TRANSFORMING PATIENT CARE THROUGH GENOMICS

A report from the NHS South East Genomic Medicine Service

November 2024

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Transforming patient care through genomics

The NHS is delivering a world leading Genomic Medicine Service.

The UK Genomics Strategy, *Accelerating Genomic Medicine in the NHS*, sets out the NHS ambition to be the first national health system in which genomics is part of routine clinical care, and central to a future, sustainable healthcare model in the NHS.

As leaders of the Genomic Medicine Service for the South East, we want to highlight how genomics is already delivering for patients, and how we hope, with your support, to be able to deliver more.

By further embracing genomics we will be able to:

- **Offer definitive and earlier diagnoses**
- **Provide targeted treatments and therapies**
- **Reduce waiting times for specialist services**
- **Reduce the NHS burden by identifying people who are not at risk of inherited diseases**
- **Support delivery of the NHS Long Term Plan, particularly its cancer ambitions**

This report highlights the achievements of the Genomic Medicine Service in the South East, and crucially outlines how we want to work with you, and your organisation, over the next year to embrace genomics, which is already impacting every aspect of the NHS across primary and community care through to secondary and specialist tertiary services.

Please get in touch, and join us on our genomics journey.

Ian Abbs

Chief Executive Officer,
Guy's and St Thomas'
NHS Foundation Trust

Jacqueline Totterdell

Group Chief Executive,
St George's, Epsom and St
Helier University Hospitals
and Health Group

WHAT IS THE SOUTH EAST GENOMIC MEDICINE SERVICE?

We deliver NHSE commissioned and centrally funded genomic testing across Kent, Medway, Surrey, Sussex and South London covering a population of 9 million people.

We are one of seven regional services working together to deliver consistent and high quality genomic testing and services across England.

Our service consists of the:

- **South East Genomic Laboratory Hub** (GLH) which delivers genomic testing for solid tumours, haematological malignancies and rare and inherited diseases
- **South East Genomic Medicine Service Alliance** (GMSA) which works with partners to embed genomics in to clinical pathways.
- A close working partnership with our two regional **Clinical Genetics Services**.

We work together to provide improved access to genomic medicine.

The benefits of genomic testing for patients can be significant. Genomics can enable:

- **Accurate diagnoses** & detailed understanding of underlying genomic causes of cancer and inherited diseases.
- **Precision medicine and personalised care**, including access to more effective treatment particularly for cancer patients, and fewer adverse side effects where genomic results can predict how people may react to treatment.
- Opportunities for **prognostics & preventative approaches** including 'cascade testing' for families to offer **earlier diagnosis, surveillance and proactive management** before disease develops.
- Identification of patients who are eligible for **clinical trials,** including the Cancer Vaccine, and access to **peer support.**

We are supporting your workforce to offer these benefits to our patients by accessing the right tests for the right patients, at the right time, through new models that support better equity of access and faster end to end pathways.

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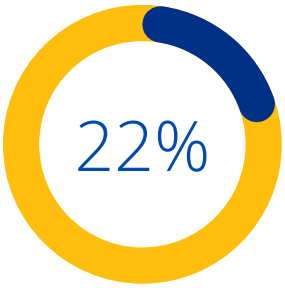
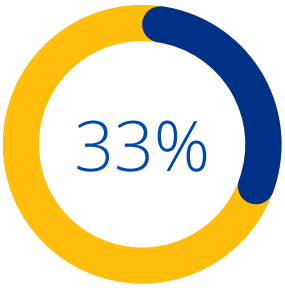
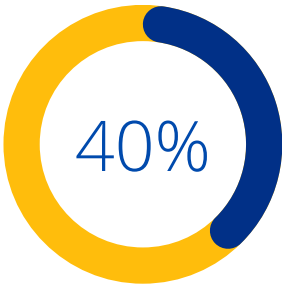
OUR 2023 STORY



increase in genomic testing in one year. We issued 108,000 genomic reports last year.

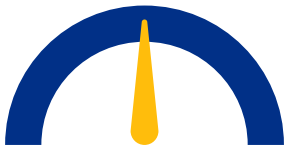


increase in cancer testing in one year thanks to new testing for breast, ovarian & prostate cancers.



increase in testing across Kent, Surrey & Sussex since last year reducing regional inequity.

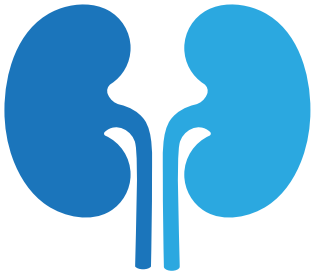
Last year we trained over 2,250 healthcare professionals about how genomics applies to their practice.



Families of over 50 people have been referred by Coroners for genetic investigation for inherited cardiac conditions.

Visited 11 NHS Trusts talking directly to clinicians about genomics.

200% increase in screening for Lynch Syndrome in bowel & endometrial cancer patients since 2021.



700% increase in genetic testing for kidney disease in the South East.

800% increase in neurological genomic testing across the South East.

We've established new genomic pathways in the South East this year including:

- Somatic and Germline **testing for BRCA gene variants** to provide access to the latest PARP inhibitor treatment e.g. Olaparib
- Somatic testing for **homologous recombination deficiency** (HRD) in patients with ovarian cancer to inform treatment decisions and optimise care.
- **ctDNA pilot testing** in Non-Small Cell Lung Cancer which, as a blood test with a 14 day turnaround time, is offering faster, less invasive diagnoses and directly contributing to personalised treatment.

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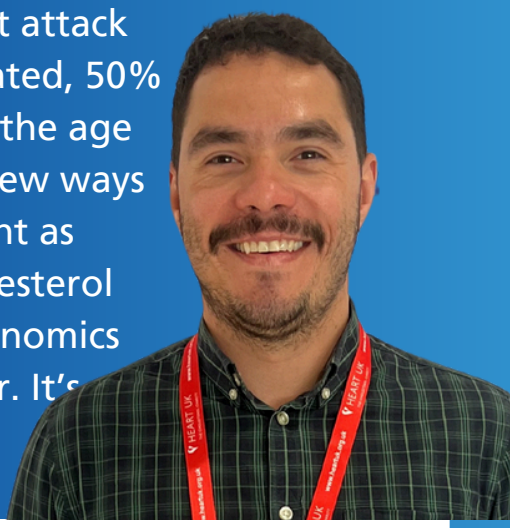
WE'RE ALREADY MAKING A DIFFERENCE TO YOUR PATIENTS

PREVENTING HEART ATTACKS & STROKES

By diagnosing and treating inherited conditions, such as Sudden Cardiac Death and Familial Hypocholesteraemia, we can actively prevent heart attacks, strokes and premature deaths.

"Our aim is to prevent our patients from having a heart attack or a stroke. If Familial Hypercholesteremia is left untreated, 50% of men and 30% of women will have a heart attack by the age of 50 and 60, respectively. It's important that we find new ways to diagnose people and get them on the right treatment as quickly as possible, which is where the idea of the Cholesterol Clinic came from. Healthcare professionals are using genomics and family histories to detect and treat patients quicker. It's already preventing premature deaths."



Pedro Bandeira is a senior pharmacist & independent prescriber specialising in lipid management at St. George's Hospital in London

What are we offering you?

Genetic testing for inherited cardiac conditions.

Early detection of people at risk of genetic cardiovascular disease.

Join our Pharmacy Professional network which supports pharmacists across the South East.

What do we need from you?

Get involved in our Sudden Cardiac Death work.

We're seeking new sites to expand our work with Coroners to identify & prevent further Sudden Cardiac Deaths.

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CANCER: PREVENTION, EARLY DIAGNOSIS AND PERSONALISED TREATMENT

Genomics is already playing an important part in helping us achieve the NHS Long Term Plan cancer ambitions in early diagnosis, personalised care and improved survival.

Genomics can:

- **Predict** when a person is at higher risk of developing cancer, enabling risk reduction interventions, stratified screening programmes, and early diagnosis if disease prevention isn't possible.
- Offer **precise cancer diagnoses** at the start of the pathway, using innovative technologies to characterise tumours, providing prognostic information and informing optimal treatment.
- Provide **access to precision medicines** early in the patient journey or enable entry into clinical trials, as well as monitor response to treatment and disease progression, together improving survival outcomes and quality of life.
- Determine when an existing cancer has heritable genomic origins, thus **allowing family members to seek predictive testing.**

In 2023/24 we delivered

52,000 tumour tests cancer patients across 23 NHS Trusts

25% increase in genomic cancer testing since 2022/23

Supported & educated colleagues to establish new testing pathways

CANCER: PREVENTION, EARLY DIAGNOSIS AND PERSONALISED TREATMENT

What are we offering you?

Comprehensive solid tumour genomic testing in line with the National Genomic Test Directory.

Regional Genomic Tumour Advisory Board (GTABs) for all eligible patients to identify those who may benefit from clinical trials.

Dedicated people to assist and advise on cancer genomic pathways.

What do we need from you?

Can you identify local histopathology leads who could work with us to streamline our end to end cancer testing pathways?

Can you, or your teams, help us to deliver the Cancer Genomic Improvement Plan? Get in touch.

Encourage your teams to attend our education & training sessions.

Ian was diagnosed with bowel cancer at an early stage.

When he had surgery to remove the cancer, doctors took a sample of his tumour alongside a blood test. Both of these underwent Whole Genome Sequencing.

The results showed that Ian may develop increased side effects and that his chemotherapy dosage would need to be adjusted.

"That test meant that I avoided many of the horrible side effects that come from chemo. I didn't get off scot-free but I count myself as very lucky."

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LYNCH SYNDROME

Lynch Syndrome is a genetic condition that increases the risk of developing cancer, predominantly colorectal and endometrial cancers, to up to 80% during a person's lifetime.

Patients can present with multiple cancers over the years, often at a young age. Although it's the most common form of hereditary cancer, 95% of people don't know they have it.

Early diagnosis of Lynch Syndrome enables targeted cancer screening, including regular colonoscopies through the national bowel screening programme to detect and treat cancers at an early stage, and to implement risk reduction treatments to prevent future cancers.

A Lynch diagnosis also results in cascade testing for family members.

We have worked with the national NHS Cancer Programme and all four Cancer Alliances to establish Lynch testing in cancer pathways across England.

Tumour screening, already taking place in oncology services, has now doubled in the South East, with 90% of colorectal and endometrial tumours now receiving reflex screening.

However, patients are then waiting up to 12 months to see Clinical Genetics to receive a Lynch genetic test. Their families wait even longer. So we are now working with Trusts and Cancer Alliances to develop mainstreamed pathways, transitioning the process of consenting, test ordering and results communication into oncology services.

This year we are funded by your Cancer Alliances to develop models and local business cases to deliver mainstreaming.

"My Mum and my brother hid their symptoms and didn't go to a Doctor until it was too late. Now we have a chance to do it differently. My sister had a preventative hysterectomy and when my niece had symptoms she went straight to the GP. Initially the Doctor told her she was too young to have cancer, but the Lynch diagnosis changed that and doctors took her more seriously. She had womb cancer but luckily they caught it in time."

Alan Baker from South London has Lynch Syndrome

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LYNCH SYNDROME

What are we offering you?

Reflex tumour screening is now in place across the South East, with 90% of endometrial or bowel cancer cases getting screened for Lynch Syndrome – meaning eligible patients can then receive onward genetic testing.

Families can benefit from cascade testing, targeted cancer screening and early diagnosis.

A regional service is in place to support mainstreaming of Lynch genetic testing, including a dedicated regional Lynch Syndrome nurse specialist and a regional Lynch Syndrome MDT.

What do we need from you?

Support us in working with your oncology services, ICBs and Cancer Alliances to develop business cases for sustainable mainstreamed models for Lynch testing in line with national guidance.

Encourage your teams to attend our education & training sessions.

Emma was diagnosed with Lynch Syndrome because her Dad had bowel cancer.

Since then, Emma has had regular colonoscopies to check for early signs of bowel cancer. She's also become more aware of her lifestyle and how to look after herself.

"I feel very grateful that I know I am being regularly monitored. If I didn't know I had Lynch Syndrome, no-one would be keeping an eye on me."

Emma Jenkins from Kent has Lynch Syndrome

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CIRCULATING TUMOUR DNA

The cancer diagnostic process can be difficult and costly.

We are part of a national pilot that uses a simple blood test to detect circulating tumour DNA (ctDNA) to identify and genetically characterise non-small cell lung cancers earlier than with existing standard of care and, in some cases, without the need for invasive biopsies.

The use of ctDNA is expanding rapidly, with the potential to change the landscape for diagnosis, treatment and monitoring of multiple cancer types.

What are we offering you?

The ctDNA pilot for lung cancer is already operating in 15 sites in the South East.

We are welcoming more hospital sites to join the pilot.

Regional genomic tumour advisory board to support results interpretation & treatment recommendations.

What do we need from you?

New pathways need local clinical leadership.

Can you support the development of your cancer CNS workforce to build the skills and capacity needed in their roles to delivery genomic testing and uptake of future cancer genomic opportunities?

David came to hospital feeling breathless. He'd never smoked however, a scan showed a large mass in his right lung which was causing his lung to collapse. He had advanced lung cancer.

Traditionally doctors would have needed to take a section of the tumour for analysis before they could start treatment. However, the ctDNA analysis allowed David's medical team to understand what type of tumour he had and how it would react to treatment. The results came back just over 2 weeks later and David started on a treatment specific to his cancer.

"This is a game changer for our lung cancer patients." Dr Alex Georgiou, Oncologist, Guys and St. Thomas' Hospital.

IMPROVING RARE DISEASE DIAGNOSTIC ODYSSEY

Many people across the South East are waiting for a diagnosis for their symptoms.

Some of them have been waiting years for an explanation so that they can access support and information. This can have a detrimental and costly effect on their downstream treatment and mental health.

Genomic testing may provide an answer. We have improved our turnaround times for rare diseases genetic testing, and we are committed to ensuring health professionals know how to order appropriate genetic testing for their patients.

What are we offering you?

Training, peer support & information to embed genomic testing in local pathways and to make appropriate genomic referrals for eligible patients.

Support to submit genomic referrals including training.

Can we come and talk to your workforce about genomics and why it is important?

What do we need from you?

Work with our Rare Disease Leads, Nursing, Midwifery and Pharmacy teams to identify and train Genomic Champions within your local services who we can work with to facilitate local training and service change.

Support your workforce to take us up our education & training opportunities.

After many years of tests, two of Mel's children were diagnosed with an ultra rare genetic mutation which causes seizures, tremors, learning difficulties and myoclonus.

Although difficult to accept their, genetic diagnosis has given them some answers and opened up new doors for the future.

They have set up a charity called Cure DHDDS to fund research into the condition.

SUPPORTING YOUR WORKFORCE

All healthcare professionals need a genomic education.

We train and educate thousands of NHS staff about all aspects of genomics.

Training opportunities range from light touch and introductory sessions, through to a Masters in Genomic Medicine or specialty tailored pathway training.

Our team offers a number of ways for people to get involved including online materials and courses, monthly virtual Lunch & Learn sessions, regular in person Grand Rounds, and bespoke study days.

Check our website for the latest opportunities.

What are we offering you?

Educational interventions that meet the needs of a range of disciplines including pharmacy.

Signposting to educational resources.

Dedicated genomic specialist nurses to support your nurses & midwives to embrace genomics.

Our new Charter of Excellence offers a framework to guide Nursing & Midwifery leaders to embed genomics into everyday practice.

What do we need from you?

Identify Genomic Champions.

Release staff time for genomic training.

Champion the uptake of the new Charter of Excellence in Nursing & Midwifery.

Share our communications about local genomic successes and patient stories with your staff.

Encourage your staff to participate in our professional networks in nursing, midwifery and pharmacy.

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NHS South East Genomic Medicine Service

OUR 2024/25 PRIORITIES

Our work spans almost every NHS speciality. Here are just a few of our priorities.

Reducing our laboratory turnaround times

Implement the Cancer Genomic Improvement Programme to deliver faster end to end turn around times

Establishing regional Cellular Pathology Genomic Centres

Creating three new national Genomic Networks of Excellence

Rapid pathogen genome sequencing for patients with suspected respiratory infections

Improving identification & outcomes for people with inherited or acquired cardiovascular disease

Genomics & Artificial Intelligence

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REDUCING OUR LABORATORY TURNAROUND TIMES

We have seen a 22% increase in genomic testing.. This is testament to the important transformation role that genomics now plays within healthcare.

We continue to prioritise urgent tests, but the increased demand had an impact on our waiting times for genomic test results, meaning that some patients waited longer than we would like.

To reduce waiting times, we put in place a range of improvements including investing in more staff, buying new equipment and examining every step of our process to find new efficiencies and seek further improvements including our intention to achieve fully automated wet lab process.

As a result, we have cleared around 90% of our backlog for non-whole genome sequencing. We've seen significant improvement in our turn around times. For example microarray testing is now delivered in 8-9 weeks compared to 40-50 weeks.

However, there are some tests such as whole genome sequencing for rare disease that are exceeding the national target. We are working hard to address these and we expect to clear our WGS backlog over the next four months.

A significant challenge of the Whole Genome Sequencing pathway is the appropriateness and completeness of referrals. We are continuing to work with our colleagues to improve pathways and ensure they are appropriately resourced to meet the increasing demand. Referrals with sufficient phenotype information (HPO terms) and the correct number of familial samples (e.g. proband and both parents; trios) are more efficient for the laboratory to process.

Our waiting times are regularly updated on our website.

If your patient is waiting for testing, but their situation changes or their treatment plan is affected, please get in touch with us to discuss on gst-tr.southeastglh@nhs.net

REDUCING OUR LABORATORY TURNAROUND TIMES

What are we offering you?

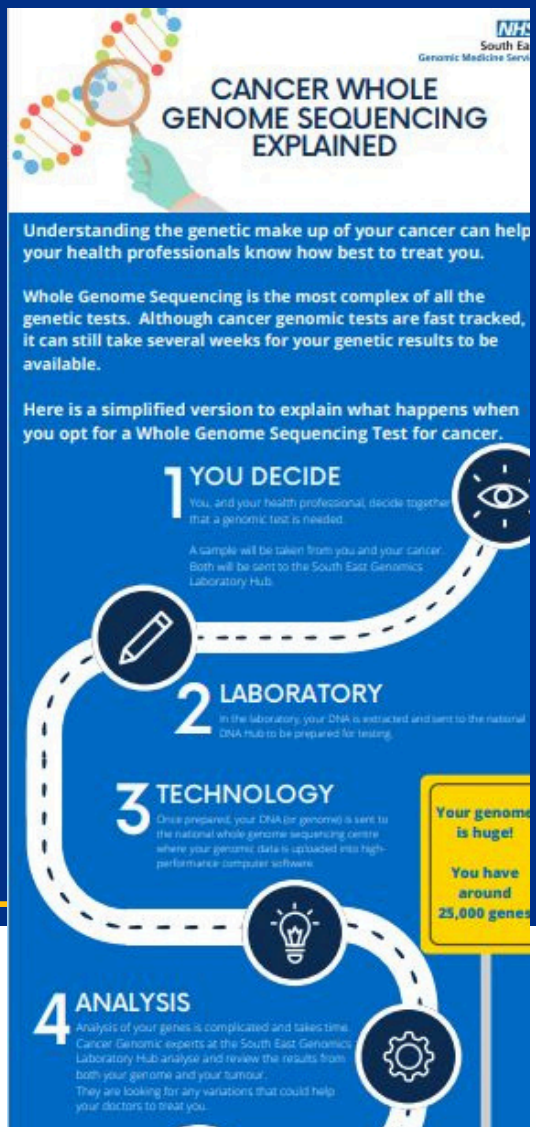
Support & information to make accurate and complete WGS referrals.

Information & training for clinicians and patients about the genomic test process.

What do we need from you?

Opportunities to educate & support your workforce.

Can you provide us with a Genomic Champion to work with us?



ESTABLISHING REGIONAL CELLULAR PATHOLOGY GENOMIC CENTRES

We are excited to announce the launch of our NEW Cellular Pathology Genomic Centres.

These new centres will reduce the time it takes to complete solid cancer tumour testing referrals and play a pivotal role in preparing samples for the NHS Cancer Vaccine Launchpad Programme, which is enabling patients to access cancer vaccine trials.

In the South East, we are establishing three new Cellular Pathology Genomic Centres:

- Guys Hospital serving South London & Sussex
- Maidstone & Tunbridge Wells Hospital serving Kent
- Berkshire Surrey Pathology Services serving Surrey & Sussex

What are we offering you?

A new centrally funded histopathology service to prepare solid cancer tissue samples for onward submission our laboratories, saving you time and money.

What do we need from you?

Opportunities to educate and support your workforce about the new service.

Can you provide us histopathology leads so that we can work together to improve the clinical pathway?

ESTABLISHING THREE NEW GENOMIC NETWORKS OF EXCELLENCE

In 2023/24 NHS England commissioned eight new NHS Genomic Networks of Excellence to develop the evidence and model of adoption for cutting edge genomic advances and technology applications that will be transformative for patients.

We are leading with our partners on three of these Networks, including delivery of several innovative programmes of work.

Respiratory metagenomics : rapid pathogen genome sequencing for patients with suspected respiratory infections

Currently patients and clinicians in intensive care wait several days to understand what is causing their infection using traditional viral or bacterial culture techniques.

We are working with NHS and academic partners, UKHSA, and the Office of Life Sciences to expand a successful pilot at Guy's and St Thomas's which offers rapid, pathogen agnostic genetic testing for intensive care patients with suspected respiratory infection.

This metagenomic testing can determine the cause of their condition and potential antibiotic resistance within seven hours, improving accurate and timely treatment for patients and providing useful public health information. The pathogen genomic data has dual benefits for national biosecurity and will be shared with UKHSA to contribute to their pilot of a national pathogen surveillance system.

This new project will expand the pilot to a further 10 intensive care units across the country by 2025.

NEW GENOMIC NETWORKS OF EXCELLENCE

Improving the identification and outcomes for people with inherited or acquired cardiovascular disease

25% of all deaths in the UK are attributed to cardiovascular heart disease making it the leading cause of death worldwide.

Early detection of people who have a high genetic risk of cardiovascular disease would prevent many premature deaths including sudden cardiac death which happens more often in young people and children.

We are working on a new project to identify people who are at risk of both inherited and acquired cardiovascular disease using genomic testing in combination with detailed clinical assessment. Once identified, these people can benefit from lifesaving treatment. We will pilot a range of approaches to understand how best to achieve this aim.

We will also be supporting the roll out of the National Sudden Cardiac Death pilot to additional sites across the region with continued support from the British Heart Foundation and NHSE.

Genomics & Artificial Intelligence

Artificial Intelligence combined with genomics, offers exciting opportunities to speed up diagnoses, further personalise treatments and release valuable staff time.

The Genomics AI Network of Excellence has been established to support the deployment of AI in genomic medicine to benefit patients. Over the next two years we will be building a national community focused on the use of AI in genomics identifying and supporting the conditions necessary to adopt AI in genomic medicine for the benefit of patients, and delivering some exemplar programmes which aim to better identify individuals with genomic conditions. We will also scope public attitudes to the implementation of AI in genomics to support efforts in building patient and public trust.

NEW GENOMIC NETWORKS OF EXCELLENCE

There are five further Genomic Networks of Excellence across the country which we are also involved in including:

Pharmacogenomics & Medicine Optimisation

Almost half of all UK adults regularly take prescription medicines and the NHS's annual budget for medicines is approximately £17.4 billion per year, with over 1.1 billion items prescribed annually.

One approach to addressing adverse or ineffective medication reactions is to leverage knowledge of an individual's genetic information to support medicines optimisation, better informing medicine selection and dosing, a concept known as pharmacogenetics.

This NHS Genomic Network of Excellence will develop the rollout of pharmacogenomics and medicines optimisation in the NHS, including furthering the rollout in primary care.

This Network, which is being led by the North West Genomic Medicine Service, will also explore how best to address the recent NICE guidance which recommends CYP2C19 genotype testing to guide the use of clopidogrel in patients who have had an ischaemic stroke or transient attack.

Haemato-Oncology

to deliver new technologies for patients with haematological malignancies.

Prenatal Genomic Medicine

to drive non-invasive prenatal genomic testing.

Circulating Tumour Biomarker testing

explore expansion of biomarker testing into a range of tumour types

Rare & Inherited diseases

will reduce genomic health inequalities & develop new testing approaches for people with a suspected rare disease.

Thank you for taking the time to read our report.

We hope you have a better understanding of how we can support one another in our shared goals.

Please share this report with your colleagues at all levels.

Get in touch and let's talk in more detail.
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