

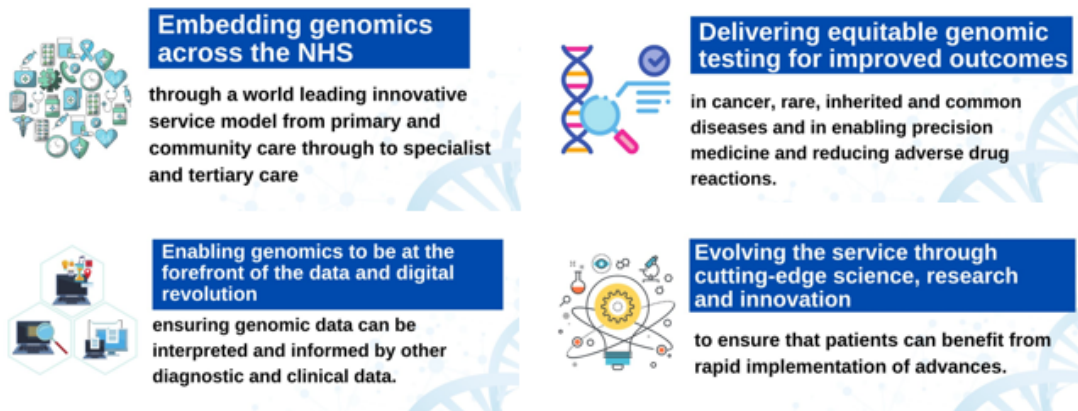
East Genomics Newsletter

November 2022

Welcome to the quarterly newsletter from East Genomics in which we share updates from the [East Genomic Laboratory Hub](#) (East GLH) and the [East Genomic Medicine Service Alliance](#) (East GMSA).

As genomic testing is becoming more mainstream, we welcome feedback on this newsletter and suggestions on what you would like included or shared via other channels. Please do drop us [an email](#).

NHS Genomics Strategy



On 12 October at the National Genomics Summit in London, Amanda Pritchard, CEO of the NHS, launched the first NHS Genomics Strategy, which sets out the ambitions for embedding genomics in the NHS over the next 5 years with **four priorities**:

- **Embedding genomics** across the NHS
- Delivering **equitable genomic testing** for improved outcomes
- Enabling genomics to be at the **forefront of the data and digital revolution**
- Evolving the service through **cutting-edge science, research and innovation**.

The full strategy, as well as an executive summary, can be viewed here [NHS England » Accelerating genomic medicine in the NHS](#), so please do have a read when you can. Easy read versions are available on request.

Health Education England has published a helpful [five top take takeaway messages](#) from the publication.

Finally, if you are interested to see some of the presentation slides from the NHSE Genomics Healthcare Summit, they can be [accessed here](#) and clicking on the relevant speaker name.

Public consultation on clinical genomics services in England

Below is a link to updated information on the public consultation on the proposed Clinical Genomics Service Specification. Whilst the consultation will formally close on Sunday 6 November, a QA document is now made available, and stakeholders can make comments during w/c 7 November if they wish.

[Public consultation on clinical genomics services in England | East Genomics](#)

National Test Directory update

New tests and amendments have now been added to the [National Genomic Test Directory](#) (Test Directory) as part of the October 2022 update.

There are currently tests covering 3,200 rare diseases and more than 200 cancer indications in the Test Directory. Please do review the Directory for the specific updates.

If you have a question or want to find out more about the Test Directory, please email england.testevaluation@nhs.net

Test order forms

Rare Disease - We recently sent out some communications to all those who have ordered Rare Disease Tests through our Laboratory before. Please note, that all test orders on samples stored in our Laboratories should be submitted using an [East GLH Rare Disease Test order form](#).

The Cambridge, Nottingham and Leicester laboratories will no longer accept email requests for the activation of tests on stored samples. The information is available on [our website](#).

DPYD tests - The test referral form for DPYD testing for 5-fluorouracil (chemotherapy) toxicity has been updated with new indications. The updated form is [available here](#).

Cambridge Genomics Laboratory update

Turn around times - We update turn around times (TATs) on our website at the end of each month (for the month prior). Please do refer to [this page](#) if you need to find out the TATs for a specific test. Unfortunately we are unable to include TATS for test carried out at other Laboratories/GLHs at this time.

Move to NHS.net emails - Cambridge University Hospitals NHS Foundation Trust is moving to NHSMail and from 1 December 2022 it will become the Trust's primary email account. This is therefore a change for staff and group email addresses at the Cambridge Genomics Laboratory. If you email a particular person or team at the Lab, please do get in touch with them or email [our general email address](#) if you have not received a new contact email address by the end of November.



Whole Genome Sequencing in Paediatric Cancer

In October, we held a joint event **Implementing Whole Genome Sequencing (WGS) in paediatric oncology practice** at the Wellcome Genome Campus in Hinxton. The event was organised by [Wellcome Connecting Science](#) and co-developed with East Genomics, The University of Cambridge and the Wellcome Sanger Institute.

The aim of the event was to raise awareness of WGS, highlight how some national and international centres are using WGS and provide an opportunity for an open discussion so that delegates could discuss issues around implementation of this particular test and consider solutions to improve uptake.

The targeted event attracted a good number of oncologists and pathologists from across England, Scotland, Wales, Northern Ireland and Ireland. One patient story was recently picked up by the BBC. Oliver, a baby who was born with a large tumour on his leg, was spared chemotherapy and surgery after WGS revealed the lump was harmless. You can read the [full story here](#).





Essential genomics information for clinicians

East GMSA recently issued a [Genomics - essential information for clinicians](#) pack to Medical Directors/Chief Medical Officers at acute trusts across our geography. The pack is aimed at clinicians and is intended to raise awareness of genomics and genetic tests available, as well as the process for requesting tests within our region and an overview of the support, training and education offered by East GMSA and other bodies.

For Departments or clinicians wishing to learn more about embedding genomic testing into their clinical pathways please contact us directly by emailing egmsa@nnuh.nhs.uk to discuss what type of additional support can be provided, such as webinars or East GMSA attendance at clinical meetings/forums.

Continuing the theme of raising awareness, East GMSA Medical Lead, Matt Hall appeared on the Nottingham University Hospitals (NUH) Clinical Research Podcast recently to talk about what genomics means in reality to healthcare professionals, and how they can access genetic medicine services - including testing for patients, in their day-to-day practice. You can listen to Matt's episode, and other episodes, on [Anchor](#), [Google](#), [Apple](#), or [Spotify](#).



Regional genomics PPV panel

East GMSA has established a Patient and Public Voice (PPV) Panel to help ensure that the views of patients, carers and families are at the heart of genomic medicine in our region. The panel's remit is broad and evolving, from reviewing patient and public-facing communications, to involvement in pathway development of genomic services in hospitals.

The panel has first-hand experience across a range of genetic conditions, as well as a wealth of skills and experience across relevant sectors. If you would like to engage the PPV panel in any aspect of your work, please contact the Panel Chair, Chris Hind, at Chris.Hind@nnuh.nhs.uk for an initial discussion.

Upcoming events



This FREE conference, organised by East GMSA is aimed at nurses and midwives - across a wide range of roles and from 'ward to board' - working in the East Midlands or East of England.

It will outline how genomics and genetic testing is helping patients in the East Midlands and East of England, the crucial role nurses and midwives are playing in embedding genomics into routine NHS care, and the role of East GMSA in supporting all of this.

For further information and to register for this event, please visit the [event website](#).

National pathology workshop: From diagnosis of cancer to diagnosis of Lynch syndrome.

Lynch syndrome (LS) is an inherited condition caused by pathogenic variants in DNA genes which result in an increased risk of colorectal, endometrial, and other cancers. It affects approximately 1 in 300/400 people, with 200,000-

300,000 people likely to have this condition in the UK. It is estimated that only 5% of people with LS have been diagnosed with this condition.

NICE guidelines recommend universal testing for LS in people with colorectal (CRC) [DG27] and endometrial cancer [DG42].

A national workshop will be held on 8 November (1 - 2pm) which will outline the responsibilities of pathologists in the Lynch syndrome testing pathway. It is aimed at pathologists, biomedical scientists and any other pathology department or Colorectal and Gynae cancer MDT staff. You can register for the [event here](#).



National nursing workshop: From diagnosis of cancer to diagnosis of Lynch syndrome

[Register here](#) to hear from Laura Monje-Garcia (National Lynch Syndrome Project Lead Nurse, North Thames GMSA), Tracie Miles (Associate Director of Nursing and Midwifery, South West GMSA), and your regional Lynch syndrome nurse/s to find out more about the testing pathway, role of the nurse, and mainstreaming germline genetic testing in routine oncology clinics.

The event will run on Tuesday 29 November (1 - 2pm) and Monday 19 December (12 - 1pm)



Genomics Education for Psychological Professions

The North West GMSA is hosting a free online webinar on Tuesday 29 November (1 - 2pm) aimed at qualified and training psychological professionals in the North West.

This webinar will provide you with an introduction to genomics and aims to develop the collaboration between genomics and psychology. To find out more and to register please [visit the website](#).



Next steps for genomics in the UK

This London conference, organised by Westminster Health Forum, on Tuesday 13 December will discuss the next steps for genomics in the UK. It will be an opportunity to explore the future of genomic transformation in the NHS, looking at screening and personalised medicine, workforce education and training, and the options for pharmacogenomics in clinical practice. Find out more and register on the [event website](#).



Quick links

[Training and courses](#)

[Resources and support](#)

[Genomics Education
Programme](#)

The October bulletin from Genomics Education Programme (GEP) can be [viewed here.](#)



Contact us

- Enquiries regarding any specific genomic tests should be directed to East Genomic Laboratory Hub (East GLH) on geneticslaboratories@nhs.net
- Enquiries in relation to transformational projects and wider engagement initiatives should be directed to the East Genomic Medicine Service Alliance (East GMSA) on egmsa@nnuh.nhs.uk

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