National Genomic Test Directory: Frequently Asked Questions

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1 Introduction

To enable the NHS to benefit from advances in genomics, NHS England and NHS Improvement launched the NHS Genomic Medicine Service (GMS) in 2018.

The NHS GMS aims to provide consistent and equitable access to cutting-edge genomic testing to England's 55 million population through consolidating existing services and improving access to the best of current NHS practice, while providing the foundation to deliver future technologies and approaches as they arise.

The NHS GMS consists of the following elements:

- **A national genomic laboratory service** delivered through a network of seven NHS Genomic Laboratory Hubs (GLHs).
- **A National Genomic Test Directory** defining the testing available within the NHS in England;
- **National Whole Genomic Sequencing provision** and a supporting informatics infrastructure developed in partnership with Genomics England;
- **An integrated clinical genetics service** inclusive of genomic counselling for rare and inherited diseases and cancer;
- **NHS Genomic Medicine Service Alliances,** to support the systematic embedding of genomic medicine in end to end patient pathways to ensure all eligible patients can access high-quality services; and
- **A national implementation, co-ordination and oversight function** within NHS England and NHS Improvement (Genomics Unit).

More information on the NHS GMS can be found on the NHS Genomics website: https://www.england.nhs.uk/genomics/

2 About the National Genomic Test Directory

The National Genomic Test Directory (Test Directory) was first published in October 2018 and 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 a test. Over time, as the evidence develops, the scope of the Test Directory is likely to expand to cover other areas, such as pharmacogenomics (how patients' genomic variation affects how they respond to the medicines they are given), and other functional genomic tests for example RNA based technologies and proteomics.

3 Updating the National Genomic Test Directory

The Test Directory will be reviewed and updated on an annual basis following the evaluation and review process implemented by NHS England and NHS Improvement (supported by the Genomics Clinical Reference Group (CRG) and clinical and scientific expert Test Evaluation Working Groups), to ensure it is up to date with the latest advances in science and technology.

The process will allow NHS England and NHS Improvement to determine which tests are available within the NHS in England and any tests that will be retired or replaced by more modern methods, such as whole genome sequencing.

The contents, structure and format of the National Genomic Test Directory is copyrighted by NHS England and NHS Improvement and therefore the content is owned, controlled and maintained by NHS England and NHS Improvement. The National Genomic Test Directory may only be amended pursuant to the robust and evidence-based process conducted and approved by NHS England and NHS Improvement.

4 Frequently asked questions

4.1 What is the National Genomic Test Directory and where is it published?

The National Genomic Test Directory (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 the test. The Test Directory is published on the NHS England and NHS Improvement website here https://www.england.nhs.uk/publication/national-genomic-test-directories/

4.2 How was the content of the Test Directory first developed?

NHS England and NHS Improvement established two expert groups – one for rare and inherited disorders and one for cancer, to develop the first publication of the Test Directory. The groups brought together clinicians, scientists, health economists, policy experts, public representatives and patient organisations. The Test Directory builds on the evaluation work that had been undertaken by the UK Genetic Testing Network and through a detailed process combining national and international evaluation approaches, emerging evidence and findings from research and the 100,000 Genomes Project and an analysis of current NHS testing activity. The first version of the Test Directory was published in 2018.

4.3 What does the implementation of a National Genomic Test Directory mean for patients?

The Test Directory is a central feature of the NHS Genomic Medicine Service. Clarifying which tests are funded by the NHS in England and creating a clear and transparent process for the ongoing evaluation of genomic tests helps to secure:

- equity of access for patients to the full range of clinically appropriate genomic tests, using the optimal technology approach, to tackle unmet need and variation;
- keep up to date with the most appropriate technology for the best diagnostic and clinical outcomes and secure comprehensive and affordable provision;
- a standardised testing approach so that all patients get the best standard of care; and
- the ability to introduce new genomic tests in the future and keep pace with the latest research and evidence.

4.4 Who is eligible for testing?

The Test Directory sets out the clinical indications where there is clear evidence for the value of genomic testing for patients. This is primarily in cancer and rare and inherited disorders. Each clinical indication included in the Test Directory has a set of testing criteria that sets out which patients are eligible for testing.

4.5 Can whole genome sequencing be ordered as part of routine care?

Whole genome sequencing (WGS) will help transform the diagnosis and treatment of cancer and rare diseases. NHS England and NHS Improvement will be offering this service to patients where there is evidence that they are likely to receive the most benefit. NHS England and NHS Improvement will continue to review the evidence and expand the number of clinical conditions eligible for WGS as the evidence becomes available.

Clinicians wishing to refer patients for WGS should first speak to their regional NHS GLH or local clinical genetics service (depending on local arrangements) who will be able to advise on the most appropriate tests available. Initially only a small number of indications (as set out in the National Genomic Test Directory) will be available for WGS though we expect to include further indications over time.

4.6 Who can order tests from the Test Directory?

The Test Directory indicates the clinical specialties able to order a specific genomic test, as many of the tests are only relevant for certain specialties e.g. clinicians from Clinical Genetics, Obstetrics, Paediatrics etc. There may be exceptional circumstances where clinicians need to order a test outside the standard repertoire.

4.7 How can tests be ordered from the Test Directory?

Clinicians wishing to request genomic tests can do so by;

- Requesting the clinical indication (name and unique code of the clinical indication), in instances where the clinical indication to be tested is known
- If the clinician is aware that some of the constituent tests which are offered as part of the clinical indication are not needed, they can specify to the lab which constituent tests are required and which are not.

Clinicians should follow local process to request genomic tests. All referrals for testing will be triaged by the local NHS GLH to ensure the most appropriate test is performed. In instances where testing is requested by the clinical indication, the NHS GLH will review the test request and relevant clinical information and select the most appropriate constituent test(s) to facilitate the test request. Testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

4.8 Where is the testing carried out?

Genomic testing for NHS patients is carried out through the seven NHS Genomic Laboratory Hubs across the country, that operate as part of a single national testing network working to common procedures and standards.

The genomic testing in the Test Directory is split into two categories:

- Core genomic tests – high volume tests that will be provided by all NHS GLHs
- Specialist genomic tests – tests that will be delivered by the NHS GLHs that have been appointed as a national specialist test provider due to required expertise. Specialist test providers deliver testing on behalf of the national network.

4.9 How is the testing in the Test Directory paid for?

NHS England and NHS Improvement fund the NHS GLHs to deliver a national genomic testing provision inclusive of the testing in the National Genomic Test Directory. All genomic tests included in the Test Directory are commissioned, and therefore funded, by the NHS in England.

NHS England and NHS improvement will set aside a portion of its budget each year to pay for implementation of changes to the Test Directory. The total funding envelope for updating the Test Directory will be determined on an annual basis. All funding decisions for updating the Test Directory will be made at the end of the evaluation process against all amendments recommended by the Test Evaluation Working Groups, when no further applications are being considered for the relevant year. The funding envelope will be allocated by the Genomics CRG based on the recommendations from the Test Evaluation Working Groups.

4.10 Can genomic tests not in the Test Directory be accessed by the NHS in England?

Only genomic tests included in the Test Directory will be commissioned by NHS England and NHS Improvement, and therefore funded, by the NHS in England and will be available through the NHS GMS.

4.11 How will the Test Directory be kept up to date?

The Test Directory will be reviewed and updated on an annual basis following the evaluation and review process implemented by NHS England and NHS Improvement, to ensure it is up to date with the latest advances in science and technology. For full information on this process and to access all supporting materials, see: https://www.england.nhs.uk/genomics/the-national-genomic-test-directory/

Through this process, applications to propose updates to the Test Directory can be submitted to NHS England and NHS Improvement, which will then be evaluated to inform the annual update to the Test Directory.

4.12 Who can submit an application to update the Test Directory?

Anyone can submit an application to update the Test Directory including but not limited to; clinicians, scientists, patient groups and charities, industry representatives, professional bodies etc.

If a member of the public or patient representative wishes to submit an amendment but requires further guidance, they can contact the Genomics Unit who can signpost to charities and/or support groups who may provide support and expertise.

4.13 What types of amendments can I propose to the Test Directory?

Applications to propose updates to the Test Directory may seek to:

- Add new clinical indications to the Test Directory
- Amend the eligibility criteria, requesting specialties, constituent tests, test targets or technologies for existing clinical indications
- Amend the content of gene panels (see point 2.12)

- Decommission a test where it is obsolete or no longer supported by clinical, scientific or economic evidence
- Move a clinical indication or test to an alternative test method e.g. to WGS

4.14 How do I suggest an amendment to the Test Directory?

The process to amend the Test Directory is outlined in the policy document "Updating the National Genomic Test Directory" which can be found here: https://www.england.nhs.uk/genomics/the-national-genomic-test-directory/

To propose a new clinical indication to be added Test Directory, please complete the application form titled Test Evaluation Application - New Clinical Indication. To amend a clinical indication or its constituent tests already available in the Test Directory please complete the application form titled 'Amendment to Existing Clinical Indication'.

All forms can be accessed via the NHS England and NHS Improvement website: https://www.england.nhs.uk/genomics/the-national-genomic-test-directory/

All applications should be sent to ENGLAND.testevaluation@nhs.net.

4.15 When do I need to submit the application?

Applications can be submitted at any time, however applications submitted after the evaluation cut-off date published on the webpage above will not be considered for the upcoming update and will be reviewed for the next available update.

4.16 Can I resubmit an application if it is not successful?

If an application is unsuccessful or further evidence becomes available, applications can be submitted with the appropriate evidence at any stage.

4.17 Who decides the amendments made to the Test Directory?

To support the process to update the Test Directory, NHS England and NHS Improvement have appointed Test Evaluation Working Groups. The Test Evaluation Working Groups are made up of experts including; clinicians, scientists, health economists, policy experts and patient representatives. There are three Test Evaluation Working Groups for; cancer, rare and inherited disease and pharmacogenomics. The Test Evaluation Working Groups will review the proposed amendments and make recommendations to the Genomics CRG who will prioritise and agree the amendments to the Test Directory each year.

Please note the pharmacogenomics group is currently being established and is not yet operational.

4.18 If an update is made to the Test Directory, when can I order the updated or new test for my patient?

Once a clinical indication or constituent test is listed on the Test Directory it can be requested by clinicians via local processes. The Test Directory will be updated annually and fully implemented by April each year.

4.19 What do I do if I think an update needs to be made urgently before the annual update?

There is a fast track application process for amendments that may be identified as requiring implementation more urgently than through the annual update to the Test Directory, that meet the designated fast track criteria. Please submit the appropriate form and complete the fast track coversheet and submit as per point 4.14.

4.20 How much evidence do I need to submit to support an application?

Submit as much evidence as is possible or available to support the application, which can include for example literature, case studies, recommendations from professional organisations, and evidence from international cohorts etc. In the event that further evidence becomes available after the application has been submitted, this can be sent to the Genomics Unit for addition to the application where possible.

4.21 How do I propose an amendment to a gene panel?

Proposed amendments to the content of NHS GMS virtual gene panels should be submitted as evidence reviews via the Genomics England PanelApp platform: panelapp.genomicsengland.co.uk

NHS England and NHS Improvement will review all evidence reviews associated with NHS GMS virtual gene panels submitted via PanelApp on a quarterly basis. This process does not apply to any NHS GMS wet lab panels. Any amendments to NHS GMS wet lab panels should be submitted via the relevant application form as per the annual application process (see point 4.14).

4.22 How do updates to the Test Directory align with the outcomes of NICE processes or other policy changes?

Updates to the Test Directory may be mandated in-year by policy decisions, such as medicine approvals by NICE or urgent policy statements issued by NHS England and NHS Improvement Specialised Commissioning. NHS England and NHS Improvement are involved in the NICE Topic Selection process to identify medicines which may proceed through the NICE technology appraisal process and include a genomic test as part of the patient pathway. Where relevant medicines are identified, the impact of implementing the genomic testing element of the patient pathway will be included as part of the technology appraisal.

NHS England and NHS Improvement undertake ongoing horizon scanning to identify such potential changes which may impact on or require a change to the Test Directory and will ensure that testing linked to such changes is available within the appropriate time scale, following policy approval.

5 Contact

If you have any questions or would like any additional information, please contact ENGLAND.testevaluation@nhs.net.