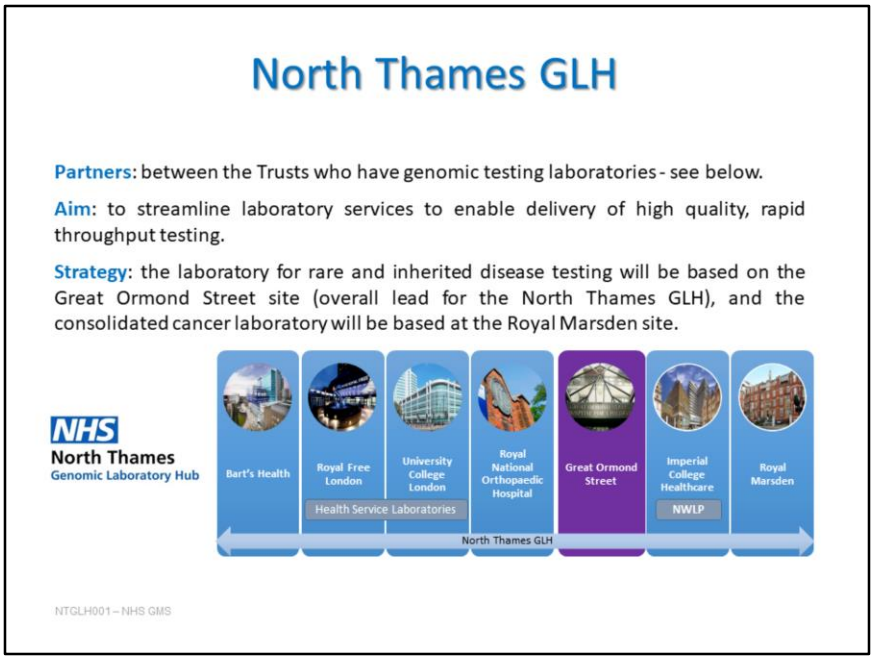
For the NHS GMS to achieve its aim to deliver an equitable, updateable, high throughput, high quality, affordable, secure and rapid 'genomic' testing service, national standards, centralised genomic laboratories and related services were recommended.

NHSE commissioned genomic testing in the NHS to be provided through a single national testing network. The network involved the streamlining of laboratories through consolidation into seven NHS Genomic Laboratory Hubs (GHLs), each responsible for coordinating and delivering services for a particular part of the country. The seven GLHs are:

1. West Midlands, Oxford and Wessex GLH
2. East Midlands and East of England GLH led by Cambridge University Hospitals NHS Foundation Trust
3. North West GLH led by Manchester University NHS Foundation Trust
4. London North GLH led by Great Ormond Street Hospital for Children NHS Foundation Trust
5. London South GLH led by Guy's and St Thomas' NHS Foundation Trust

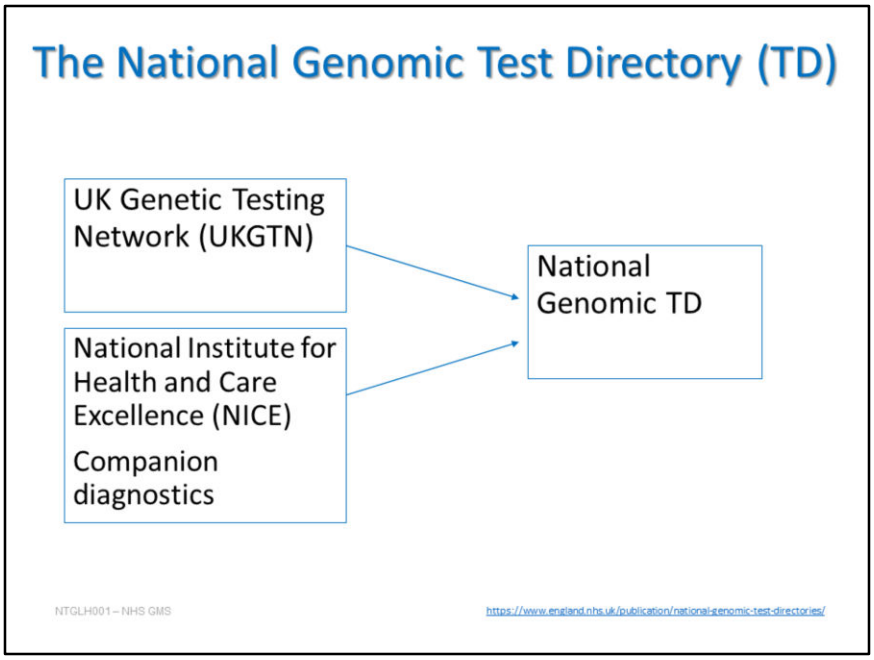
6. South West GLH led by North Bristol NHS Trust
7. Yorkshire and North East GLH led by The Newcastle upon Tyne Hospitals NHS Foundation Trust

For more information see, https://www.england.nhs.uk/genomics/genomic-laboratory-hubs/



For news from the North Thames GLH, follow us on Twitter @NorthThamesGLH (https://twitter.com/norththamesglh?lang=en) and contact us at, gos-tr.norththamesglh@nhs.net.

Health Service Laboratories (HSL) is the laboratory service provider for Royal Free and UCL, https://www.hslpathology.com/ and the North West London Pathology (NWLP) is the laboratory service provider for Imperial College, http://pathology.imperial.nhs.uk/



From 2001 to 2018 the UK Genetic Testing Network (UKGTN) was a national advisory organisation for NHS clinical genetic testing services. The UKGTN evaluated and recommended genetic tests for rare and inherited disorders for the NHS across the UK, by publishing the NHS Directory of Genetic Disorders/Genes for Diagnostic Testing. However, the UKGTN did not have an equivalent for cancer genomic testing. Instead this testing offered within the NHS had evolved over time in-part driven by NICE medicine assessments and the identification of companion diagnostics. From October 2018 the National Genomic Test Directory (TD) replaced the UKGTN, specifying the genomic tests that are commissioned by the NHS in England. Over time, as the evidence develops, the TD will also include other functional genomic tests for example RNA based technologies and proteomics.

The 2020/2021 National Genomic TD has been released, which specifies the genomic tests that 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. See, https://www.england.nhs.uk/publication/national-genomic-test-directories/. For more information on the TD see, https://www.england.nhs.uk/wp-content/uploads/2018/08/national-genomic-test-directory-faqs.pdf

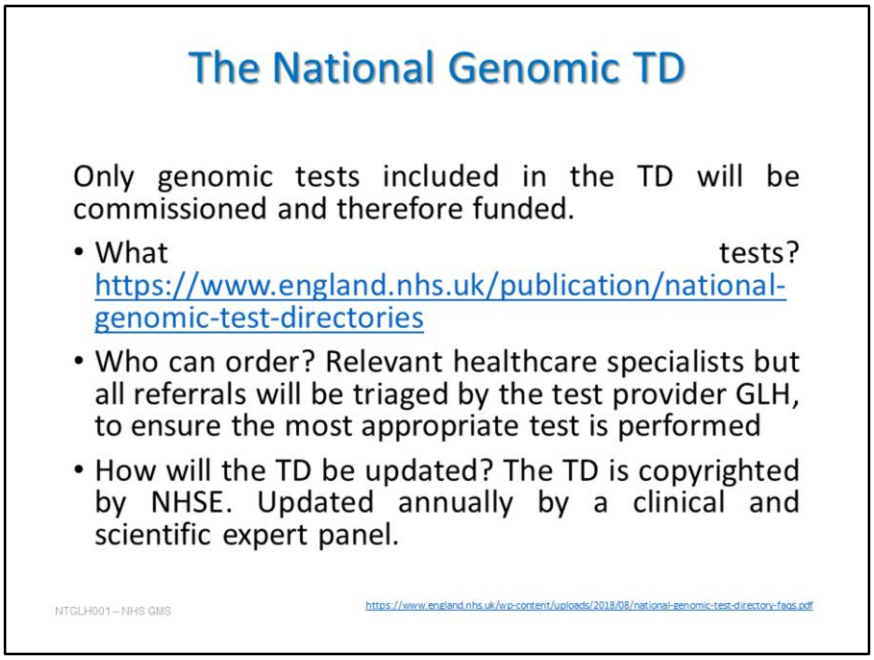
The National Genomic TD

The TD will contain:

- Tests available in the NHS in England; one TD for rare and inherited disorders and one TD for cancer
- Technology platform by which the testing will be delivered
- Clinical eligibility to access a test
- Funding arrangements for the test.

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

For more information see module, NTGLH002_Ordering from the Test Directory



As many of the clinical tests are only relevant for certain specialties e.g., clinicians from Clinical Genetics, Obstetrics, Paediatrics etc, the TD indicates the specialties who will be able to order a specific genomic test. There may, however, be exceptional circumstances where clinicians need to order a test outside of the standard repertoire. In such circumstances, please contact the local test provider laboratory.

The TD will be updated on an annual basis, and NHSE working with colleagues from the Devolved Nations and supported by a Clinical and Scientific Expert Panel, will:

(1) Implement a clear and transparent process for the future evaluation of new genomic tests.
(2) Determine which tests are available within the NHS and any tests that will be retired or replaced by more modern technology, such as whole genome sequencing.
(3) Publish the UK approach to evaluating genomic tests
(4) Publish a policy outlining the approach to commissioning and funding the tests.

The National Genomic TD

Funding:

- The TD will clearly indicate the commissioner responsible for funding each genomic test
- Testing eligible patient's in cancer, and rare and inherited disorders will be funded by NHSE
- Whole genome sequencing will be funded by NHSE
- Cancer genomic testing is included under the National Tariff Payment System and is included within the HRG tariff payment for an individual patient

https://www.england.nhs.uk/wp-content/uploads/2018/08/national-genomic-test-directory-faqs.pdf

The National Genomic TD

How to order from the TD and how to consent for genomic tests are described in separate modules:

- NTGHL_002 Ordering from the Test Directory
- NTGHL_003 Whole Genome Sequencing consent.

Advice and educational resources

North Thames Genomic Laboratory Hub

- Follow us: @NorthThamesGLH
- Contact us at: gos-tr.norththamesglh@nhs.net

Further education

- https://www.genomicseducation.hee.nhs.uk/
  Free online course -5 weeks, 2 hours per week
- https://www.genome.gov/about-genomics/fact-sheets
- https://www.futurelearn.com/courses/the-genomics-era
- https://geneticsunzipped.com/blog/2019/3/4/008-getting-ready-for-genomic-medicine

Contacts and information

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Includes slides adapted from:
https://www.genomicseducation.hee.nhs.uk/supporting-the-nhs-genomic-medicine-service/

https://www.england.nhs.uk/wp-content/uploads/2018/08/national-genomic-test-directory-faqs.pdf

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