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East Genomics Newsletter

October 2021

Welcome to the first newsletter from East Genomics. In this newsletter, we will share information on what's been happening at the East Genomic Laboratory Hub together with progress of a new arm of the service - the East Genomic Medicine Service Alliance (East GMSA). Over the summer we launched a **new website**, mentioned below, where you can access all the referral forms and information you need; we would welcome your feedback.

As many of you may already be aware, the **National Genomic Test Directory** has been updated this week. This includes some changes to a few Rare and Inherited Disease tests, as well as Cancer tests. Here we will explain what those updates mean for you as healthcare professionals within the East and East Midlands region.

National Genomic Test Directory – October update

This week, updates to the National Genomics Test Directory have been made available to view on the NHS England website.

The updates (the first of three updates expected before April 2022) includes new and amended tests on both the Rare and Inherited Disease Test Directory and eligibility criteria, as well as the Cancer Directory.

The expectation is that each GLH will work to deliver the updated test directory within three months of publication; thank you in advance for your patience with the Genomic Medicine Service as these updates are made operational across

the country.

For the **rare and inherited disease test directory**, the most significant change is the inclusion of additional clinical indications for Whole Genome Sequencing (WGS). These broadly affect Neurology, Ophthalmology, Immunology, and Cardiology and can be viewed via the table provided on our East Genomics website. New or amended tests are noted within the directory (Column G) and also within the Eligibility Criteria document (a change log is also detailed on page 387).

For the **cancer test directory**, some changes have been introduced to the haemato-oncology section. However, these relate mainly to nomenclature alterations and to the amendment of entries to reflect pre-existing routine practice.

Within the Solid Tumour (adult) section, a test used for the identification of Lynch Syndrome - known as MLH1 promoter methylation - is now included for patients with endometrial cancer (Test number M215.2); this test is therefore now centrally funded. A handbook with helpful flowcharts (see page 8, Option 1) has been developed to support the implementation of the Lynch Syndrome testing and surveillance pathways, should this be useful. There have also been some revised gene targets for select solid cancer indications and tests that are duplicated have been removed.

Thank you to those who have contributed to these updates. NHSE recognise this is the first time the directory has been updated; if you have any comments on the latest version please email england.testevaluation@nhs.net

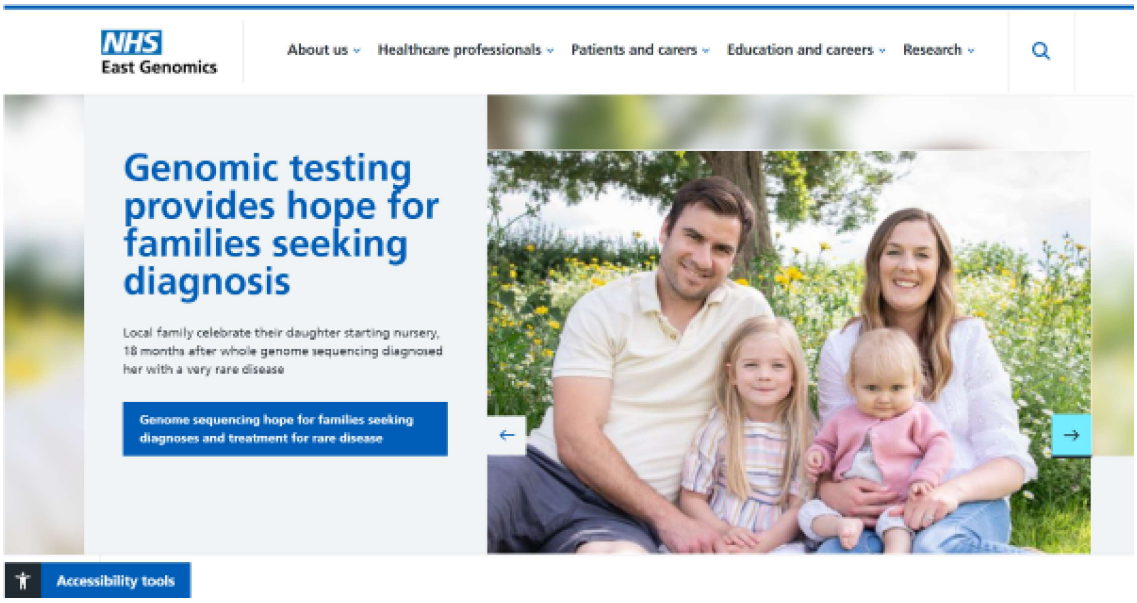
If you have any questions in relation to test orders in the region please don't hesitate to get in to contact with us, on:

- emee.glh@nhs.net for any WGS enquires
- geneticslaboratories@nhs.net for any other enquiries

Future updates to test directory – deadline date

- The deadline for the receipt of applications for the next update to the National Genomics Test Directory has been extended to **5pm on 29 October 2021**.

- Further information concerning applications and updates to the Test Directory is available on the NHS England website
- If you have any questions or require any further information at this stage, please email england.testevaluation@nhs.net



New website launched!

Our new website is now launched on www.eastgenomics.nhs.uk. The new site has been developed to:

- provide a functional GLH site for clinicians
- include all essential information for ordering tests plus educational content
- incorporate information and updates from East GMSA as well as East GLH
- house useful information for patients and the public
- promote news and support the organisation's educational and recruitment priorities

We will continue to develop the new site and add material that will be of use to our users. If you have any feedback or suggestions at any time please do email lorna.hamblin@addenbrookes.nhs.uk.

Following the launch of our new website www.eastgenomics.nhs.uk, we have also now rebranded our Twitter account to @East_Genomics. If you don't already follow us, please do.

Cambridge Genomic Laboratory Hub - major refurbishment work to aid high throughput of tests

We are extremely proud to have recently completed major building works at our lead laboratory based at Cambridge University Hospitals NHS Foundation Trust. This will help us deliver an enhanced genomic service for the East of England and East Midlands.

East GLH is now well placed to provide rapid turnaround of tests and a higher throughput. The work has included:

- integration of our haemato-oncology team alongside our rare disease and cancer teams
- an automated DNA extraction facility, as well as a separate sequencing hub with a dedicated area for next generation sequencing
- an increase to our 'dry' laboratory capacity so that we can accommodate more staff and have a dedicated administration area in the department
- the development of an efficient one-way system for staff and samples, designed to also prevent contamination

The new and improved laboratory has attracted several key stakeholders since it has been completed including staff from the Department for health and Social Care (DHSC), Office for Life Sciences (OLS), as well as celebrity Jean-Christophe Novelli and his wife Michelle.

As well as taking a detailed tour of the laboratory, Jean-Christophe and Michelle spoke to staff and shared their experience of genomic testing of their youngest son.

East Genomic Medicine Service Alliance (East GMSA)

The East Genomic Medicine Service Alliance (GMSA) is now starting to take

shape. This part of East Genomics has been set up to embed genomic medicine across the NHS; it will ensure all eligible patients are able to access appropriate genomics tests and the personalised treatment and research opportunities that this brings.

This will be delivered through a programme of workforce transformation together with national and regional projects. Current national projects cover various topics: Lynch syndrome, sudden cardiac death, pharmacogenomics, familial hypercholesterolaemia (FH), a pathology accelerator programme to support rapid cancer diagnostics, and embedding genomics across nursing and midwifery practice. Local projects include: the development of cloud based Phenotips to improve access to the test directory, supporting WGS consenting in paediatrics, and raising awareness of prostate cancer to address inequity of care for those at high risk.

The GMSA covers the same geographical region as the East GLH. The four GMSA partners are:

- Cambridge University Hospital NHS Foundation Trust
- University Hospitals of Leicester NHS Trust
- Nottingham University Hospitals NHS Trust
- Norfolk and Norwich University Hospitals NHS Foundation Trust

Further information on the East GMSA will be made available on our website in due course.

Regional Genomics survey

Eastern AHSN

NHS East Genomics

Regional Genomics Survey – coming soon!

We will soon be sending out a survey to help us capture information that will influence our workforce education and training efforts as well as communications strategies in relation to genomics across the East of England

and East Midlands.

Please do look out for the email; we will also make it available on our website next week. Your response and further circulation of the survey will help us achieve equity of access for patients across our region.

Keen to learn more about Whole Genome Sequencing (WGS)?
Genomics England and Health Education England are running an online course, starting on 18 October (open for enrolment until 19 December) if you would like to find out more about WGS. You will hear from leading expert clinicians and researchers working in cancer, rare disease and infectious disease about how WGS is helping them find answers for patients.

The course is delivered through a range of articles, videos, animations and discussion forums facilitated by expert mentors. For more information or to sign up, please visit: https://www.futurelearn.com/courses/whole-genome-sequencing

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Patient information leaflets on Whole Genome Sequencing available in several languages

Translations of the whole genome sequencing (WGS) patient information leaflets as well as the Record of Discussion form have been produced from NHSE. These have been uploaded to our website:

- Record of discussion forms
- Patient information leaflets (Cancer, Rare Disease and Easy Read versions)

Languages available are: Polish, Punjabi, Urdu, Bengali, Gujarati, Arabic, Chinese, Portuguese, Spanish and Welsh.

We are currently looking into the cost of producing further languages to serve our communities across the whole East Genomics catchment area. If you do have any specific needs please do email us.

NHS GMS Research Collaborative

The NHS Genomic Medicine Service (NHS GMS), Genomics England and the National Institute of Health Research have joined forces to set up the NHS GMS Research Collaborative. The collaborative will improve healthcare by supporting and facilitating genomic research that has the potential to deliver better outcomes for patients now and in the future.

Proposals can be made from all Individuals and interested parties (including commercial partners) to access expert advice and support from colleagues working in the NHS GMS to help develop their healthcare projects. In addition to a range of other benefits, the NHS GMS Research Collaborative can provide access to the clinical and laboratory infrastructure available in the NHS to support genomic research through the regional NHS GMS Alliances and the National Genomic Research Library.

You can find out more about the NHS GMS Research Collaborative, its guiding principles and how to make a submission here.

If you have any questions in relation to this newsletter or any of the content, please email lorna.hamblin@addenbrookes.nhs.uk

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