# Keeping pace with cancer:

## Accelerating access to genomic testing through the NHS Genomic Medicine Service

FUTURE HEALTH

This independent report was sponsored by Johnson & Johnson and authored by Future Health. The views and conclusion in the report are those of Future Health and should be attributed as such. Future Health takes full responsibility for the content of the report and associated publications.

# CONTENTS

# ABOUT THE AUTHOR

Richard Sloggett is the Founder and Programme Director of Future Health. He was previously a Senior Fellow at Policy Exchange and from 2018-19 was Special Advisor to the Secretary of State for Health and Social Care. Richard is a regular commentator in the national media on health and social care and has been named as one of the top 100 people in UK healthcare policy by the Health Service Journal. During his time with the Secretary of State, Richard worked across Whitehall, the NHS and local government on major policy decisions including the NHS Long Term Plan and the Prevention Green Paper. Alongside his work at Future Health Richard is undertaking his doctoral thesis in preventative healthcare systems at the University of Liverpool.

# ABOUT FUTURE HEALTH AND THIS REPORT

Future Health is a public policy research centre focused on creating healthier, wealthier people, communities and nations. Future Health publishes regular research papers across its three policy research programmes of health prevention, health technology and the links between improvements in health and economic growth.

This work is part of the health technology programme.

Future Health completed a series of interviews with national policymakers and local system leaders to inform the research. These were conducted between November 2023 and May 2024.

Data analysis was undertaken in January-February 2024 and covered Q2 23-24 data from the NHS Genomic Medicine Service available here.

# GLOSSARY OF ORGANISATIONS/TERMS[1]

### NHS Genomic Medicine Service

The NHS Genomic Medicine Service aims to harness the power of genomic technology and science to improve the health of the population and deliver on the commitments in the NHS Long Term Plan to be the first national health care system to offer whole genome sequencing as part of routine care.

### The National Genomic Test Directory

The National Genomic Test Directory (Test Directory) outlines the range of genomic tests that are commissioned by NHS England for patients in England. The Test Directory covers the full repertoire of testing from targeted testing, to panel next generation sequencing and up to the level of whole genome sequencing. NHS England, supported by a genomics clinical reference group and test evaluation working groups, reviews the Test Directory on an annual basis.

### NHS Genomic Laboratory Hubs (GLHs)

The NHS GLHs are seven consolidated laboratory networks with defined geographies that operate as part of a national genomic laboratory network, commissioned by NHS England to deliver the genomic testing outlined in the National Genomic Test Directory. This includes the delivery of specialist rare and inherited disease, cancer and pharmacogenomic testing.

### NHS GMS Alliances

Seven NHS GMS alliances were established in December 2020. An NHS GMS Alliance is a collective made up of a small number of key NHS providers with a recognised track record and expertise in genomics working in partnerships to support the strategic systematic embedding of genomic medicine in end-to-end clinical pathways and clinical specialities for a given population. They drive this embedding across all providers within their geography from primary and community care to secondary and tertiary care.

### Clinical Genomics Service

NHS England commissions seventeen NHS clinical genomic services (NHS CGSs), including genetic medical consultants, specialist trainees and genetic counsellors. They deliver a clinical genomic and counselling service that directs the diagnosis, risk assessment and lifelong clinical management of patients of all ages and their families who have, or are at risk of having, a rare genetic or genomic condition, including, inherited cancer.

---

[1] Adapted from NHS England. *Accelerating Genomic Medicine in the NHS*. 2022. Available at: https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/#appendix-1-overview-of-the-nhs-gms

### Genomic Networks of Excellence

Eight innovative networks to develop the evidence and model of adoption for cutting edge genomic advances and technology applications that will be transformative for patients.

### Genomics Clinical Reference Group (CRG)

The Genomics CRG has been convened to support implementation of the GMS. Through its professional, patient and public representation, the Genomics CRG carries out the following functions:

- Advise on clinical policy and strategy for genomics, including implementation of NHS Long Term Plan commitments and future development of the NHS GMS
- Oversee a clear and transparent process for annual review of the National Genomic Test Directory (supported by three test evaluation working groups covering rare and inherited disease, cancer and pharmacogenomics)
- Support activities to raise awareness and embedding of genomics across all clinical specialties
- Support driving improvements in personalised medicine
- Advise on, review and develop guidance and service specifications (including the clinical genetics service specification)

### Genomics England

In a number of areas, the NHS works with Genomics England – a company established and wholly owned by the Department of Social Care in 2013 to deliver the 100,000 Genomes Project. Genomics England provide key services to NHS England and to the NHS GMS, managed through a Master Service Agreement that underpins the delivery of whole genome sequencing in the NHS.

### Health Education England Genomics Education Programme

Health Education England and specifically the Genomics Education Programme support the upskilling of the multi-professional workforce in genomics. This includes through the development of resources, educational materials and best practice sharing to increase education and training. Health Education England is now part of NHS England.

### Community Diagnostic Centres

Offer patients a wide range of diagnostic tests closer to home and greater choice on where and how they are treated, reducing the need for hospital visits and helping them to receive potentially life-saving care sooner.

### Cancer Alliances

Cancer Alliances bring together clinical and managerial leaders from different hospital trusts and other health and social care organisations, to transform the diagnosis, treatment and care for cancer patients in their local area.

# EXECUTIVE SUMMARY

Speaking at the Conservative Party Conference in October 2018 the Secretary of State for Health and Social Care Matt Hancock announced that the NHS Genomic Medicine Service would start rolling out access to genomic testing for those with rare cancers and seriously ill children.

The Secretary of State noted that the UK was 'leading the world, and I'm incredibly excited about this technology because of its potential to change lives for the better.'[2] In the years since, the Government and NHS England have set out a series of national plans and strategies to increase the patients eligible to benefit from such testing, including cancer patients.

Nearly six years on there are positive signs that the number of genomic tests for cancer that are available and those being undertaken is increasing. For solid tumour cancers the number of tests on the National Genomic Test Directory has increased from 111 to 140 since 2021.[3] Based on recent growth our analysis finds that the system can be expected to deliver 400,000 solid tumour cancer tests a year by the end of 2025.

The NHS has completed the first year of its new genomic medicine strategy, *Accelerating Genomic Medicine in the NHS*, and has put in place measures to address key enablers for change such as patient and public engagement, workforce training, regional networks of excellence and new governance structures to bring together the constituent parts of regional services.

However in making these strides, there are signs of a service, like the wider system it is in, under pressure. It is difficult to gauge an accurate picture of how such pressures are playing out both nationally and regionally given the limited data on the system currently published. Regional data on turnaround times for tests where available, anecdotal stories and media reports have though all highlighted some difficulties.

Part of this pressure is based on increasing innovation and the complexity of testing. The expertise of the Genomic Laboratory Hubs (GLHs) – where a significant amount of testing takes place – was primarily in rare diseases rather than in cancer and as innovation in genomic testing for cancer has expanded across single-gene, panel tests and whole genome testing the capability and capacity of the system has been stretched. More and more tests are being added to the National Genomic Test Directory, including updates 'in year', with local services not always equipped to deploy them; and there are variations in clinical pathways in when tests are being ordered. Workforce, data infrastructure and capital challenges are also checking rates of progress.

[2] https://www.ukpol.co.uk/matt-hancock-2018-speech-to-conservative-party-conference/

[3] https://www.england.nhs.uk/publication/national-genomic-test-directories/

Such challenges comes with a clinical and patient cost. Delays to genomic testing for cancer, result in delays to diagnosis which in turn lead to poorer outcomes.

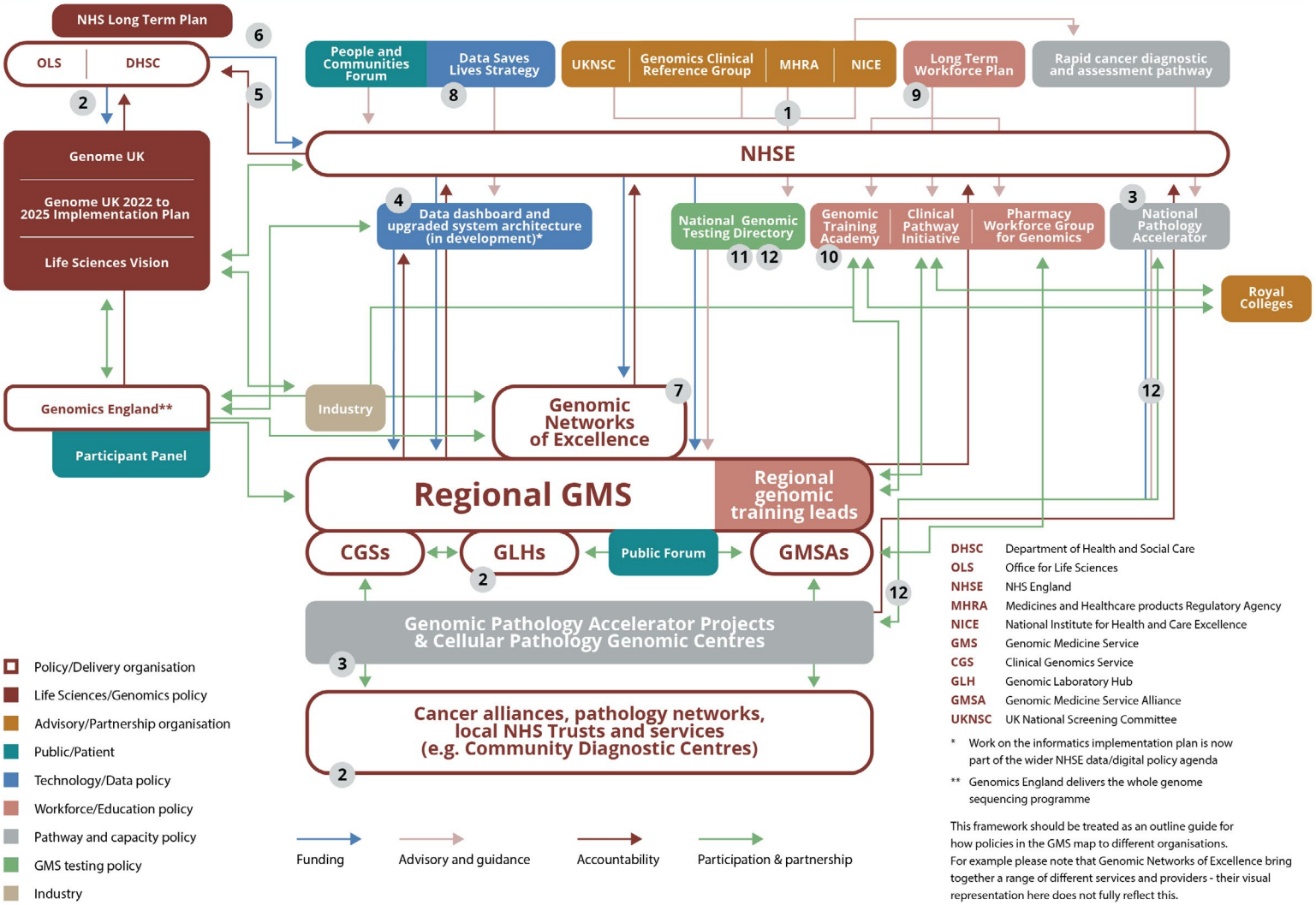
The next Government will face a plethora of challenges across health and social care but improving cancer services will inevitably be near the top of the in-tray. Increasing the capacity and the faster delivery of cancer testing to support improvements in diagnosis and treatment needs to be an immediate system priority.

This report argues for more investment in the NHS Genomic Medicine Service as part of a wider health system shift to prevention. Capacity can be increased by further scaling up GLHs and through better connecting the Genomic Medicine Service with local pathology services, and reforming contracting arrangements to enable greater flexibility and partnership on where testing is delivered. Building the clinical science workforce and new workforce models – supported by new technologies – can help increase capacity and improve turnaround times. More iterative and agile updates to clinical guidelines can help embed good practice on when genomic testing is used in cancer patient pathways and move to a 'genomics-first' approach to cancer testing. Better data on how the system is performing can improve transparency and drive investment and upgrades.

The science of genomic testing for cancer is moving quickly and public need is rising – the number of UK cancer cases is set to reach half a million by 2040 and UK cancer outcomes continue to lag behind other comparable countries. The work of the NHS Genomic Medicine Service is critical in helping us in keeping pace with cancer, saving lives and supporting a much needed shift to prevention; but to do so will now require concerted investment and prioritisation.

# SUMMARY OF NHS GENOMIC MEDICINE SERVICE POLICY FRAMEWORK AND RECOMMENDATIONS FOR ACTION

## Structure of NHS Genomic Medicine Service policy framework



# SUMMARY OF NHS GENOMIC MEDICINE SERVICE POLICY FRAMEWORK AND RECOMMENDATIONS FOR ACTION

## Recommendations by organisation

| | DHSC | NHSE | Regional GMSs |
|---|---|---|---|
| **Accountability and performance** | With OLS publish an annual report with metrics on progress in implementing the Genome UK Strategy **(5)** | Publish monthly performance dashboard for the Genomic Medicine Service **(4)** | Provide data to support the development of the Genomic Medicine Service dashboard **(4)** |
| **Capacity** | Ensure the Genomic Medicine Service is resourced to deliver the necessary increase in cancer testing capacity required **(2,6)** | Explore new contracting and funding arrange-ments to maximise cancer testing capacity between GLHs and other local services **(2)** | Be enabled to maximise local testing capacity outside of GLHs to improve turnaround times for cancer testing **(2)** |
| **Pathways** | Evaluate the uptake and impact of the GeNotes course **(11)** | Publish an update on Genomic Pathology Accelerators and Cellular Pathway Genomic Centres by the end of 2024 **(3)**. Ask each GLH to report back on where a small sample of genomic cancer tests from the Test Directory are being used in pathways to identify variation. Use such data to support moves to deliver a 'genomics first' approach to cancer testing **(12)** Publish data on the uptake and feedback on the GeNotes course **(11)** | |
| **Horizon scanning** | | Complete and publish an annual horizon scanning process (with other agencies e.g. NICE and MHRA) for new genomic tests and set out a clear process for updating the Genomic Test Directory 'in year' **(1)** | |
| **Workforce** | Continue to invest in the healthcare science workforce through the NHS Long Term Workforce Plan and ensure that the Genomic Medicine Service has the staff needed to deliver for patients **(9)** | Regularly review workforce models to ensure they utilise the full capabilities of the workforce including biomedical scientists. **(9)** Publish the numbers participating in and feedback from participants in genomic training programmes **(10)** | |
| **Data** | Use NHS Productivity Plan to upgrade the IT and data capability of the Genomic Medicine Service. Fund and scale evidenced based solutions developed through the Genomic Networks of Excellence **(6)** | Invest in the Genomic Medicine Service dashboard **(4).** Provide quarterly updates on progress in implementing the Data Saves Lives strategy **(8)** | Collect data on the economic impact of new technological innovations in the Genomic Medicine Service, through Genomic Networks of Excellence **(7)** |

# BACKGROUND – THE INCREASING IMPORTANCE OF GENOMICS WITHIN UK HEALTH POLICYMAKING

Genomics and advances in molecular biology have accelerated in the last twenty years particularly following the work of the Human Genome Project.[4] The Project which ended in 2003 led to concerted action internationally across Governments, health systems, clinicians and scientists on how such understanding and the innovation it led to could be successfully adopted to improve human health.

The late 1990s and early 2000s saw an associated rise in importance in genomics within UK health policymaking. In December 1999 the Human Genetics Commission was established, which aimed to provide strategic advice to Government on developments in human genetics.[5] In April 2001 Secretary of State for Health Alan Milburn announced plans to create new Genetic Knowledge Parks across the UK to be centres of clinical and scientific excellence.[6]

In a subsequent speech in January 2002, Milburn highlighted how the NHS was particularly well placed to benefit from advances in genomics: 'I believe there is no other health care system in the world better placed to harness the potential of genetic advances than the National Health Service. The values on which the NHS is based – providing care for all on the basis of need, not ability to pay – are uniquely suited to capturing the benefits of the genetics revolution.'[7]

To coincide with the end of the Human Genome Project the Government published a White Paper *Our Inheritance, Our Future*. In the Foreword, the Prime Minister set out his determination that 'the National Health Service should be able to respond to these advances so the benefits of genetics and the more personalised and improved healthcare it will bring are available to all.'[8]

The Government's three year review of progress with the White Paper pointed to a series of examples where progress had been made including rolling out new born screening programmes for sickle cell disease and cystic fibrosis.[9]

However a 2009 House of Lords Committee report argued that the 2003 White Paper's model – focused on rare single-gene disorders under the care of clinical geneticists in regional centres – had come before further technological advances in screening[10] and greater understanding of the genetic causes of major diseases.[11][12]

---

4 https://www.phgfoundation.org/media/137/download/Public%20health%20in%20an%20era%20of%20genome-based%20and%20personalised%20medicine.pdf?v=1&inline=1

5 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1127190/

6 https://www.theguardian.com/science/2001/apr/19/genetics.politics

7 https://www.theguardian.com/science/2001/apr/19/genetics.politics

8 https://www.nuffieldtrust.org.uk/sites/default/files/2019-11/genetics.pdf

9 https://www.wired-gov.net/wg/wg-news-1.nsf/0/B84ABCE02FD523188025742D0031C804?OpenDocument

10 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7717495/

11 https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10703.htm

12 https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10703.htm

The Committee called for a new White Paper to facilitate the introduction of advances in genomic science into clinical practice.[13] They also recommended that the Government set out clear actions for preparing commissioners and providers for using genetic tests.[14] The 2006 Cooksey Review into UK health research funding had identified 'translational research as an area of weakness and warned that the UK was at risk of failing to reap the full economic, health and social benefits that public investment in health research should generate. '[15] Two key gaps were identified: first, the translation of ideas from basic or clinical research into development of new products and new approaches to treatment of disease and illness; and, second, the use of those new products and approaches in clinical practice.'[16]

In response to the Lords Committee, the Government set up the Human Genomic Strategy Group.[17] The Group produced a report in January 2012 calling for a White Paper which 'sets out overarching policy direction on genomic technology adoption in the NHS.'[18] The report also recommended that the NHS Commissioning Board (which was to then become NHS England) should take a lead in commissioning genomic services in the NHS, including:

- Bringing forward proposals for the establishment of a strategic network to deliver expert advice on the strategic development of genomic and genetic services
- Developing national tariffs for genetics and special pathology tests, and ensuring that the cost of genetics diagnostics is included in the clinical specialty pathway
- Developing, in collaboration with commissioners, the UK Genetic Testing Network and NICE, a robust process for the evaluation of clinical validity and utility of all genetic and genomic tests and markers and setting minimum national quality standards
- Ensuring that NICE Diagnostics assess the validity, utility and quality of all new molecular tests, e.g. for cancer, with input from all relevant specialties including pathology
- Putting in place agreements that require data from tests carried out by NHS-commissioned laboratories – in the NHS or private sector – to be made available to nationally designed research databases within a framework that ensures patient confidentiality and data protection[19]

The Group also recommended that the Department of Health and the NHS Commissioning Board (NHS England) should work together to set-up a service delivery model for genetic and genomic testing 'with the objective of putting in place a network consisting of Genomic Technology Centres, Biomedical Diagnostic Hubs and Regional Genetics Centres.'[20]

---

13 https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10711.htm
14 https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10711.htm
15 https://assets.publishing.service.gov.uk/media/5a7c49bc40f0b6321db382e2/0118404881.pdf
16 https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10706.htm
17 https://assets.publishing.service.gov.uk/media/5a7c38c640f0b67d0b11faec/dh_132382.pdf
18 https://assets.publishing.service.gov.uk/media/5a7c38c640f0b67d0b11faec/dh_132382.pdf
19 https://assets.publishing.service.gov.uk/media/5a7c38c640f0b67d0b11faec/dh_132382.pdf
20 https://assets.publishing.service.gov.uk/media/5a7c38c640f0b67d0b11faec/dh_132382.pdf

In December 2012 Prime Minister David Cameron launched the 100,000 genomes project, aiming to sequence 100,000 genomes from NHS patients over the next five years.[21]

In July the following year Genomics England was established as a wholly owned limited company to deliver the project. The organisation had five main aims:

- To benefit patients by providing clinical diagnosis and, in time, new or more effective treatments;
- To provide new scientific insights and discovery;
- To accelerate the uptake of genomic medicine in the NHS;
- To stimulate and enhance UK industry and investment; and
- To increase public knowledge and support for genomic medicine[22]

The 2017 *Life Sciences Industrial Strategy* highlighted UK BioBank and Genomics England as large scale infrastructure projects that had put the UK in a leading position on life sciences globally. The strategy called for continued support for genomics in medicine including advancing proposals by the Chief Medical Officer (CMO) for increasing access to genomic testing and screening.[23]

This referred to the important 2016 annual report from then CMO Dame Sally Davies which was dedicated to genomics and called for wider access to genomic tests in the NHS.[24] In her report Davies argued that 'as with other specialties, genomic services developed as 'cottage industries' built on regional expert presence and local interests and funding' and that it was time now to move on from this way of doing things and 'build a national, first class service that is scalable, futureproof, and delivers value for money.'[25]

21 https://publications.parliament.uk/pa/cm201719/cmselect/cmsctech/349/34906.htm

22 https://publications.parliament.uk/pa/cm201719/cmselect/cmsctech/349/34906.htm#footnote-293

23 https://assets.publishing.service.gov.uk/media/5a74aeaced915d7ab83b5b0b/LifeSciencesIndustrialStrategy_acc2.pdf

24 https://assets.publishing.service.gov.uk/media/5a82b85fe5274a2e87dc2a4a/CMO_annual_report_generation_genome.pdf

25 https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/631043/CMO_annual_report_generation_genome.pdf


# INTRODUCTION – BUILDING THE NHS GENOMIC MEDICINE SERVICE

The NHS proposals for increasing access to genomic testing through the creation of an NHS Genomic Medicine Service were published in March 2017.

The NHS England Board paper noted that the NHS had been actively involved since the start of the 100,000 Genome project through establishing 13 NHS Genomic Medicine Centres (GMCs) working across geographical areas of approximately 3 to 5 million people and involving over 85 hospital trusts.[26] However the paper highlighted the scale of challenge in mainstreaming genomic testing into the system stating that 'significant transformational change is required across the NHS to embed genomic medicine into routine care. From the way consent is taken, processes for collecting and handling samples, the role of clinical genetics within clinical specialities, to the informatics infrastructure enabling data to be brought together, shared and analysed.'[27]

Successful implementation of the service would according to the paper include: a new networked genetic laboratory and genomic medicine service infrastructure, a comprehensive genomic testing strategy for the NHS, a joint-commissioning model with clinical commissioning groups (CCGs) to achieve standardisation, equity of access, economies of scale and the high quality testing required for the future and require a further evolution of the NHS informatics environment, particularly through capital investment.[28]

An April 2018 report from the House of Commons Science and Technology Committee raised concerns about the progress of digital and data infrastructure and medical training to support the effective delivery of the NHS Genomic Medicine Service.[29] In its evidence gathering the Committee found support for the creation of regional hubs to deliver the tests in the National Genomic Test Directory but differences in opinion over the optimal number of sites and their integration with other services and design. For example, the Royal College of Pathologists suggested that the GLHs should be co-ordinated with NHS Improvement's plans to establish a consolidated network of 29 pathology hubs, emphasising the expertise required to prepare tumour samples for genome sequencing.[30]

In October 2018 Secretary of State for Health and Social Care Matt Hanock launched the NHS Genomic Medicine Service. The announcement included the expansion of the 100,000 Genomes Project to 1 million genomes sequenced by the NHS and UK Biobank over five years. The announcement formed part of a wider ambition to sequence five million genomes by bringing together world-leading industry experts such as UK Research and Innovation, the NHS and other partners.[31] The subsequent 2019 NHS Long Term Plan committed to 'extend the use of molecular diagnostics and, over the next ten years.... routinely offer genomic testing to all people with cancer for whom it would be of clinical benefit, and expand participation in research.'[32]

---

26 https://www.england.nhs.uk/wp-content/uploads/2017/03/board-paper-300317-item-6.pdf

27 https://www.england.nhs.uk/wp-content/uploads/2017/03/board-paper-300317-item-6.pdf

28 https://www.england.nhs.uk/wp-content/uploads/2017/03/board-paper-300317-item-6.pdf

29 https://publications.parliament.uk/pa/cm201719/cmselect/cmsctech/349/349.pdf

30 https://publications.parliament.uk/pa/cm201719/cmselect/cmsctech/349/349.pdf

31 https://www.gov.uk/government/news/matt-hancock-announces-ambition-to-map-5-million-genomes

32 https://www.longtermplan.nhs.uk/wp-content/uploads/2019/08/nhs-long-term-plan-version-1.2.pdf

This was followed by a Government genomics strategy, *Genome UK,* published in 2020 which set out the main elements of the new Genomic Medicine Service. This included:

- The development of a network of National GLHs to deliver an integrated genomic testing system supported by a National Genomics Informatics Service
- A comprehensive National Genomic Test Directory of genomic tests for specified cancers and rare diseases that encompasses the entire testing repertoire from WGS to single gene tests
- Work by NHS England and NHS Improvement to further transform pathways of care and create the multi-disciplinary teams and cross-professional infrastructure that will be critical for the future
- A new NHS Genomic Medicine Service Research Collaborative led by NHS England and NHS Improvement, the NIHR, and Genomics England will establish a coordinated approach to embedding research and discovery to advance clinical care ensuring patients gain maximum benefit from research and innovation, with breakthroughs enabling prevention of ill-health, earlier diagnosis, more effective treatments, better outcomes and faster recovery

This was aligned with the 2021 *Life Sciences Vision* which called for unlocking the power of genomics at scale across the UK to help 'create the most advanced and integrated genomic research healthcare ecosystem in the world, underpinned by the latest science and technology, in order to drive better health outcomes, early detection, diagnosis and treatment of disease, and innovative research.'[33] Professor Sir Mike Richards' review of diagnostics had highlighted the challenges of getting new diagnostics, including genomic testing, into the NHS and had called for greater co-ordination between national bodies and agencies through the NHS Accelerated Access Collaborative to realise this.[34]

A three year implementation plan for the genomics strategy was published later in 2022 which included a series of investments including: funds to tackle health inequalities in genomic medicine; money for a new innovative cancer programme to evaluate cutting-edge genomic sequencing technology; the use artificial intelligence to analyse genomic data alongside digital histopathology and radiology images.[35]

In October 2022 NHS England published their strategy for embedding genomic testing within the NHS over the next five years. The strategy had four objectives:

- Embedding genomics across the NHS, through a world leading innovative service model from primary and community care through to specialist and tertiary care

---

33 https://assets.publishing.service.gov.uk/media/612763b4e90e0705437230c3/life-sciences-vision-2021.pdf

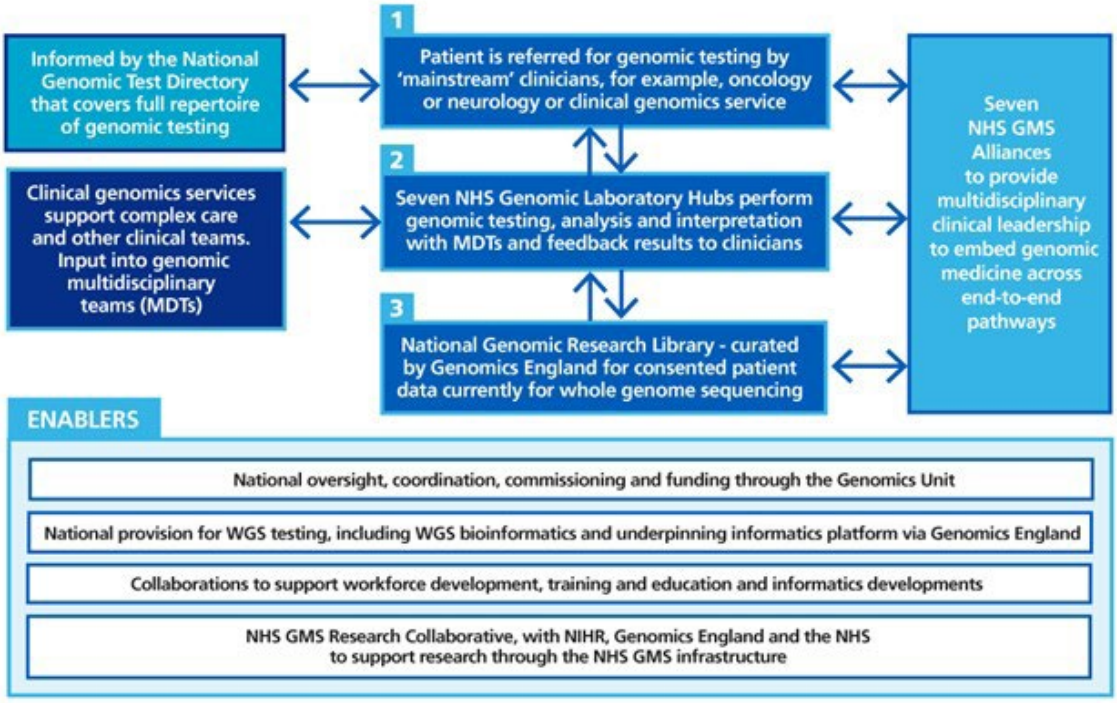
34 https://www.england.nhs.uk/wp-content/uploads/2020/11/diagnostics-recovery-and-renewal-independent-review-of-diagnostic-services-for-nhs-england-2.pdf

35 https://www.gov.uk/government/publications/genome-uk-2022-to-2025-implementation-plan-for-england/genome-uk-2022-to-2025-implementation-plan-for-england

- Delivering equitable genomic testing for improved outcomes in cancer, rare, inherited and common diseases and in enabling precision medicine and reducing adverse drug reactions
- Enabling genomics to be at the forefront of the data and digital revolution, ensuring genomic data can be interpreted and informed by other diagnostic and clinical data
- Evolving the service through cutting-edge science, research and innovation to ensure that patients can benefit from rapid implementation of advances[36]

The design of the NHS Genomic Medicine Service was set out in the strategy, see figure 1 below.

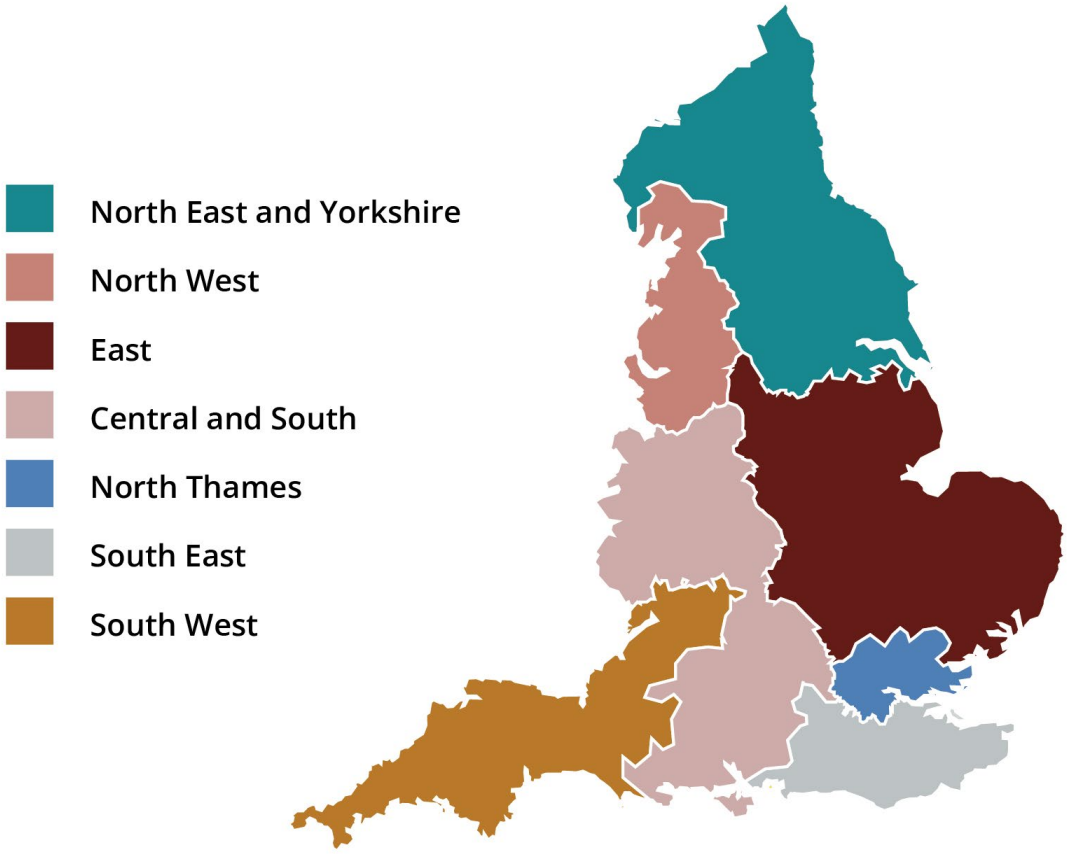
*Figure 1: The structure of the NHS Genomic Medicine Service*[37]




The seven GLHs are regionally located as set out in figure 2 on the next page.

[36] https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/
[37] https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

*Figure 2: The NHS Genomic Laboratory Hubs*[38]




The size of patient populations varies between GLHs. The largest is the central and south GLH which covers 10 million people, 45 NHS Trusts and 14 Integrated Care Systems (Integrated Care Systems). The smallest is the South West GLH which covers a population of 4 million, 18 NHS Trusts and 7 ICSs.

[38] https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

*Figure 3: NHS Genomic Medicine Service: Geographies and populations*[39]

The NHS Genomic Medicine Service: Geographies

| Geography | Patient population | NHS trusts | NHS ICSs |
|---|---|---|---|
| North East and Yorkshire | 8 million | 34 NHS trust | 4 ICSs |
| North West | 7 million | 34 NHS trusts | 4 ICSs |
| Central and South | 10 million | 45 NHS trusts | 14 ICSs |
| East | 8 million | 32 NHS trusts | 13 ICSs |
| North Thames | 7 million | 36 NHS trusts | 11 ICSs |
| South East | 8 million | 29 NHS trusts | 8 ICSs |
| South West | 4 million | 18 NHS trusts | 7 ICSs |

[39] https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

# STATE OF PLAY - THE NHS GENOMIC MEDICINE SERVICE AND GENOMIC CANCER TESTING

The number of new cancer cases in the UK is set to reach half a million by 2040.[40] Despite some progress, UK cancer survival rates continue to lag behind other countries including Australia, Canada and Norway.[41]

The 2020 Richards' Review into diagnostics noted the need to expand genomic testing for cancer to meet the NHS *Long Term Plan* commitment to diagnose 75% of people with cancer at stage one or two by 2028.[42] As science advances and our understanding of the disease increases, genomic testing presents an opportunity to make new progress in the earlier diagnosis and improved and more personalised treatment of cancer. The different forms of genomic testing for cancer are summarised in figure 4 below.

*Figure 4: Summary of different types of genomic tests for cancer*[43]


40 https://news.cancerresearchuk.org/2023/11/28/cancer-research-uk-launches-ambitious-manifesto/

41 https://www.ucl.ac.uk/news/2024/feb/uk-cancer-treatment-falls-behind-other-countries#:~:text=Lower%20use%20of%20chemotherapy%20and,15%20years%20behind%20leading%20countries

42 https://www.ucl.ac.uk/news/2024/feb/uk-cancer-treatment-falls-behind-other-countries#:~:text=Lower%20use%20of%20chemotherapy%20and,15%20years%20behind%20leading%20countries

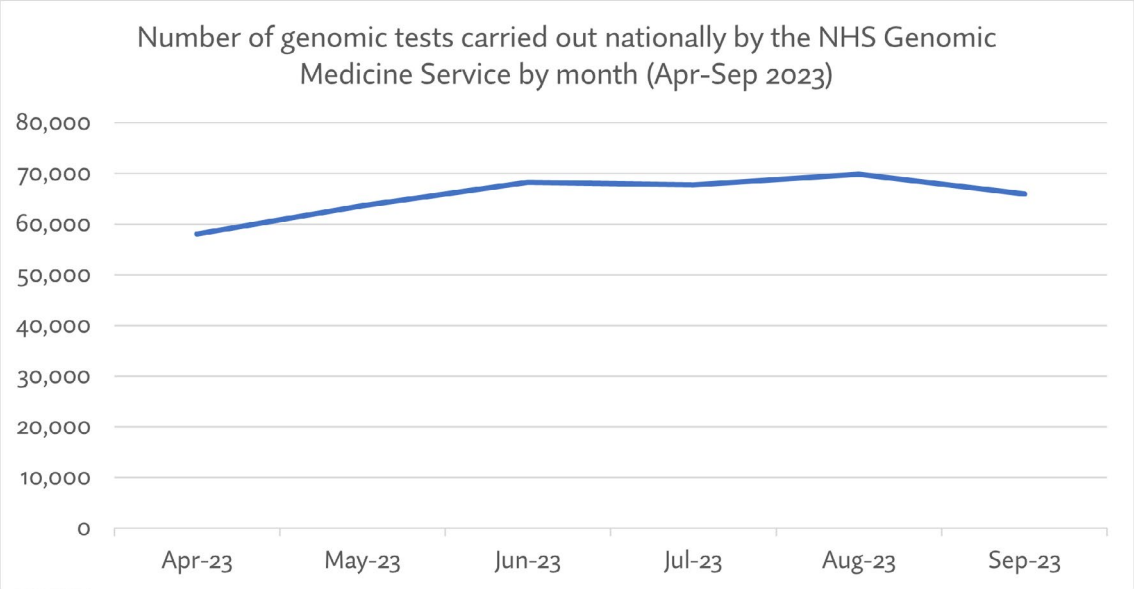
43 https://www.genomicseducation.hee.nhs.uk/genotes/knowledge-hub/different-approaches-to-gene-sequencing/

Ensuring clinicians can access the latest approved genomic tests rapidly will form an increasingly important part of cancer care. As it approaches its six year anniversary the NHS Genomic Medicine Service is central to realising these ambitions for improved cancer patient outcomes.

In April 2023 media reports highlighted long delays for patients waiting for certain genome sequencing results through the Genomic Medicine Service. Long waiting lists, outdated infrastructure and shortages of scientists and clinical geneticists were all cited as reasons for these delays.[44][45]

In December 2023 NHS England published statistics on the number of tests being performed by the Genomic Medicine Service (see figure 5 below).[46]

*Figure 5: Number of genomic tests carried out nationally by the NHS Genomic Medicine Service by month, April-September 2023*




These data show a slight increase in genomic testing over the six month period. In September 2023 65,883 tests were conducted, 7,843 more tests than in April 2023, representing a 13.5% increase. The September figure was a slight reduction on the highest month recorded – which was June – when 68,199 genomic tests were undertaken.

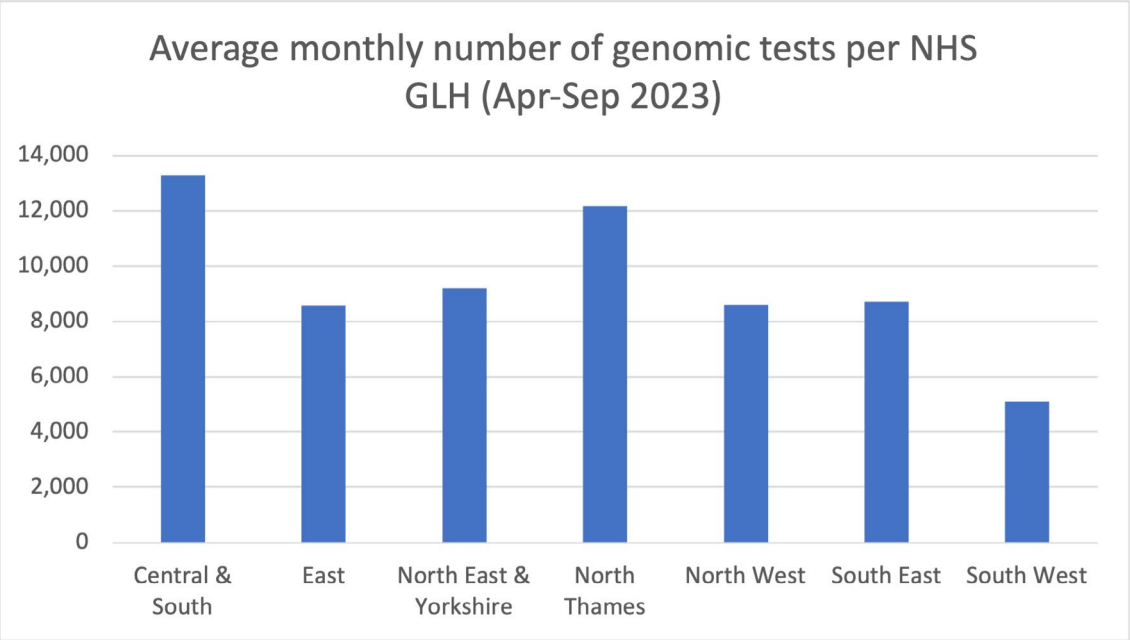
Regionally the number of tests conducted varies. On an average monthly basis Central and South GLH conducted the most tests with an average of 13,276 tests. South West GLH conducted the fewest number with an average of 5,111.

44 https://www.theguardian.com/society/2023/apr/03/backlog-in-nhs-genome-service-leaves-families-facing-long-wait-for-results

45 https://www.thetimes.co.uk/article/nhs-backlog-patients-it-system-genetic-test-diagnosis-2023-2tj7rksfp

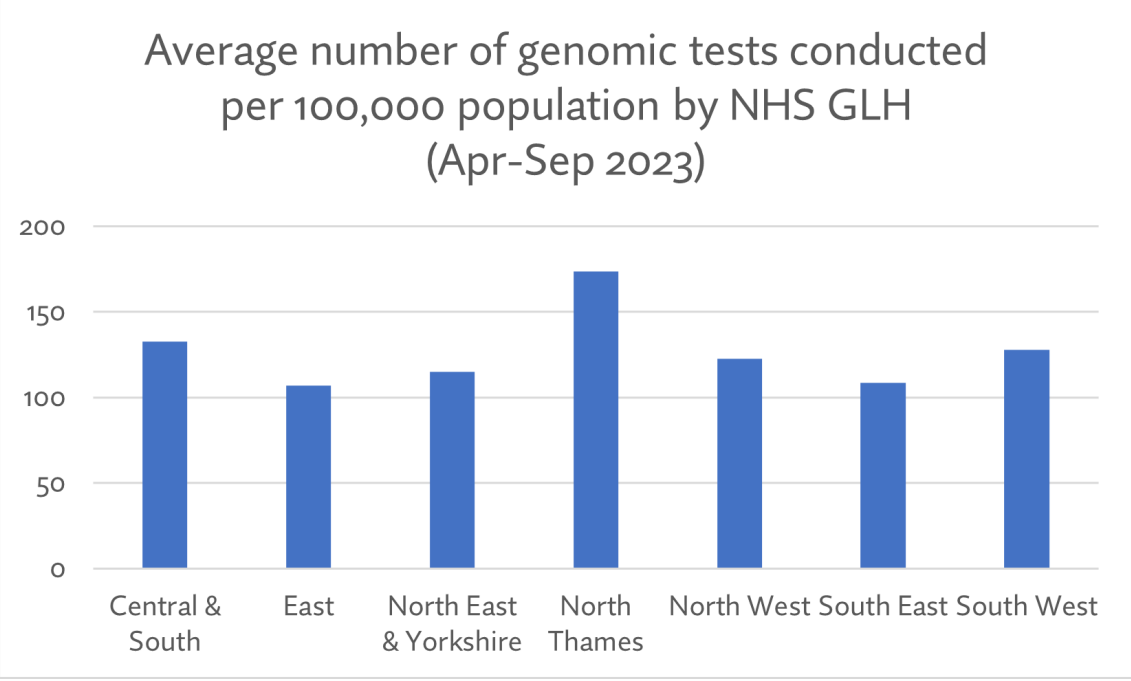
46 https://www.england.nhs.uk/statistics/statistical-work-areas/genomic-testing-activity/ (note that updated activity data was published in mid March 2024 and covered the April-December 2023 period)

*Figure 6: Average monthly number of genomic tests per NHS GLH April-September 2023*




This variation is perhaps not surprising given the different populations covered by each of the GLHs. When the number of tests is divided by the population served, a different picture emerges as seen in figure 7 below.

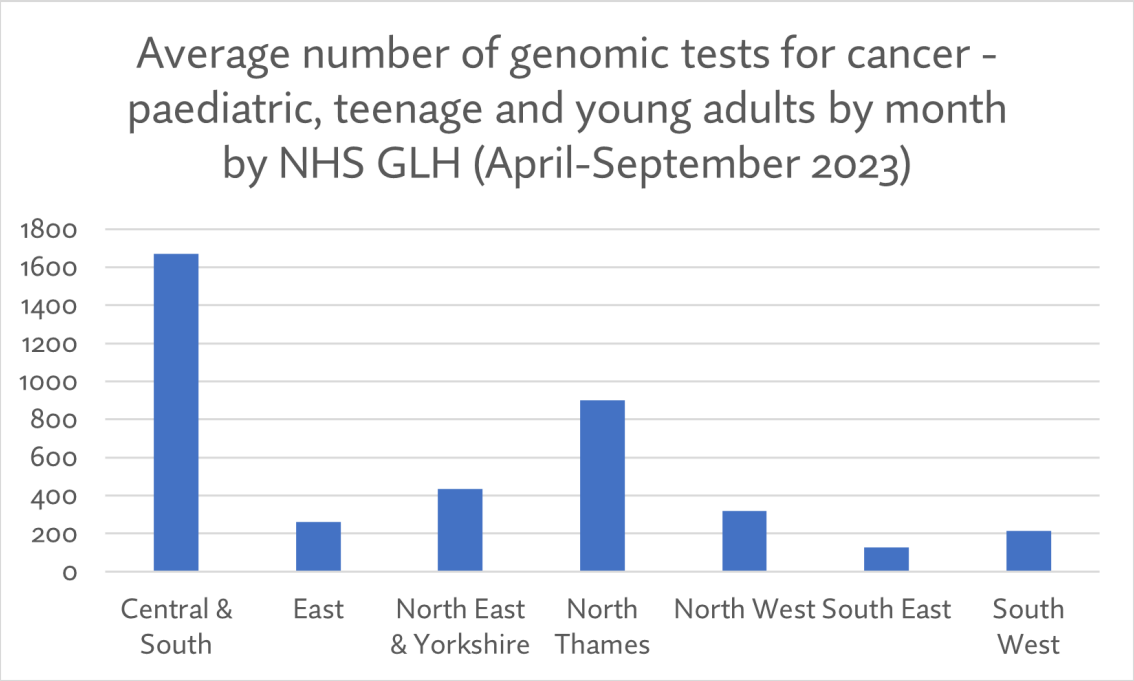
*Figure 7: Number of genomic tests per 100,000 population by GLH*



On the basis of tests per 100,000 population North Thames GLH is conducting the highest number of genomic tests, 174 per 100,000 population each month. By contrast the East and South East GLHs are carrying out the fewest, 107 and 109 tests per 100,000 population respectively each month. It is important to note that not all GLHs are able to carry out all genomic tests in the National Genomic Test Directory, with particular expertise and capacity sitting within individual GLHs. Testing capacity is as a result shared across the system, with some GLHs carrying out tests on populations served directly by other GLHs.

For example when looking at genomic testing for paediatric, teenage and young adult cancer testing, there is a 13 fold variation in the average number of monthly tests conducted between Central and South GLH (1670 monthly tests) and South East GLH (126 monthly tests).

*Figure 8: Average number of genomic tests for cancer - paediatric, teenage and young adults by month by NHS GLH (April-September 2023)*




However when looking at genomic testing for two major cancers (colorectal and lung), the variation in genomic testing between GLHs is far less pronounced.

*Figure 9: Average number of monthly genomic tests for colorectal cancer by NHS GLH, April-September 2023*

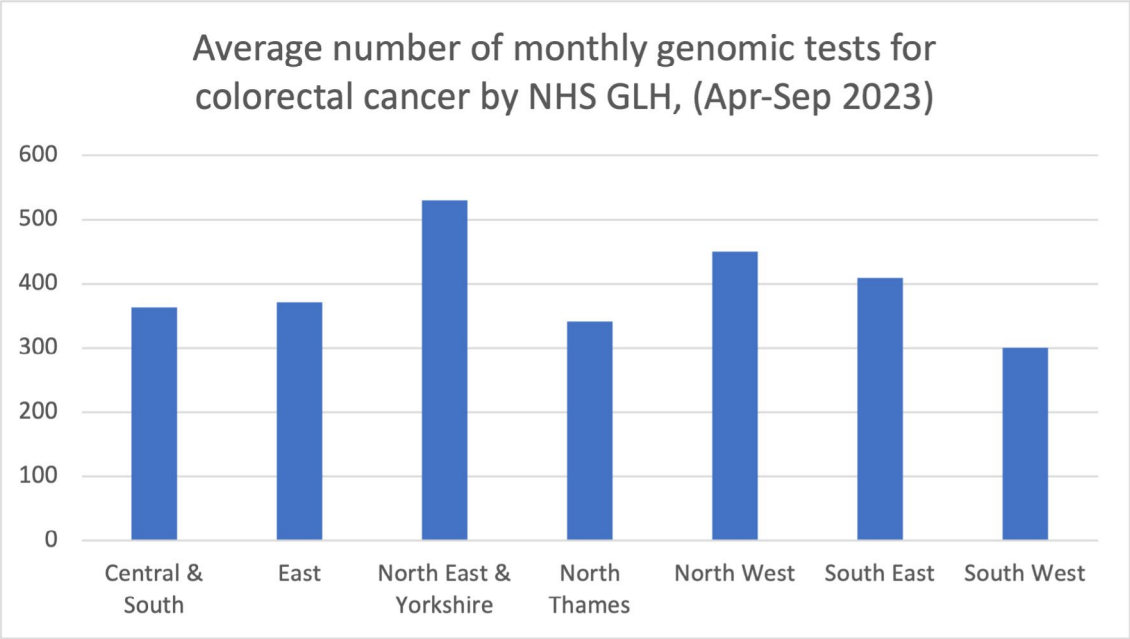
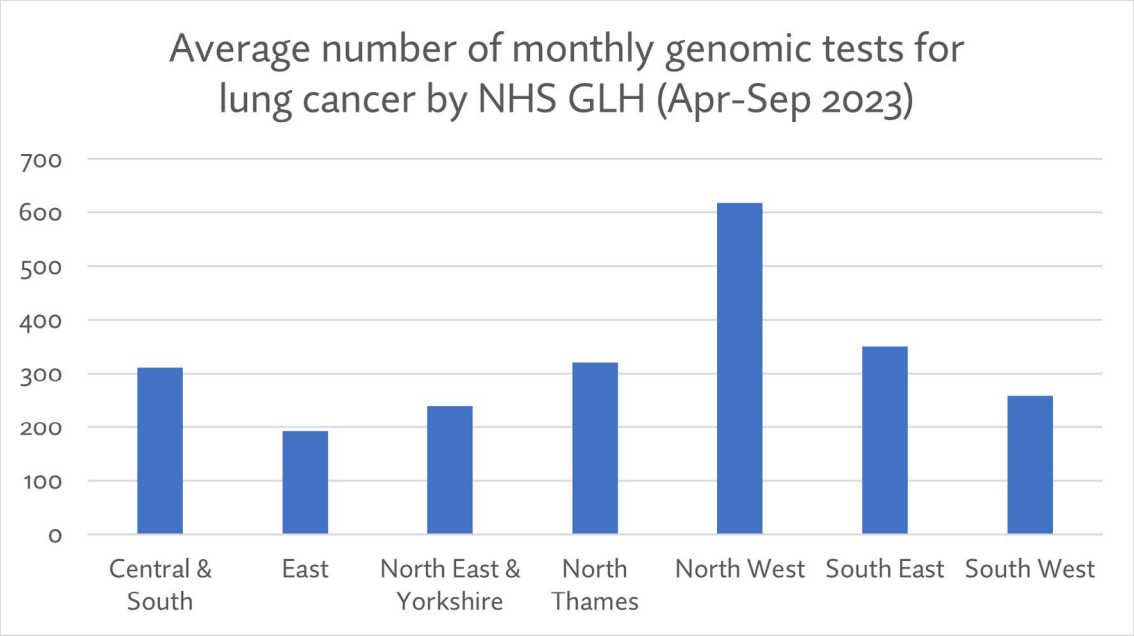


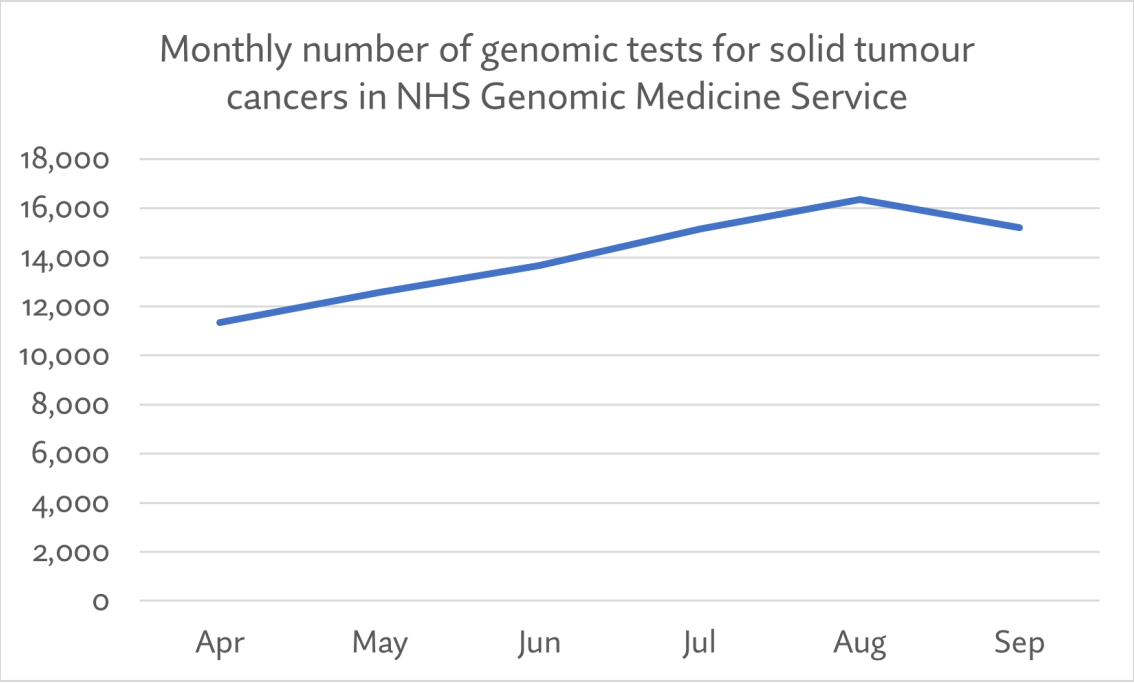

*Figure 10: Average number of monthly genomic tests for lung cancer by NHS GLH, April-September 2023*



For colorectal cancer the variation in average genomic testing per month between the GLH with the lowest and highest number of tests is only 1.76, for lung cancer the variation is slightly higher at a multiple of 3.22.

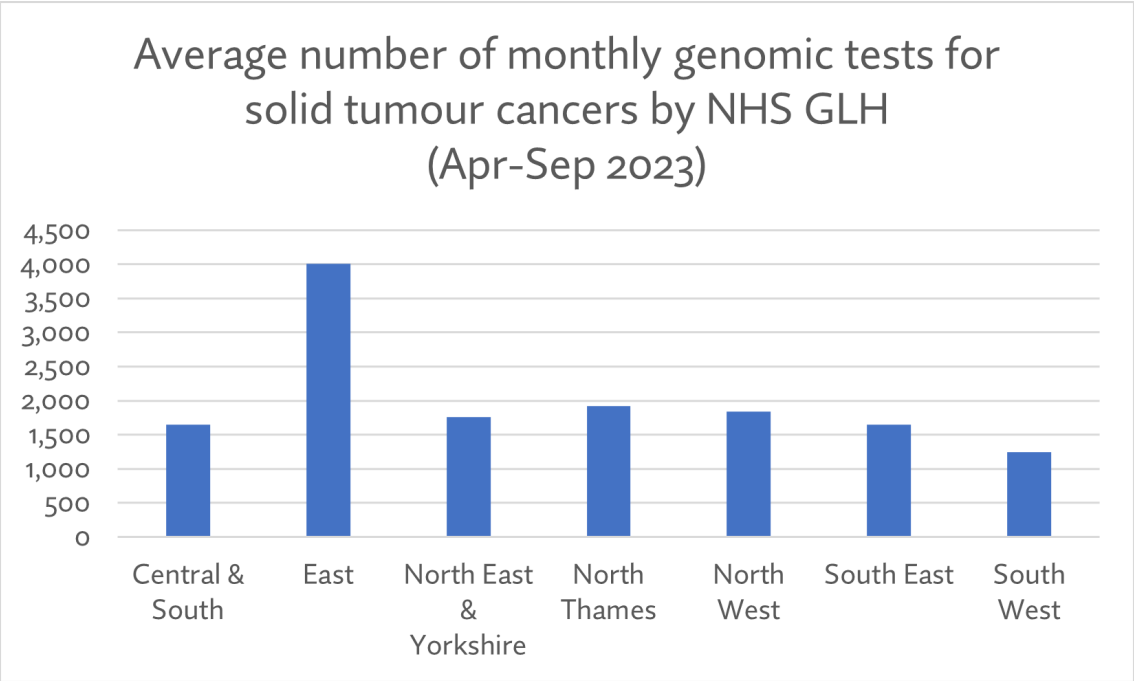
Across solid tumours there is evidence that the NHS Genomic Medicine Service is increasing capacity overall. The number of genomic tests for solid tumour cancers nationally has increased by 22% in the six month period in the data, though dropped slightly from an August high of 16,347 tests.

*Figure 11: Monthly number of genomic tests for solid tumours across the NHS Genomic Medicine Service*




If this six monthly data was replicated over a whole year, the NHS would be conducting nearly 170,000 genomic tests annually for solid tumour cancers. If the growth trajectory witnessed here, was replicated over the next two years, the number of genomic tests for solid tumour cancers annually would more than double, to over 400,000 tests a year, nearly 35,000 per month. Currently the average number of tests for solid tumours cancer by NHS GLH is 2,000 per month. East GLH conducts the highest average number of tests, with over 4,000 tests a month completed, this is twice as many as the next GLH, North Thames 1,916. The South West GLH, serving the smallest population carries out the fewest tests of the seven GLHs.

*Figure 12: Average number of monthly genomic tests for solid tumour cancers by NHS GLH (Apr-Sep 2023)*



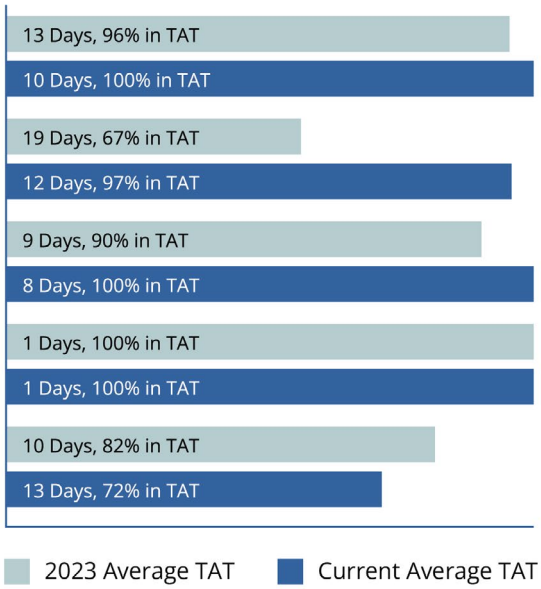

NHS England's *Accelerating Genomic Medicine Strategy* commits to the publication of activity data and turnaround times for the Genomic Medicine Service.[47] Currently such data is not easily available. Some GLHs are publishing their data on genomic testing turnaround times. For example the South West NHS GLH and East NHS GLH have published the following on their performance against turnaround times for solid tumour cancers on their websites.

[47] https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

*Figure 13: NHS GLH turnaround times for genomic cancer testing*

## South West GLH [48]

| Category | NHSE TAT Target |
|---|---|
| Tumour NGS Panels | 21 Days |
| Tumour Gene Fusion Panel | 21 Days |
| Targeted Molecular Analysis | 14 Days |
| DPYD | 5 Days |
| FISH | 14 Days |




## East GLH [49]

| Clinical Urgency | Example Test | NHSE Target TAT (Days) | Nottingham | Cambridge | Leicester |
|---|---|---|---|---|---|
| Urgent - Rapid | FISH for solid cancers | 14 | 85% | 96% | 100% |
| Urgent - Rapid | MGMT promoter methylation | 14 | N/A | 100% | N/A |
| Urgent - Rapid | BRAF | 14 | 100% | N/A | N/A |
| Urgent - Rapid | NGS - targeted DNA | 21 | 10% | 98% | 100% |
| Urgent - Rapid | NGS - targeted RNA | 21 | N/A | 98% | N/A |
| Urgent - Rapid | Pan cancer - DNA | 21 | N/A | 57% | N/A |
| | Pan cancer - RNA | 21 | N/A | 55% | N/A |
| Urgent - Rapid | FISH for solid cancers | 21 | 63% | N/A | N/A |
| Non-urgent Standard | MSI testing & MLH1 promoter methylation | 42 | 0% | 95% | N/A |

[48] https://www.nbt.nhs.uk/south-west-genomic-laboratory-hub/swglh-quality (Accessed April 2024)

[49] https://www.eastgenomics.nhs.uk/about-us/quality/service-turnaround-times/ (Accessed April 2024)

Gaining a complete picture of the turnaround times of the GLHs is not possible. The data from these two GLHs – which is welcome – highlights variation in the meeting of the NHS England Turnaround Times depending on the category of tests assessed. Though the data is limited and difficult to compare. For example FISH Tests for solid cancers are expected to be turned around within 14 days. In the South West the average Turnaround Time in 2023 for such tests was 10 days, with 82% of tests meeting the NHS target. In East GLH no average data is recorded, but data is provided at a location level and shows compliance rates. For FISH tests for solid cancers this shows that whilst Leicester is delivering 100% of tests within the Turnaround Time, Nottingham is delivering 85%.

Whilst the number of tests being conducted has risen so has the number of approved tests on the National Genomic Test Directory – putting pressures on the system. For example the number of tests for solid tumour cancers has increased from 111 to 140 since 2021.[50]

Published material and correspondence within the NHS Genomic Medicine Service highlights the pressures that services are under to meet published turnaround times.

The North West Genomic Medicine Service notes on its website that 'following the recent introduction of new elements of the test directory a number of services are reporting results outside of the below guidelines.'[51] In June 2023 East GLH noted that there would be delays to the Inherited Cancer genomic testing service due to 'ongoing pressures across our genomic services and increases in testing volume.'[52] In November 2023 a letter to GLH users in the North East and Yorkshire GLH cited challenges around national and local reconfiguration and staffing levels as the source of ongoing delays to turnaround times.[53]

## Summary

- There is evidence that testing capacity within the NHS Genomic Medicine Service is increasing, with a 13.5% increase in overall genomic tests conducted across the six months of data analysed. For cancer the number of genomic tests has increased by 22% over the same period
- The volume of testing per population served by each GLH varies. North Thames GLH is conducting the highest estimated number of genomic tests, 174 per 100,000 population each month. By contrast the East and South East GLHs are carrying out the fewest, 107 and 109 estimated tests per 100,000 population respectively each month

50 https://www.england.nhs.uk/publication/national-genomic-test-directories/ , v8 vs version 1

51 https://mft.nhs.uk/nwglh/quality/laboratory-test-service-turnaround-times/

52 https://buckup-cuh-production.s3.amazonaws.com/documents/EastGLH_Letter_Inherited_Cancer_Delays_June2023.pdf

53 https://www.leedsth.nhs.uk/a-z-of-services/the-leeds-genetics-laboratory/ , number2 November 2023

- Regionally GLHs are processing different volumes of genomic cancer tests based on capability and capacity. There is a 13 fold variation in the number of monthly tests conducted for paediatric, young people and teenage cancers. Variations for solid tumour cancers such as lung cancer and colorectal cancer are less substantive
- The number of approved tests on the National Genomic Test Directory is increasing. The number of approved tests for solid tumour cancers has increased by 26% since 2021
- It is not possible to fully assess the performance of the NHS Genomic Medicine Service against national targets for turnaround times for genomic tests as the data is not published
- Correspondence within the NHS Genomic Medicine Service and regional data published by GLHs shows a series of pressures and challenges facing the system in meeting national turnaround time targets

# NEXT STEPS FOR THE NHS GENOMIC MEDICINE SERVICE – ACCELERATING PROGRESS IN CANCER PATIENT ACCESS TO GENOMIC TESTING

NHS England's strategy *Accelerating Genomic Medicine in the NHS* focused on four areas for embedding genomics in the NHS:

- Co-creating services, infrastructure and an operating model with patients and the public
- Developing a sustainable infrastructure across testing, clinical services and research and innovation
- Building greater clinical and professional leadership and developing the capacity and capability of the workforce
- Developing national and international collaborations and partnerships[54]

A series of actions were committed to within the first year of the strategy. In response to a written parliamentary question in summer 2023 Minister Will Quince noted that the NHS was on track to deliver on the commitments made in the strategy:

'To date, the NHS Genomic Medicine Service is on track to deliver the commitments outlined in the strategy for 2023/24, including continuing to review the latest scientific evidence to inform and update the National Genomic Test Directory; launching a rapid whole genome sequencing service in October 2022; exploring the introduction of innovative genomic sequencing techniques, which can be applied to a range of clinical applications, including cancer; continuing to extend the use of molecular diagnostics and increasing the number of tests offered; establishing a NHS Data and Digital Board; and progressing work to establish NHS Genomic Networks of Excellence, that will play a key role in bringing together stakeholders to accelerate evidence generation to support future commissioning decisions for innovative genomics services in the NHS.'[55]

The most recent update to the NHS England Board noted that patient access to genomic testing was continuing to increase and that Genomic Networks of Excellence had now been established.[56] A previous update in July 2023 had highlighted a series of workshops aimed at ratifying pathways and turnaround times for genomic tests for solid cancers.[57]

The commitments and a high-level summary of progress against them is summarised in the table on the next page.

54 https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/
55 https://questions-statements.parliament.uk/written-questions/detail/2023-05-09/184143/
56 https://www.england.nhs.uk/long-read/operational-performance-update-7-dec-23/
57 https://www.england.nhs.uk/long-read/annex-operational-performance-update/

*Table 1: Summary of progress in meeting year one commitments in Accelerating Genomic Medicine in the NHS*

| Theme | Priority | Commitment | Progress |
|---|---|---|---|
| Patient and public engagement | Co-creating services, infrastructure and an operating model with patients and the public | The NHS will continue to work at a national and regional level to raise awareness of genomics, including the benefits and challenges, and support ongoing and transparent public dialogue on issues relating to genomics. | The NHS Genomic Medicine Service website notes that the service has a People and Communities Forum: https://www.england.nhs.uk/genomics/governance/<br><br>There is though little public information on the Forum and its activities. A recent advert was posted to recruit a new independent Chair for the Forum.[58] The NHS Genomic Medicine Service X (formerly twitter) account has not been active since May 2023.[59]<br><br>Regionally GLHs and GMSAs have set-up their own patient and public forums and are recruiting to them, Examples include:<br><br>North East and Yorkshire: https://ney-genomics.org.uk/about-genomics/patient-and-community-forum/<br><br>North Thames: https://norththamesgenomics.nhs.uk/patients-and-public/patients-and-public-involvement/<br><br>South East: https://southeastgenomics.nhs.uk/patients-and-public/#how-to-get-involved<br><br>However it is difficult to ascertain how effective and wide-ranging such engagement is. Some of the initiatives are not well publicised and it is difficult to determine what impact such engagement is having. |

[58] https://www.jobs.nhs.uk/candidate/jobadvert/M9990-24-0184?location=LS2%207JS&language=&page=23

[59] https://twitter.com/nhsgms?lang=en

| Theme | Priority | Commitment | Progress |
|---|---|---|---|
| Governance and structures | Developing a sustainable infrastructure across testing, clinical services and research and innovation | The NHS GLHs, NHS GMSAs and clinical genomics services will establish integrated NHS Genomic Medicine Service governance boards with appropriate partnerships and leadership to drive forward the embedding of genomics into the wider NHS. | Integrated models for delivering the Genomic Medicine Service at a regional level, between the main constituent parts (GMSA, GLHs and clinical genetics services) appear to have been implemented. Examples include:<br><br>Central and South Genomics:<br><br>https://centralsouthgenomics.nhs.uk/who-we-are/<br><br>North East and Yorkshire:<br><br>https://ney-genomics.org.uk/person/<br><br>However from public sources it is difficult to discern if formal governance arrangements and boards have been established. |
| Workforce | Building greater clinical and professional leadership, and developing the capacity and capability of the workforce | Over the next year, NHS England and the Health Education England Genomics Education Programme are working together to develop a Genomic Training Academy. | The Genomic Training Academy has been established but information on its operation and progress to date is limited.[60] Health Education England (now part of NHS England) has an extensive library of materials aimed at educating the healthcare workforce on genomics.[61] |

[60] https://www.genomicseducation.hee.nhs.uk/about-us/gtac/

[61] https://www.genomicseducation.hee.nhs.uk/

| Theme | Priority | Commitment | Progress |
|---|---|---|---|
| Data | Developing an interoperable informatic and data infrastructure that enables the NHS to use and share genomic data appropriately to improve patient care | Over the next year, the NHS will develop shared data standards.<br><br>During 2023, the NHS working with partners such as Genomics England will publish a genomics informatics implementation plan. | The proposed genomics informatics implementation plan has not yet been published with work instead re-geared to align with wider NHS England activity on data transformation policy. |
| Uptake of innovation | Enriching existing and developing new NHS Genomic Medicine Service relationships to support innovation and the generation of evidence for adoption and improvements in health and care | NHS England will, as part of the evolving NHS GMS alliance infrastructure, establish 'NHS genomic networks of excellence'. | Eight Genomic Networks of Excellence have been established<br>• Prenatal genomic medicine<br>• Circulating tumour biomarker testing<br>• Haemato-oncology<br>• Rare and inherited disease<br>• Severe presentation of infectious disease<br>• Improving the identification and outcomes for individuals with inherited and acquired cardiovascular disease<br>• Pharmacogenomic and medicines optimisation<br>• Genomics artificial intelligence (AI)[62]<br><br>The most recent update to the NHS England Board noted that they were expected to deliver work packages in early 2024.[63] |

[62] https://www.england.nhs.uk/genomics/nhs-genomic-networks-of-excellence/

[63] https://www.england.nhs.uk/long-read/operational-performance-update-7-dec-23/

Progress has been made across the five areas expected to be delivered within the first year of the NHS Genomic Strategy, but from public sources the level progress would appear to be mixed.[64]

Genomic Networks of Excellence have been established, a training academy set-up, integrated governance models and patient and public forums built. However a genomics informatics implementation plan has not been published and in some areas such as patient and public involvement and the operation of governance arrangements it is difficult to appraise the success or otherwise of efforts to date from public sources. In other areas such as the Genomics Training Academy and Genomic Networks of Excellence it is too early to make a judgement on their impact.

*Accelerating progress in unlocking access to genomic testing for cancer patients in the NHS*

As set out above, data from the NHS Genomic Medicine Service demonstrates that it is steadily increasing testing capacity and a national strategy is in place to further develop the system so that more cancer patients can benefit from genomic testing.

However the service currently faces a series of challenges and barriers to delivering on its ambitions. Many of the issues facing the Genomic Medicine Service are reflective of the wider challenges across the NHS, including workforce pressures, capacity backlogs, disjointed data and technological systems and variable implementation of clinical pathways.

### Increasing testing capacity

As set out in the previous chapter it is difficult to determine the full scale of the pressures within the Genomic Medicine Service. Whilst the number of tests conducted is increasing, published correspondence from GLHs, turnaround time data from specific regions and media reports indicate a system under pressure.[65] In particular the shift from single tests to panel tests is putting additional strain on the system due to the added complexity and time for interpretation involved. Wider capital challenges in the NHS, including inappropriate or outdated buildings and unreliable courier services were other challenges identified at a recent Institute of Biomedical Science roundtable.[66]

Challenges in the system are not new. Before the pandemic there was evidence of difficulties for services in meeting turnaround times for genomic testing. For example, a spotlight report on molecular testing in advanced lung cancer found that median turnaround times from tissue acquisition to EGFR mutation result was 18 days against a target within the National Optimal Lung Cancer Pathway (NOLCP) of ten days.[67] [68] These are complex next generation sequencing tests which require specialist expertise; creating challenges for systems in processing them within the set turnaround times.

---

64 NHS England has confirmed to Future Health that all 36 actions committed to through the strategy have progressed but not all progress is documented in the public domain

65 https://www.theguardian.com/society/2023/apr/03/backlog-in-nhs-genome-service-leaves-families-facing-long-wait-for-results#:~:text=The%20Genomics%20Medicine%20Service%2C%20launched,to%20allow%20more%20targeted%20treatments.

66 https://www.ibms.org/resources/news/time-to-test--six-key-challenges-facing-cancer-testing/

67 https://www.hqip.org.uk/wp-content/uploads/2020/01/REF100_NLCA_Spotlight-Molec-Test_FINAL-TYPESET_web_20200108.pdf

68 https://www.roycastle.org/app/uploads/2019/07/Lung_Cancer_Implementation_Guide_August_2017.pdf

A review of lung cancer services by NHS England's Getting It Right First Time Programme published in 2022 noted improvements in meeting the standard, but highlighted that the target was being met by less than four in ten services.[69]

Looking ahead there are concerns about the level of capacity in the Genomic Medicine Service to conduct more and more genomic tests and the process whereby tests are added to the testing directory. Approved tests are included in the National Genomics Test Directory which is expected to be updated annually, but since October 2021 the cancer directory has been updated 11 times in 27 months, meaning an average update every 2.5 months.[70] Such iterative updates create challenges for local services in ensuring that the right infrastructure and resource is in place to deliver the new tests.

NHS England has established a 'fast track process' for amendments to the Test Directory, this includes errors that need to be corrected, actions in emergencies (e.g. pandemic) and changes in guidance, clinical policies or carrying out statutory responsibilities.

Whilst horizon scanning for likely updates to the Test Directory with critical agencies such as NICE and MHRA is ongoing - an annual outlook should be published of possible updates to the Test Directory that can support regional systems in local planning.[71]

The existing regional services upon which the Genomic Medicine Service was built had expertise primarily in rare diseases rather than cancer testing which was located elsewhere in the system. There were also concerns expressed at the time of the service being established that the creation of separate Genomic Medicine Service hubs would present challenges in keeping developments in genomic technology closer to patients through local pathology networks.[72]

The centralised regional hubs have delivered an increase in cancer genomic testing, but there is evidence that they are struggling to meet national turnaround times. Anecdotal evidence has highlighted local services undertaking their own cancer genomic tests outside of the Genomic Medicine Service to avoid long turnaround times.

69 https://gettingitrightfirsttime.co.uk/wp-content/uploads/2022/09/lung-cancer-overview-article-FINAL.nhse_.pdf

70 https://www.england.nhs.uk/publication/national-genomic-test-directories/

71 https://www.england.nhs.uk/genomics/the-national-genomic-test-directory/

72 https://data.parliament.uk/writtenevidence/committeeevidence.svc/evidencedocument/science-and-technology-committee/genomics-and-genome-editing-in-the-nhs/written/71574.pdf

The scale of such practice however is difficult to ascertain. It is important to be clear that the cause of any delays in turnaround times is complex. NHS England is undertaking a number of steps to monitor this across the NHS GLHs, including through implementing patient level contract monitoring (PLCM) to facilitate a national approach to reporting and validating activity data and turnaround times and undertaking a quarterly assurance process with each NHS GLH to monitor improvements in turnaround times to ensure these are being met in every region and for all patients.

The solution is to ensure that where appropriate, tests are being done around the needs of patients, with GLHs working with other local services, such as local pathology networks and Community Diagnostics Centres to deliver additional cancer testing capacity. The Genomic Pathology Accelerator programme and development of Cellular Pathology Genomic Centres is aimed at supporting this, and NHS England should report back on the progress of these programmes in expanding testing capacity and reducing turnaround times by the end of 2024.

Where such testing is delivered outside the Genomic Medicine Service, funding needs to be in place to support such capacity which is itself constrained with workforce shortages from technical laboratory staff to consultants and demands for an ever increasing number of tests linked to an ageing population and increasing burden of disease.[73]

Part of the challenge in delivering this will be different contractual arrangements for the different services involved. The NHS Genomic Medicine Service is funded through a block contract with an activity based element to cover testing growth on top. By contrast local pathology services have more diverse funding streams from local primary and secondary commissioners.

Moving from a block and activity growth contract between NHS England and regional GLHs to a more activity led contract for the NHS Genomic Medicine Service with an ability for groups of providers regionally to work together to deliver increased testing rates is a potential route forward to enable this. Though it is important to note that such a change would almost certainly need additional resource overall in order to be delivered.

[73] https://thebiomedicalscientist.net/science/transforming-histopathology

## Recommendations

**Recommendation 1:** An annual multiorganisational horizon scanning exercise of potential genomic tests (including NHS England, NICE and MHRA) to be added to the directory should be undertaken and published. The number of updates 'in year' to the National Genomic Test Directory should be minimised where possible

**Recommendation 2:** NHS GLHs should be enabled to work closely with other local testing services to maximise cancer testing capacity and improve turnaround times. NHS England should explore new contracting and funding arrangements for increasing genomic testing capacity for cancer and to ensure Trusts and local services do not undertake such activity at their own financial risk. The UK Government will need to support such changes through providing the necessary resource for the service re-design and expansion of capacity proposed

**Recommendation 3:** By the end of 2024, NHS England should publish a Board level update on progress with the Genomic Pathology Accelerator Programme and the development of Cellular Pathology Genomic Centres and their impact on increasing genomic testing capacity and reducing turnaround times for tests

### Monitoring and improving service performance

The increasing importance and use of genomic testing within cancer patient pathways means that there is an urgent need for better public data on the performance of the NHS Genomic Medicine Service. The publication of testing volume data in December 2023 was an important step forward. But NHS England should now go further and as part of investment in core service data infrastructure and capability publish a monthly dashboard for the NHS Genomic Medicine Service that covers the number of genomic tests provided across each of the tests in the National Test Directory and standardised turnaround times data so that a comparison can be made across the seven regional services. The breakdown of the different population groups accessing tests should also be reported so that inequities in access between different groups is understood and can be addressed. Qualitative data from clinicians and patients using the service should also be developed and incorporated into the dashboard. This could also include how often services have provided advice to clinicians on what tests to request.

Where performance is outside expected levels, NHS England and the relevant regional service should publish a public recovery plan with clear timelines for improvement.

In December 2022, the Government's implementation plan for *Genome UK* included a set of high-level metrics to quantify long-term changes in the genomics environment and measure progress against the strategy's ambitions. The high level

metrics included a focus on monitoring whether genomic testing was available to all people with cancer where it would be clinically beneficial. The document committed to explore how such data could be broken down regionally and by specific test into a baseline report.[74]

Whilst regular reporting on progress is delivered to Ministers and the NHS England leadership, no public baseline report has been produced, making it difficult for the public to understand the performance of the Genomic Medicine Service and the progress on the actions agreed in the strategy.

Ministers working with the OLS should urgently bring forward a final agreed list of metrics to ensure that the ambitions of the strategy are delivered and any issues regarding progress are identified and quickly addressed.

### Recommendations

**Recommendation 4:** NHS England should publish a monthly performance dashboard for the Genomic Medicine Service covering data across the full range of genomic tests, including number of tests and turnaround times.[75] This should be delivered as part of NHS England's investment in data capability and also include qualitative feedback from those using the service including clinicians and patients. Services performing outside agreed performance levels should publish an agreed recovery plan with timelines for improvement clearly set [76]

**Recommendation 5:** DHSC and OLS should publish a baseline report with agreed high level metrics assessing progress in implementing the *Genome UK* Strategy. This should include a set of metrics assessing the access of cancer patients to clinically beneficial genomic tests through the Genomic Medicine Service

## Investing in and connecting data and technological systems

Despite the development of the national NHS Genomic Medicine Service in 2018, and the creation of a national network for sharing processes and practice, the foundations and day to day operation of the service remain regional, with variable digital systems and capability. Such practice is longstanding and not uncommon. A Getting It Right First Time assessment of NHS pathology services noted labs referring tests to other labs on paper in 2021.[77]

[74] https://assets.publishing.service.gov.uk/media/639842e8e90e077c2b945347/genome-uk-annex-a-high-level_metrics.pdf

[75] The original commitment in the NHS Genomic Strategy was for quarterly reporting which is being delivered. Over time the ambition should be for monthly data to be published

[76] Such discussions and plan development does take place between NHS England and relevant GLHs, but recovery plans should be made publicly available

[77] https://gettingitrightfirsttime.co.uk/wp-content/uploads/2022/05/Pathology-4May22j.pdf

*Accelerating Genomic Medicine* highlighted the variability of regional GLH IT systems noting different levels of digital maturity and in places the ongoing use of paper based records as barriers to improving the performance and operation of the NHS Genomic Medicine Service.[78]

This followed the Government and NHS health data strategy *Data Saves Lives* which committed to integrating genomic data and feeding it back into clinical care by the end of December 2023.[79]

The use of paper based systems is a barrier to delivering on turnaround time targets, and in also publishing up to date data on the performance and pressures within the system as set out above.

In the Spring Budget 2024 the Chancellor committed to a £3.4 billion Productivity Plan for the NHS, this included:

- £1 billion to transform the use of data to reduce time spent on unproductive administrative tasks by NHS staff, enabling more than £3 billion of savings over five years
- £2 billion to update fragmented and outdated IT systems across the NHS, reducing 13 million hours wasted by doctors every year and enabling up to £4 billion of savings over five years

The funding pledged will also support all NHS Trusts in implementing an electronic patient record by March 2026 and lay the ground work for the uptake of new technologies such as artificial intelligence.[80]

In 2020 the PHG Foundation set out a series of ways in which artificial intelligence was playing an expanding role in genomic medicine, in both the research and clinical areas. In the clinical domain, artificial intelligence was identified as supporting time sensitive analysis, interpreting test results and genetic counselling. [81] The report was clear that despite the excitement in AI, such technology on its own would not transform genomic medicine without investment in 'workforce training, considered implementation, patient and public engagement, robust ethical appraisals, and other types of technologies, for example non-AI statistical techniques.'[82] It also noted that other disciplines such as medical imaging, predicting acute illness, and handling repetitive administrative tasks were likely to see a more substantial earlier benefit from the introduction of AI than genomics.[83]

---

78 https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/#appendix-1-overview-of-the-nhs-gms

79 https://www.gov.uk/government/publications/data-saves-lives-reshaping-health-and-social-care-with-data/data-saves-lives-reshaping-health-and-social-care-with-data ; it is unclear if this commitment has been met

80 https://assets.publishing.service.gov.uk/media/65e8578eb559930011ade2cb/E03057752_HMT_Spring_Budget_Mar_24_Web_Accessible__2_.pdf

81 https://www.phgfoundation.org/publications/reports/artificial-intelligence-for-genomic-medicine/

82 https://www.phgfoundation.org/publications/reports/artificial-intelligence-for-genomic-medicine/

83 https://www.phgfoundation.org/publications/reports/artificial-intelligence-for-genomic-medicine/

*Accelerating Genomic Medicine* committed to pilot new technologies such as AI and machine learning to support the improved operation of the Genomic Medicine Service.[84] The creation of new Genomic Networks of Excellence creates a way forward to accelerate the testing and adoption of these new technologies across the Genomic Medicine Service.[85] The funding announced in the Spring Budget 2024, which will not come on stream until 2025/2026 presents an opportunity to upgrade data infrastructure and scale the adoption of proven technologies across the system.[86]

It will be important that the pilot programmes through the Genomics Networks of Excellence collect a range of evidence not only on their effectiveness in direct service performance but also their wider economic impact - as they have committed to. Collecting such evidence will help with making an ongoing case for investment in new technologies in the Genomic Medicine Service to the Treasury in future years.

**Recommendations**

**Recommendation 6:** Funding for the NHS Productivity Plan in the 2024 Budget should be used to upgrade the IT and data capability of the Genomic Medicine Service. Funding should also be assigned to support the effective evaluation and adoption of schemes piloting the use of innovative new technologies through the recently announced Genomic Networks of Excellence

**Recommendation 7:** Regional Genomic Networks of Excellence should ensure that they collect a range of data including economic impact data on the introduction of new innovations, such as automation and artificial intelligence, aimed at improving the operation of the Genomic Medicine Service to help support future investment cases with Government

**Recommendation 8:** NHS England should provide quarterly updates on progress in implementing the commitments in the health data strategy, *Data Saves Lives*. Where targets are missed or off track, actions should be set out for addressing the delays in their delivery

84 https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/#appendix-1-overview-of-the-nhs-gms

85 https://www.england.nhs.uk/genomics/nhs-genomic-networks-of-excellence/#:~:text=The%20NHS%20Genomic%20Networks%20of,towards%20rapid%20informing%20commissioning%20decisions

86 There also may be opportunities to use funds flowing back into Government and the NHS from the Voluntary Scheme for Branded Medicines Access and Growth (VPAG) here too: https://www.gov.uk/government/publications/2024-voluntary-scheme-for-branded-medicines-pricing-access-and-growth

## Easing workforce pressures

Cancer Research UK's report, *Estimating the cost of growing the NHS cancer workforce in England by 2029* indicates that a 45% staff increase is needed across seven cancer-related professions to meet Health Education England's (HEE) aim to provide world-class services for cancer patients by 2029.[87]

One of the major challenges in the system is the need to expand the workforce. Workforce challenges translate into delays in interpreting tests and longer turnaround times.

The Royal College of Pathologists (RCPath) workforce census, *Meeting Pathology Demand* found that just 3% of histopathology departments have enough staff to meet clinical needs.[88]

The NHS *Long Term Workforce Plan* did not set out detailed future projections for the scientific workforce and omitted certain professionals (such as biomedical scientists). However it did highlight a 65% expansion of training places through the three-year Scientist Training Programme in 2022/23.[89]

Action to maximise the capability and capacity of the existing workforce will be particularly important given current pressures. There are opportunities to embed biomedical scientists more closely within clinical teams to support dedicated and relevant tasks, as well as including pathologists as members of multidisciplinary cancer teams.[90]

Advances in the technological capability of the system and in particular the use of new innovations such as AI as set out above, could help support the healthcare science workforce and increase service output levels. Any such blend of technology and workforce in carrying out such tasks will though need to be carefully appraised by NHS England in collaboration with relevant Royal Colleges.

Clinical staff also need to be trained in the latest genomic technologies and developments to support the effective delivery of the genomics service. A 2023 survey of 50 oncologists by Tutika et al found that 38.7% has received no formal genomics training and 92.7% identified a need for additional genomics training as part of their role.[91]

The NHS *Long Term Workforce Plan* did acknowledge the need to upskill the workforce in genomics and noted the work of the Genomic Training Academy as the primary vehicle for this.[92]

87 https://www.cancerresearchuk.org/sites/default/files/estimating_the_cost_of_growing_the_nhs_cancer_workforce_in_england_by_2029_october_2020_-_full_report.pdf

88 https://www.rcpath.org/discover-pathology/public-affairs/the-pathology-workforce.html

89 https://www.england.nhs.uk/wp-content/uploads/2023/06/nhs-long-term-workforce-plan-v1.2.pdf

90 https://www.ibms.org/resources/news/time-to-test--six-key-challenges-facing-cancer-testing/

91 https://pubmed.ncbi.nlm.nih.gov/36697012/

92 https://www.england.nhs.uk/wp-content/uploads/2023/06/nhs-long-term-workforce-plan-v1.2.pdf

In January 2024 NHS England published a three year strategic framework for upskilling pharmacists in genomics so that genomics can be part of mainstream medicines optimisation work.[93] As training activities through the Genomics Training Academy and associated programmes are rolled out, NHS England should publish regular feedback on participant views on the usefulness and impact of the training and adjust training programmes based on the feedback received. This work can be co-ordinated through regional HEE Genomic Leads who are working with GLHs and GMSAs - who are each producing a Workforce Strategy and People Plan.

### Recommendations

**Recommendation 9:** As regional workforce strategies and people plans are developed, the NHS Genomic Medicine Service and GMSAs should in partnership with the Royal Colleges and professional bodies ensure that workforce models within the Genomic Medicine Service are regularly reviewed and reflect the latest developments in the capability of digital technology and innovation. Such models should also support joined-up multidisciplinary working that maximises the capabilities of the existing workforce, such as biomedical scientists, around the needs of patients. UK Government should continue to invest in the healthcare science workforce through the NHS *Long Term Workforce Plan*

**Recommendation 10:** NHS England should publish quarterly feedback from participants on genomics training programmes undertaken through the Genomic Training Academy and annually set out how it is adjusting training programmes based on the feedback received

## Transforming clinical pathways and service design

*Accelerating Genomic Medicine* set out three objectives for ensuring that the Genomic Medicine Service was incorporating the latest genomic tests.

In the more immediate term (1-3 years) this included a need to 'transform clinical pathways and service models to embed genomics where there is the greatest impact on clinical outcomes and pathway efficiencies.' It also included a focus on driving 'equity in access to genomic testing.'[94]

For cancer the strategy noted that 'pathways will need to be redesigned to enable for example the collection of biopsies for cancer genomics early in the diagnosis pathway to ensure the timely return of genomic test results to inform clinical care and decision making.'

[93] https://www.england.nhs.uk/long-read/pharmacy-genomics-workforce-education-and-training-strategic-framework/

[94] https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/#appendix-1-overview-of-the-nhs-gms

The Strategy highlighted that the NHS Genomic Medicine Service will need to work with partners such as cancer alliances and pathology networks to deliver rapid turnaround times for certain types of cancer where timely results are needed to inform diagnosis and treatment.[95]

In the longer term (3-5 years) the strategy notes that the NHS will 'explore the utility of genomic testing to support population screening for cancer, including reviewing the evidence from population based studies to detect cancer earlier, such as the research trial of the GRAIL Galleri test, and piloting to expansion of BRCA testing in higher risk populations.'[96]

A summary of the current process for ordering genomics tests within the NHS Genomic Medicine Service for solid tumour cancers is set out on the next page:[97]

*Figure 14: Process for ordering solid tumour cancer tests – North East and Yorkshire Genomic Medicine Service*[98]


95 https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

96 https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/#2-delivering-equitable-genomic-testing-for-improved-prediction-prevention-diagnosis-and-precision-medicine ; NHS England provided an update on the GRAIL trial in May 2024: https://www.england.nhs.uk/blog/an-update-on-the-ongoing-nhs-galleri-trial/

97 https://ney-genomics.org.uk/testing/solid-tumours-cancer/

98 Please note that alongside the visual process described and in certain cases consultant biomedical scientists independently report cases with equivalence of pathologists

There are however inconsistencies over when genomic tests are carried out within cancer pathways.

A study published in January 2024 by Raffaella et al in the American Cancer Society's, A Cancer Journal for Clinicians noted that genomic tests remained underutilised in routine cancer care despite leading to better outcomes by informing cancer risk, prognosis, and therapeutic selection. One of the contributing factors to this the authors noted was 'a lack of understanding of their clinical utility and the difficulty of results interpretation by the broad oncology community.'[99] This is seen in demands for existing tests remaining high even when new tests emerge.[100]

The Government's *Genome UK implementation plan* included a commitment for Health Education England (HEE) (now part of NHS England) to 'support clinicians to use the test directory by developing 2 massive open online courses aligned to the rare disease and cancer genomic pathways and continuing the development of their GeNotes resource.'[101]

GeNotes consists of 2 tiers. Tier 1 is mapped to the test directory and supports the clinician to choose the right genomic test for the right patient at the right time and navigate the test directory and its supporting resources. Tier 2 is 'the knowledge hub', providing an extended learning opportunity for clinicians engaging with the resource and content.

Performance was to be judged by (a) the number of people accessing the course and (b) the evaluation of the course. The Government and NHS England should publish performance against these metrics, along with other metrics in the *Genome UK implementation plan*.[102]

Within the NHS the Clinical Pathway Initiative (CPI) is a collaboration between NHS England and the Academy of Medical Royal Colleges (AMRC) to help integrate genomic medicine into clinical practice.[103] The Initiative has five aims:

- Introduce a unified approach to the integration of genomic medicine across the different specialties;
- Harness and share expertise from around the country;
- Identify the workforce development and education needs;
- Highlight gaps in resource provision to support education and training needs; and
- Avoid duplication of effort around resource development

A number of pathway projects have been launched through the CPI, however from the published list to date the main focus of work appears to be within rare disease testing rather than in cancer testing.[104]

---

99 https://acsjournals.onlinelibrary.wiley.com/doi/10.3322/caac.21825

100 https://assets.roche.com/f/172650/x/cd8bca05d8/m-gb-00007998_precision_meds_report_june_22.pdf

101 https://assets.publishing.service.gov.uk/media/6398387e8fa8f55300899840/genome-uk-2022-to-2025-implementation-plan-for-england.pdf

102 The National Genomics Education Team and the NHS GMS Alliances are working together to collect this data

103 https://www.genomicseducation.hee.nhs.uk/the-clinical-pathway-initiative/

104 https://www.genomicseducation.hee.nhs.uk/the-clinical-pathway-initiative/

There have been specific projects aimed at improving cancer pathways. In 2017 NHS England published an optimal pathway for lung cancer based on the input of an expert clinical group. The pathway was revised in 2020 and 2023 and aimed at delivering improved outcomes for lung cancer – which has some of the poorest outcomes of all cancers – through meeting the Faster Diagnosis Standard of diagnosing or ruling out cancer within 28 days following a referral.[105] Within the optimal pathway, services are expected to provide a maximum 10-day turnaround time for molecular profiling.[106] However compliance and uptake of the guideline, overseen by regional cancer alliances, is unclear. This work forms part of a series by NHS England to develop timed pathways across a number of major cancers to meet the faster diagnosis standard.[107] These pathways include short audit tools for services to assess whether they have the capacity and capability to deliver the core requirements of the pathway.

NHS England's Genomic Pathology Accelerator Programme aims to embed the national exemplar pathways for a range of tumour types across regional services.[108] The development and roll out of Cellular Pathology Genomic Centres is expected to improve the co-ordination of cancer pathways locally.'[109]

However guidance for when genomic tests should be deployed within cancer pathways can be uncertain. For example, for tumour agnostic therapies the timing for when to undertake the genomic test is not included in National Institute for Health and Care Excellence (NICE) clinical guidelines, leading to variation in where in the clinical pathway genomic testing is applied. One possible way of tracking the usage of genomic tests in pathways in the short term could be through selecting a small sample of tests from the Test Directory and asking each GLH to collect data on their use in pathways to identify variations in practice. Longer term such data should form part of the NHS Genomic Medicine Service dashboard.

NICE has committed to make its clinical guidelines 'living useful and useable' as part of a new five year strategy.[110] Instead of reviewing and updating each guideline five years after it is published, evidence underpinning the individual or groups of recommendations will be more regularly monitored. One of the first areas identified for this more ongoing monitoring is breast cancer.[111]

105 https://www.england.nhs.uk/long-read/implementing-a-timed-lung-cancer-diagnostic-pathway/#:~:text=The%20National%20Optimal%20Lung%20Cancer%20Pathway%20(NOLCP)%20provides%20a%20detailed,reconfiguration%20can%20facilitate%20NOLCP%20implementation

106 https://www.cancerresearchuk.org/sites/default/files/national_optimal_lung_pathway_aug_2017.pdf

107 https://www.england.nhs.uk/publication/rapid-cancer-diagnostic-and-assessment-pathways/

108 https://centralsouthgenomics.nhs.uk/transformation-projects-patients/genomics-pathology-accelerator-project-gpap/

109 https://www.eastgenomics.nhs.uk/about-us/genomic-medicine-service-alliance/Transformation_Projects/GPAP/

110 https://www.nice.org.uk/about/what-we-do/our-programmes/nice-guidance/nice-guidelines/maintaining-and-updating-our-guideline-portfolio

111 https://www.nice.org.uk/hub/indevelopment/gid-hub10003

Part of the planned ongoing updates to this guideline are based on developments relating to genetic testing for people with early and locally advanced breast cancer. As NICE expands its topic selection for such iterative updates it will be important that other major cancers feature prominently in the expanded list of topics given both the burden of disease and opportunities of diagnostic innovation coming from the latest genomic tests. Next Generation Sequencing panel testing presents an opportunity to move to a 'genomics first' approach to cancer testing, where genomic variants relating to particular cancers are able to be tested at the point of diagnosis.[112]

There have been a series of recent announcements demonstrating progress in delivering greater genomic testing earlier in cancer pathways. In March 2024 the NHS announced the expansion of a pilot programme to use blood tests to test people with suspected lung cancer after a CT scan. The existing approach involves tissue biopsies first being used to confirm a diagnosis of lung cancer and samples then being sent for genomic testing. The NHS decision moves the use of genomic testing earlier in the lung cancer pathway.[113]

In March 2024 NICE guidelines for ovarian cancer were also revised and finalised with a greater focus on improving earlier access to genetic testing for those with or at risk of having a pathogenic variant associated with ovarian cancer.[114]

To meet its commitment within the *Accelerating Genomic Medicine* strategy for transforming cancer pathways over three years, NHS England should seek to co-ordinate the range of initiatives already under-way and work with key actors such as the medical royal colleges, professional bodies and NICE to ensure that guidelines for major cancer pathways are regularly updated to reflect the latest evidence on genomic testing. Over the three year timeline such work should aim to move services closer towards a 'genomics first' approach to cancer testing.

[112] https://assets.roche.com/f/172650/x/cd8bca05d8/m-gb-00007998_precision_meds_report_june_22.pdf

[113] https://www.england.nhs.uk/2024/03/thousands-more-lung-cancer-patients-to-get-innovative-blood-test-as-part-of-nhs-pilot/

[114] https://www.nice.org.uk/guidance/ng241/chapter/recommendations#pathogenic-variant

## Recommendations

**Recommendation 11:** To improve the use of the National Genomic Test Directory and the application of genomic testing for cancer in care pathways, the Government and NHS England should publish data on performance against (a) the number of people accessing the GeNotes course and (b) the evaluation of the course as set out in the Genome UK Implementation Plan

**Recommendation 12:** NHS England should seek to collect data on when in cancer pathways genomic tests are being used. Initially each GLH should be asked to report back on where a small sample of tests from the Test Directory are being used in pathways to identify variation. Over time such information should be embedded within the NHS Genomic Medicine Service Dashboard. As part of its 1-3 year commitment on transforming cancer pathways to adopt genomic tests, NHS England should seek to co-ordinate the range of initiatives already under-way and work with key actors such as the medical royal colleges, professional bodies and NICE to ensure that guidelines for major cancer pathways are regularly updated to reflect the latest evidence on genomic testing.[115] Over the three year timeline such work should have a set aim to move services closer towards a 'genomics first' approach to cancer testing

[115] Work is underway to do this through the Cancer Genomic Oversight Group, which is co-chaired by the NHS Genomics Programme and NHS Cancer Programme

# CONCLUSION

Advances in genomic testing present opportunities for transforming our ability to diagnose and treat cancer earlier, improving patient outcomes and saving lives.

It is nearly six years since the NHS Genomic Medicine Service was launched to take advantage of such innovation and more closely embed and scale genomic testing for cancer within the NHS.

A number of early commitments made through the 2022 national *Accelerating Genomic Medicine* Strategy have been delivered – at least in part. There are positive signs that testing capacity and workforce capability is increasing. Newly established regional Genomic Networks of Excellence should also help ensure that new evidence is built which can support the adoption of new innovations to improve the operation and performance of the system.

However the scale of the challenge facing the NHS Genomic Medicine Service as it approaches its sixth anniversary should not be underestimated. Whilst it is difficult to accurately assess delays on turnaround times for tests, published data, correspondence and anecdotes indicate a system, like the wider NHS in which it operates, under strain. With more and more tests being added to the National Genomic Test Directory and as innovation in this area accelerates, whether capacity, workforce, processes and skills can keep-up is uncertain. Services are still suffering from disjointed technology and systems, patient and public engagement work is difficult to appraise and the use of genomic testing within clinical pathways is variable.

To deliver more genomic testing for more cancer patients will now require concerted effort to change clinical pathways to embed genomic testing earlier, working more flexibly to expand and share testing capacity, building and learning from workforce educational initiatives and greater transparency on service performance.

It will also require the next Government to prioritise investment in the service to match the ambitions that it has been set and support wider efforts at a greater preventative shift in healthcare system delivery.

FH | FUTURE HEALTH