NHS England

2018/2019 final draft National Genomic Test Directory FAQ

August 2018

1 Contents

1 Introduction

To enable the NHS to benefit from advances in genomics, NHS England's *Next Steps on the NHS Five Year Forward View* signalled the intention to create a national NHS Genomic Medicine Service. The future service will build on the existing provision of NHS clinical genetic services and the NHS contribution to the 100,000 Genomes Project.

The NHS Genomic Medicine Service 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 future NHS Genomic Medicine Service will consist of the following elements:

- a national genomic laboratory network made up of seven Genomic Laboratory Hubs;
- a National Genomic Test Directory (Test Directory) to underpin the genomic laboratory network covering rare and inherited disorders and cancer and from single genes to whole genome sequencing;
- national whole genome sequencing provision and the underpinning data and informatics infrastructure, including a Clinical Interpretation Pipeline secured through a partnership with Genomics England;
- an integrated clinical service (built by restructuring the existing clinical genetics service) and an evolved role of the NHS Genomic Medicine Centre infrastructure; and
- a national coordinating and oversight function within NHS England (the Genomics Unit).

2 About the Test Directory

For the last 15 years the UK Genetic Testing Network (UKGTN) has provided leadership in this area by publishing the NHS Directory of Genetic Disorders/Genes for Diagnostic Testing, which evaluated and recommended genetic tests for rare and inherited disorders for the NHS across the UK. However, there was not an equivalent for cancer genomic testing. Instead the testing that has been offered within the NHS has evolved over time in-part driven by NICE medicine assessments and the identification of companion diagnostics.

From October 2018 the National Genomic Test Directory will specify 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. Over time, as the evidence develops, the Test Directory will also include other functional genomic tests for example RNA based technologies and proteomics.

The Test Directory will signal the:

- testing available in the NHS in England for rare and inherited disorders and cancer;
- technology platform by which the testing will be delivered;
- clinical requirements for access to the test; and
- funding arrangements for the test.

The first Test Directory will be published for 2018/19 to become operational from October 2018. The Test Directory will specify the testing delivered by the new Genomic Laboratory Hubs that will become operational from October 2018.

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

3 Frequently asked questions

3.1 Where has the Test Directory been published?

The final draft 2018/19 Test Directory has been published on the NHS England website and can be found here https://www.england.nhs.uk/commissioning/spec-services/npc-crg/group-e/e01/.

3.2 How was the content of the Test Directory developed?

NHS England established two expert groups – one for rare and inherited disorders and one for cancer. The panels brought together clinicians, scientists, health economists, policy experts, public representatives and patient organisations. The Test Directory builds on the evaluation work that has 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.

3.3 What do these changes mean for patients?

There is variation in the approach to the commissioning and funding of genomic tests across England, creating inequity as not all eligible patients are currently able to access appropriate testing. Clarifying which tests are funded by the NHS using the Test Directory and creating a clear and transparent process for the ongoing evaluation of genomic tests will help 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 to get the best possible 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

3.4 Who is eligible for testing?

The Test Directory will set 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 qualify for testing. For some clinical indications, these will focus testing on cases where the likelihood of an inherited condition and clinically actionable findings is higher than is currently required by some local practice. The eligibility criteria for rare and inherited disorders is not currently included in the Test Directory but will be published shortly.

As the evidence base develops then 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).

3.5 Can whole genome sequencing be ordered as part of routine clinical care from October 2018?

Access to whole genome sequencing will begin to be planned from October 2018 and be operational from January 2019. If required alternative technology platforms, such as whole exome sequencing or large panels, will be available in the interim. Alternative testing for whole genome sequencing will be reflected in the final version of the Test Directory that will be published ahead of October 2018.

3.6 How can test be ordered from the Test Directory?

From October 2018 an online interactive test ordering tool will be available to enable clinicians to search and order the genomic tests they require. The test order tool will provide a list of the appropriate tests that are included in the National Genomic Test Directory, based upon clinical indications entered by the requesting clinician. After selecting a genomic test, a downloadable template specifically for that genomic test will be provided for the requesting clinicians, which will include and require completion of the minimum data and the laboratory that the sample needs to be sent.

From April 2019 the interactive test ordering tool will be integrated into the National Genomics Informatics System (NGIS) and tests will be orderable online.

3.7 Who will be able to order tests from the Test Directory?

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

3.8 Where is the testing going to be carried out?

Genomic testing for NHS patients will be carried out at genomic laboratories across the country. NHS England is consolidating the current NHS genomic laboratories into a single national testing network of seven Genomic Laboratory Hubs 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 Genomic Laboratory Hubs
- Specialist genomic tests – specialist tests that will be delivered only by the Genomic Laboratory Hubs that have been appointed as a National Specialist Test Provider due to the fact that they have the required quality and expertise

3.9 How will the genomic testing in the Test Directory be paid for?

Currently there is variation in the approach to the commissioning and funding of genomic tests across England, creating inequity as not all eligible patients are currently able to access appropriate testing. The Test Directory will clearly indicate the commissioner responsible for funding each genomic test. In summary testing for NHS patients in England are funded as follows:

- *Testing for rare and inherited disorders* will be funded nationally by NHS England
- *Whole genome sequencing* will be funded nationally by NHS England
- *Cancer genomic testing* is included under the National Tariff Payment System (excluding whole genome sequencing which NHS England is funding nationally) and are included within the HRG tariff payment for an individual patient. For example EGFR testing for lung cancer, along with oestrogen (+/- progesterone) receptor status and HER2 testing for breast cancer have been established tests within the NHS for a number of years
- *Testing for inherited cancer*, for example inherited breast cancer at high familial risk, will be funded by NHS England

3.10 How will the directory be kept up to date?

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

The Test Directory will be updated on an annual basis and NHS England, working with colleagues from the Devolved Nations, will implement a clear and transparent process for the future evaluation of new genomic tests, supported by a Clinical and Scientific Expert Panel, and to 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.

The UK approach to evaluating genomic tests will be published and NHS England will also publish a policy outlining the approach to commissioning and funding the tests.

3.11 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, and therefore funded, by the NHS in England and will be available through the Genomic Laboratory Hubs. NHS England will put in place a process to annually update the

Test Directory in order to keep up to date with the latest advances in science and technology.

3.12 Will a final version of the Test Directory be published before October 2018?

A final version of the Test Directory will be published ahead of October 2018. This will include the changes required to reflect the fact that whole genome sequencing will be available from January 2019.

4 Contact

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