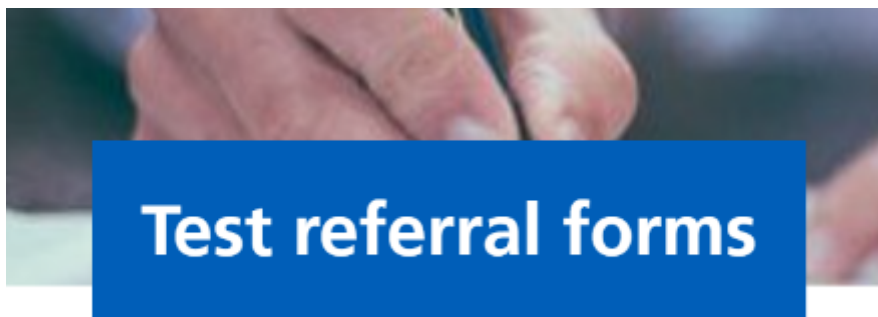


East Genomics Newsletter

February 2022

Welcome to the quarterly update 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 well as national updates and online training opportunities that we would like to highlight.

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](#).



East Genomics website developments

With more clinicians ordering genomic tests, we've made a few changes to the East Genomics website to make it easier and quicker for clinicians to order their required tests.

Whilst there are many clinicians new to genomic testing, in particular WGS, there are also many who know what test they want to order and what forms they need to use. We've therefore developed a 'test referral form' index page for those clinicians who are able to go directly to the forms they need.

New page link: [Referral forms Index page | East Genomics](#)

Clinicians who aren't too familiar with the ordering process, can still go into the relevant ([Rare and Inherited Disease](#), [Cancer \(Solid Tumour\)](#), [Haematological Malignancy](#) or [Pharmacogenomic](#)) pages where there is further detail.

Feedback is always welcome to enable us to make further improvements. Please drop us [an email](#) at any time.

National Genomic Test Directory updates

Rare and inherited disease - additional tests

Whole genome sequencing (WGS) is now available for a number of additional clinical indications. The [National Genomic Test Directory for Rare and Inherited Disease](#) has had two updates at the end of 2021 to include these tests, now delivered by WGS, within **cardiology, immunology, renal and ophthalmology**.

Additional clinical indications within the neurology specialism will soon be transitioning to WGS; we will be in touch to provide training and guidance.

A list of when certain tests and indications will transition to WGS is available on our [Rare Disease WGS web page](#). The page also includes updated instructions and flowcharts for ordering, test order forms and record of discussion forms.

We expect further updates to the directory in the Spring and will share information with the relevant clinicians as soon as necessary.

If you have any questions, feedback or wish to request additional training for ordering WGS please [email us](#) and include 'Rare Disease WGS' in the subject heading.

Cancer updates

The most significant amends to the [National Genomic Test Directory for Cancer](#) update in December were the addition of tests for new clinical indications to help inform therapy choice or structural variant detection.

These included new tests for: **oesophageal cancer (M236), gastric cancer (M237, small bowel cancer (M238) and thyroid hurtle cell carcinoma (M239)**.

Other new tests changes have been made to the Test Directory and further updates are expected by the end of the March. Like the other GLHs, we strive to provide these new tests as soon as possible and will keep all NHS Trusts in the region informed.



Study reveals the benefits of ‘whole genome sequencing’ for children with cancer

In November, our Scientific Lead for Solid Cancer, Dr Patrick Tarpey, presented the findings of a study on the utility of Whole Genome Sequencing in children at the National Cancer Research Institute (NCRI) Festival.

The study, part of the 100,000 Genomes Project, involved 36 children treated at Addenbrooke’s hospital in Cambridge and included 23 different types of solid tumour. Careful analysis of the WGS data revealed several potentially important variants, a number of which were hard to spot. This information helped to influence the diagnosis and/or treatment of many of these patients.

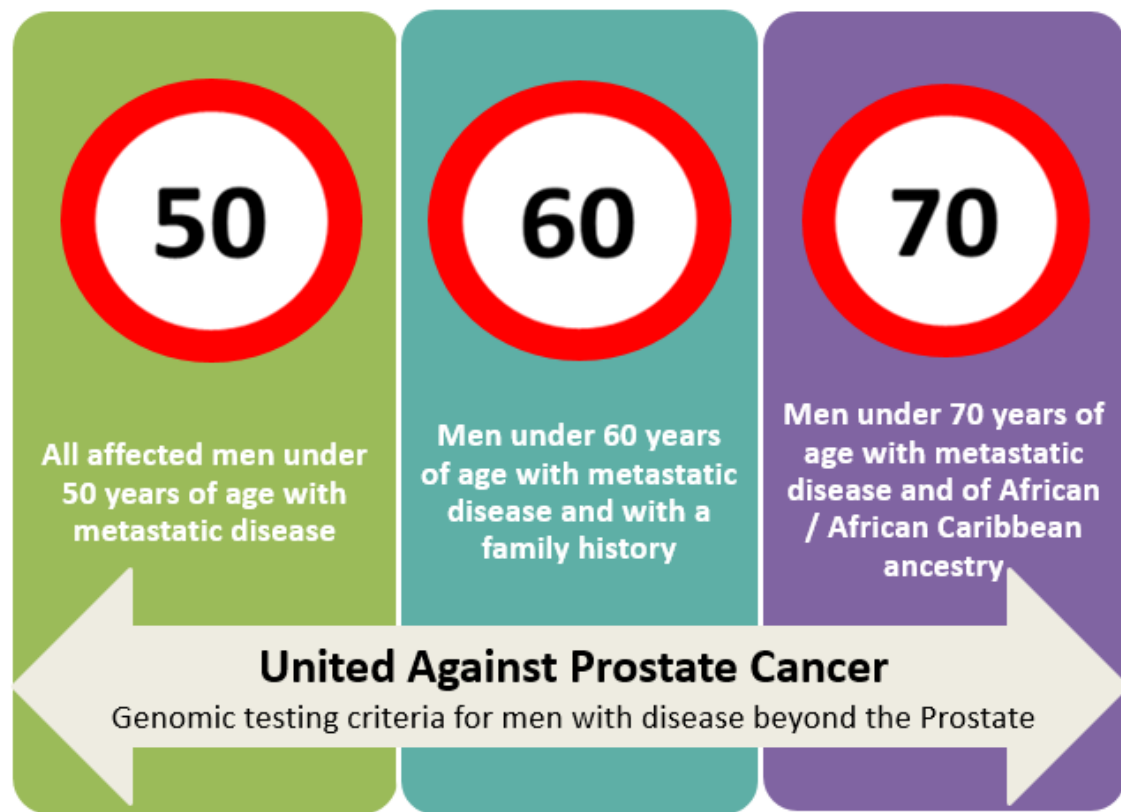


Dr Patrick Tarpey
Lead Scientist for Cancer

“Our results suggest that offering whole genome sequencing to all children with cancer will:

- provide more accurate diagnosis and prognosis
- show whether there could be any hereditary cancer risk
- and improve treatment options.”

The research generated international press coverage. You can read about the study [here](#). We have also shared detail of how WGS helped confirm the diagnosis and treatment for [one of the patient's in the study.](#)



United Against Prostate Cancer

The 'United against prostate cancer' project, led by the University Hospitals of Leicester (UHL), aims to improve prostate cancer pathways and support for patients; it will tackle disparities and help provide equitable access to prostate services.

The work is part of a national pilot project which has a focus on establishing genetic testing of prostate tumour tissue samples to help identify the causes of this disease. This could be useful in planning a patient's treatment and/or determining if their relatives could also be at an increased risk of breast, ovarian or prostate cancer.

From April 2022 we aim to offer testing across the whole region for all affected men under 50 years of age, under 60 years of age with a family history or under the age of 70 and of African/African Caribbean ancestry with metastatic disease.

Read more about this initiative from the East Genomic Medicine Service Alliance (East GMSA) [here](#).

Regional Monogenic Diabetes Clinical Network

East GMSA has established a regional Monogenic Diabetes Clinical Network,

which will meet bi-monthly, starting Tuesday 22 February.

The Network will provide an ongoing supportive learning environment for clinicians who have undertaken Monogenic Diabetes training and are keen to embed genomics within their local pathways.

Meetings will focus on shared learning, including case-based discussion, patient stories, subject matter expert guest speakers, journal article reviews and networking.

Diabetes specialist nurses across the region with an interest in MD are invited to join the network by emailing marie.baker@nnuh.nhs.uk. If you have not managed to do the training, you can find this [here](#).

EDUCATION & TRAINING FOR CLINICIANS

Patient Choice in genomics - courses now available

Two e-learning courses on Patient Choice have been developed for NHS healthcare professionals who are ordering whole genome sequencing for patients with a suspected rare, inherited condition or cancer.

Developed by NHS East GLH education and training lead Dr Gemma Chandratillake, the courses explore the Patient Choice framework through textual, video and interactive content. Available on Health Education England's e-Learning for Healthcare (e-LfH) platform, you can select either the course on testing for a [rare, inherited disease](#), or that [for cancer](#).

From Niche to Necessity: Genomics in Routine Care

- Earlier this year, NHSE held a three-part webinar series to help staff find out more about genomics, how it is transforming patient care, what it will mean for healthcare professionals and further educational opportunities now available.
 - The webinars, produced alongside Health Education England (HEE) Genomics Education Programme and RCNi are useful for a wide range of staff interested to learn more about genomics. The recordings are available on demand via the [HEE website](#).
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Advanced webinar series on 'How to read a cancer genome'

The Genomics Education Programme (GEP) recently organised a special three-part educational programme on '**how to read the cancer genome**' with Professor Serena Nik-Zainal.

The webinars, on the 'Basics of cancer genomics', 'Tertiary analysis beyond driver mutations' and 'Walking through examples of WGS holistic interpretation together' are available on the [Genomics Education Programme website](#).

These advanced level webinars are aimed at those working in oncology (clinical and medical oncologists, specialist nurses, pathologists and researchers) and genomics (clinical geneticists, clinical scientists and genomic counsellors).



E-learning courses developed to help clinicians order genetic tests for patients with breast cancer or inherited cardiac conditions

Clinical Geneticists and Genetic Counsellors at Cambridge University Hospitals (CUH) have developed several short e-learning modules to help healthcare professionals across the East of England and East Midlands offer genomic testing to patients.

Two modules are now live and are now being rolled out across the region to oncologists and cardiologists. They focus on hereditary (or germline) genetic testing in patients with breast cancer and testing for those with inherited cardiac conditions.

You can self enrol for the [online modules here](#). A further module for primary care clinicians is expected to be launched later in the year.

Virtual Cancer Genetics Course

Guys and St Thomas's NHS Foundation Trust has developed a virtual course for healthcare professionals working in primary care and specialist settings including oncology, breast care, gynae-oncology, gastroenterology and screening services.

Participants will be equipped with the basics of cancer genetic counselling and sessions will include approaches to genetic testing, management of hereditary cancers and consent taking. For further information, please visit [their webpage](#).

GeNotes: genomic notes for clinicians

Subject matter experts across the NHS are working in collaboration with HEE's Genomics Education Programme to launch GeNotes as an educational resource for healthcare professionals. GeNotes will provide clinicians with educational information at the point of need (In the Clinic), with opportunities for extended learning (Knowledge Hub).

The work is currently in development and working groups have already been set up in a number of specialties. If you would like to see your clinical specialty included and would be willing to contribute to the working group, further information and contact details can be [found here](#). GeNotes is expected to be made public in 2022.

GeNotes will form an important part of [The Framework Project Initiative](#) which is a step-by-step method to identify workforce development and education needs aligned to patient pathways.



Healthcare professional survey

**Ensuring equity of
access to the
Genomic Medicine
Service**

Regional Customer Survey

We are currently carrying out a regional customer survey to help us gain insight into the knowledge and use of genomic testing by healthcare professionals across East of England and East Midlands.

If you have not yet taken this short survey it would be really helpful if you could take five minutes to share your knowledge and awareness of genomic testing. This will help shape and improve our education and communications plans and ultimately help ensure equity of access to testing.

The survey is available on [our website](#). Thank you.

Rare Disease Day – Monday 28 February

Rare Disease Day offers an opportunity to highlight the work being done to deliver better care for patients living with a rare disease and the families that support them. If you have a passionate clinician delivering outstanding care for patients or a patient representative who can shed light on their experience then please share these on your social channels.

The NHS Genomic Medicine Service as well as East Genomics will be sharing video and content from patients, clinicians and others on their Twitter accounts ([@NHSgms](#) and [@East_Genomics](#)) so please feel free to share these.

There are some interesting infographics and materials on the [Rare Disease Day website](#).

Please send future newsletter content or suggestions to
lorna.hamblin@addenbrookes.nhs.uk

