Sequencing Success: Genomics for cancer research and care

Cancer Research UK's position on genomic research and healthcare in the UK

November 2024

Full Report

CANCER RESEARCH UK

Contents

About this report

Reference

This report should be referred to as follows:

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Author

Lead authors of this report are:

Emily Eagles, Policy Advisor

Alice Hilborn, Policy Advisor

Benjamin Jones, Policy Advisor

Joe Kiely, Policy Advisor

Helena Kipling, Health Innovation Policy Manager

Contact

Comments, questions or feedback should be sent to *emily.eagles@cancer.org.uk.*

About Cancer Research UK

We're the world's leading cancer charity dedicated to saving and improving lives through research. We fund research into the prevention, detection, and treatment of more than 200 types of cancer through the work of over 4,000 scientists, doctors, and nurses. Over the last 50 years, we've helped double cancer survival in the UK and our research has played a role in around half of the world's essential cancer drugs. Our vision is a world where everybody lives longer, better lives, free from the fear of cancer.

Our values

Our values help guide our behaviour and culture in an ever-changing world, building on the best of what we do today and what we aspire to be in the future. They unite and inspire us to achieve our ambitious plans and our mission of beating cancer, together.

1. Introduction

Over the last decade, genomics has led to many exciting innovations in cancer diagnosis and treatment and is becoming increasingly important for cancer research. Genomics is central to building our understanding of the fundamental biological nature of cancer and is the key to developing more precise diagnostics and treatments for cancer. Genomics also provides a powerful under-pinning for many areas of innovation, which could transform cancer research and care. By looking at the molecular biology of an individual tumour, cancer can be diagnosed more accurately, and more effective targeted treatments can be developed with reduced adverse side effects for patients.

This policy report has been shaped by insights from engagement with people affected by cancer, researchers, health and industry professionals, policymakers, academics, and charities. CRUK would like to acknowledge and thank those who gave their time and expertise through interviews or participation in online surveys. CRUK would like to extend special gratitude to the patients and people affected by cancer who shared their views through our engagement panels, online surveys, and involvement in an engagement event co-facilitated with useMYdata.

Our recommendations can be found throughout this report. The full list can be found in the *Sequencing Success: Executive Summary.*

2. Landscape

2.1 What is genomics?

Genomics refers to the study of someone's genome. A genome is made up of an individual's complete set of DNA, and sections of DNA are called genes. Cancer is a disease that results from changes in genes that impact on how a cell functions. These changes, also known as variants, can be acquired during someone's life or inherited.

Somatic Variants	Most cancers are caused by these variants that can be acquired at any point during a person's life and are only present in cancer cells.
Germline Variants	These are variants that are present in all cells of the body and can be passed on. In around 5% of cases, an individual has germline genetic variant(s) that substantially increases their chance of developing cancer[1].

Genomic medicine uses the insights from someone's genomic testing to inform their clinical healthcare. There are two main ways in which genetic testing is used for cancer. This may either be through looking at variants in the cancer cells – known as somatic testing or through looking at someone's germline DNA – known as germline testing. In a cancer context, genomics can be used to indicate prognosis, offer a more complete diagnostic picture, or indicate whether there is a clinical trial that could benefit a patient. For some patients, genomics can also be used to determine eligibility for treatments targeted to an individual's tumour type and could lead to better outcomes for that patient. Genomic testing can also play an important part in managing care for individuals and family members who have a higher inherited risk of developing cancer. It can also inform how a patient might respond to treatment or whether they are at risk of adverse impacts from certain treatments.

Advances in genomic testing have huge potential to drive the next cycle of research and innovation, enabling us to identify new genetic variations and markers associated with cancer. This knowledge will enhance our ability to assess cancer risk, detect it earlier, and discover new preventative measures, as well as accelerating the development of more precise and less harmful diagnostics and treatments for people with cancer.

2.2 The picture across the UK

In 2020, the UK government published a national 10-year strategy for genomic healthcare titled 'Genome UK: the future of healthcare'[2] with the vision to solidify the UK's position as a world leader in discovery and translational genomic research. This was followed by the development of shared commitments for UK-wide implementation between 2022 to 2025. This strategy recognised that the rapid pace of discovery driven

by cancer genomics requires a health system - supported by nationally coordinated, harmonised genomic services - that is agile and can quickly deliver testing and translate findings into clinical impacts. Each nation holds devolved responsibility for strategy implementation.

Progress has been made across the UK but at different paces, so genomics services vary depending on where you live. These differences in service development are creating difficulties with collaboration and sharing of best practice, learnings, and data between the nations. Our concerns about equity of access to genomic health services within and between the nations of the UK are explored in detail below and throughout this statement.

The following figures aim to illustrate the basic structure of the genomic systems in each nation of the UK (e.g., the Genomic Medicine Service in England) and show simplified versions of very complex systems. There are brief descriptions of the roles of key actors in England, Scotland, Wales and Northern Ireland, and the overarching system components that comprise these genomic healthcare systems.

Figure A: Genomics in England

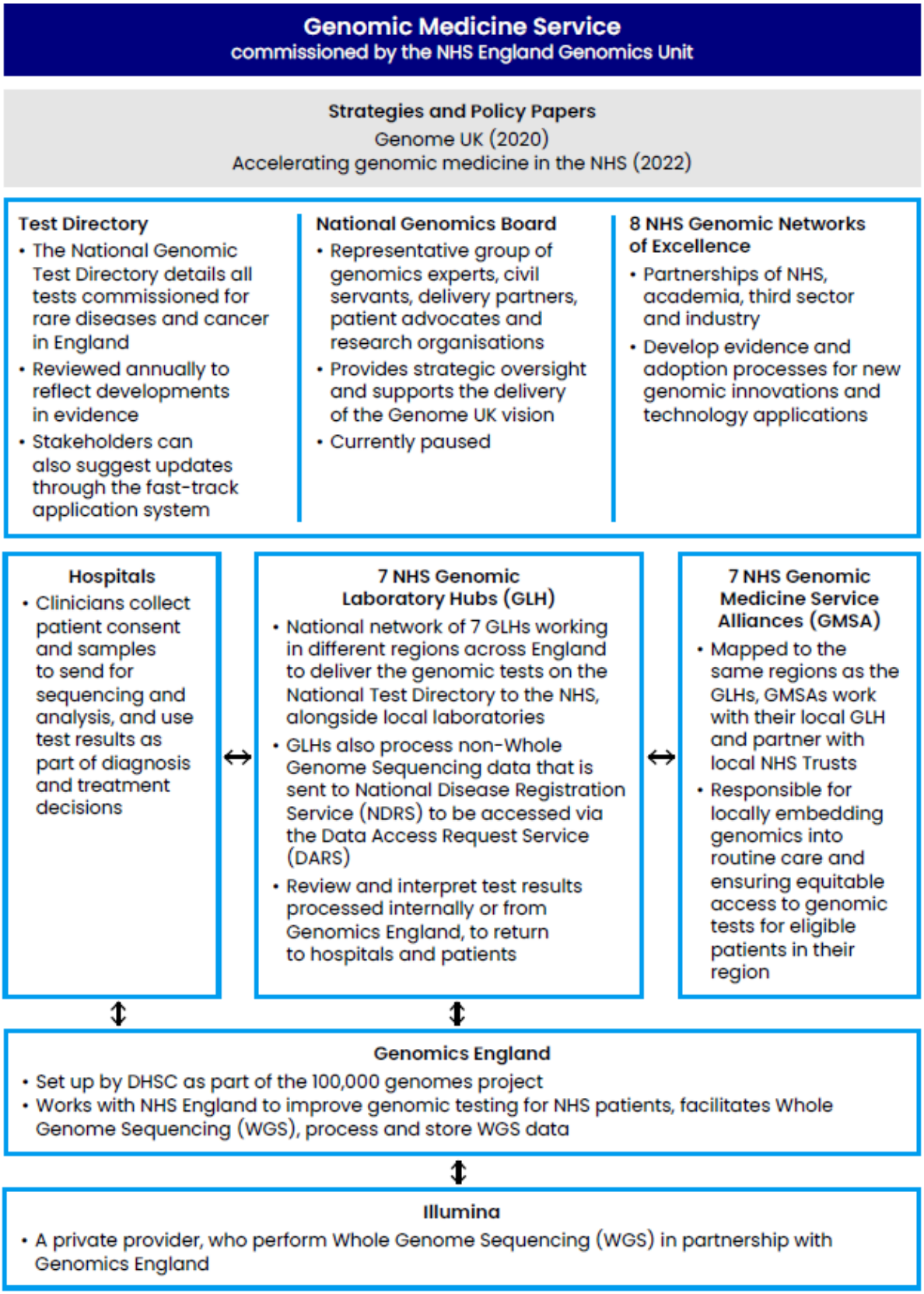


Figure B: Genomics in Scotland

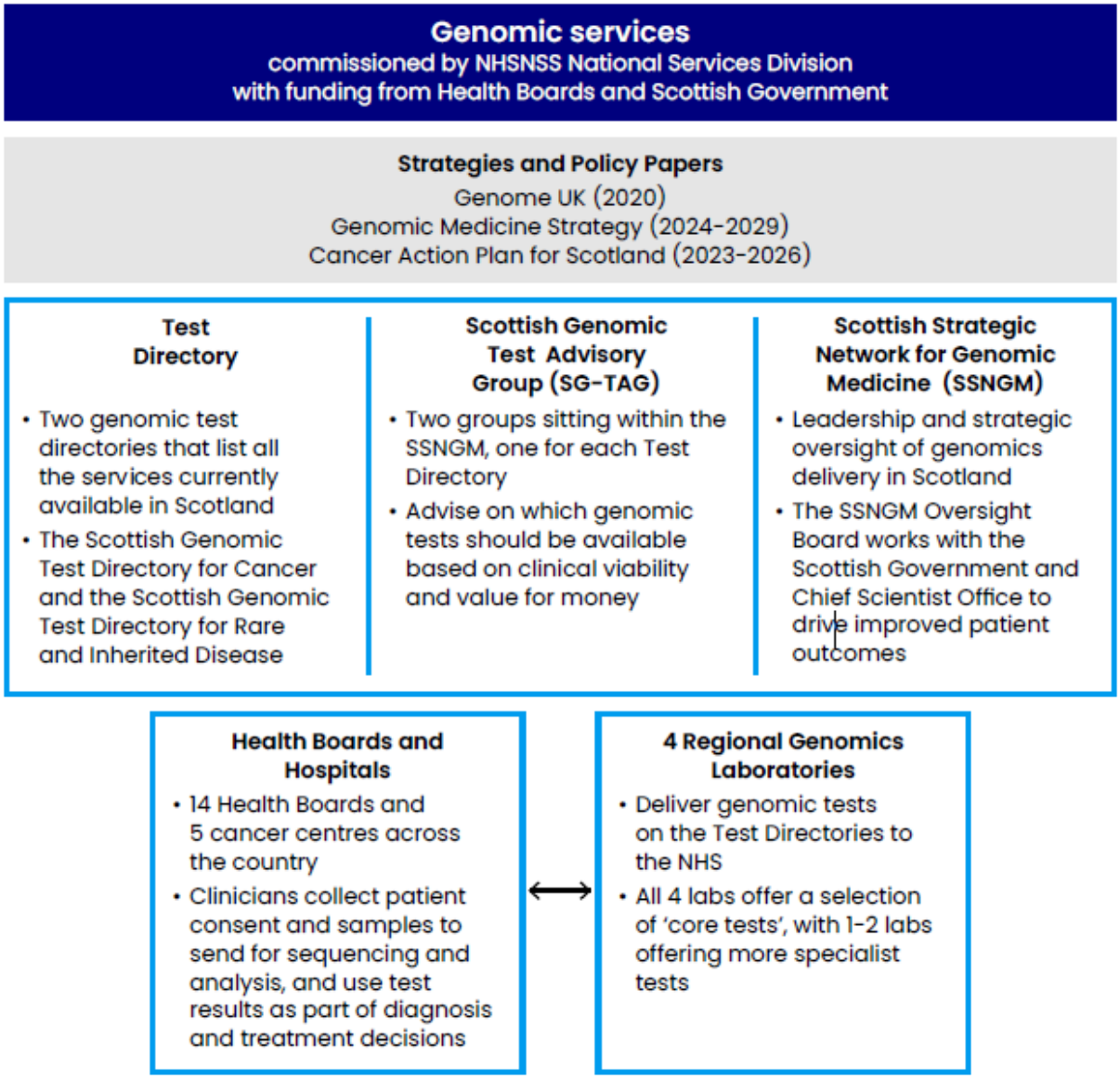


Figure C: Genomics in Wales

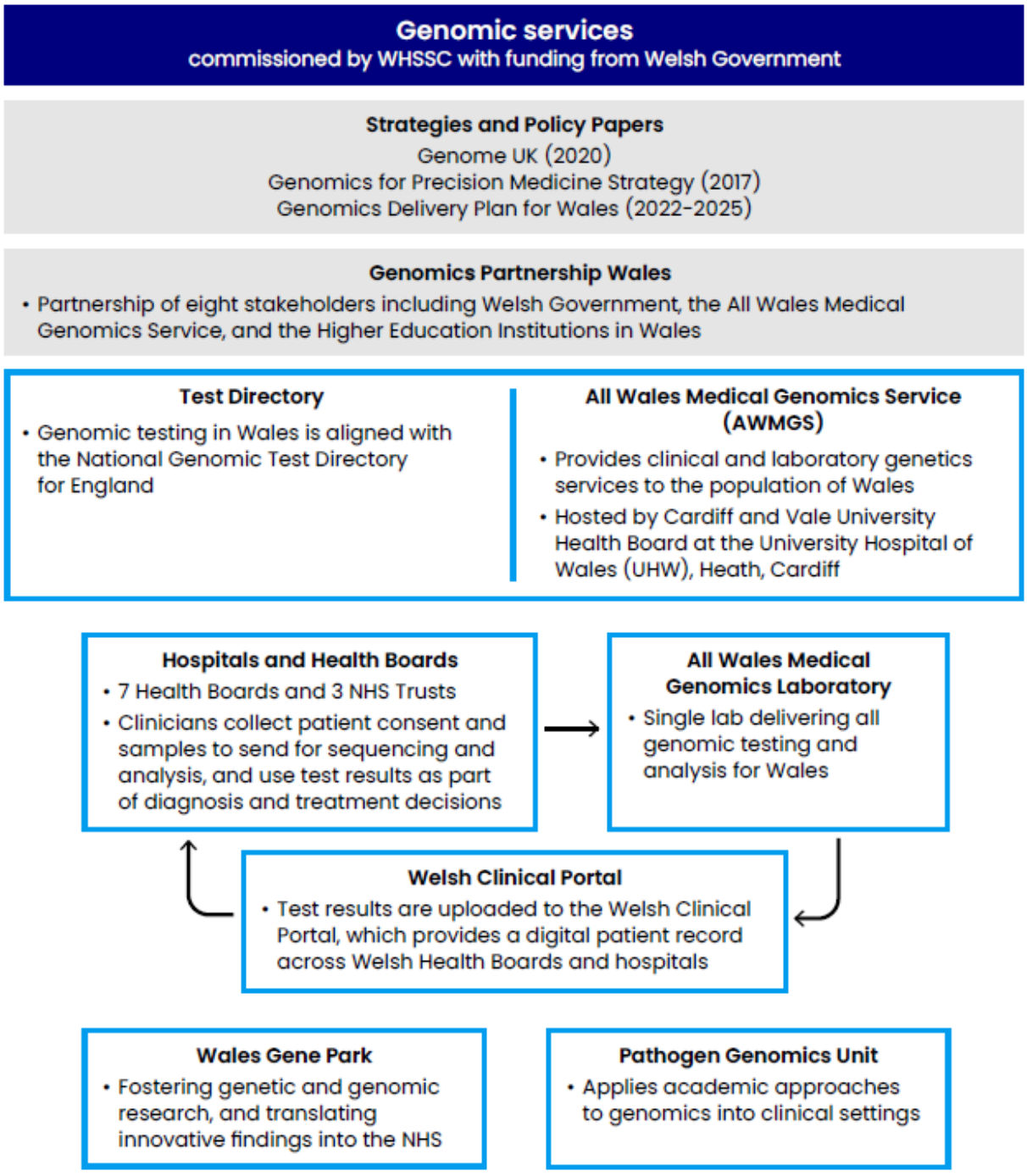
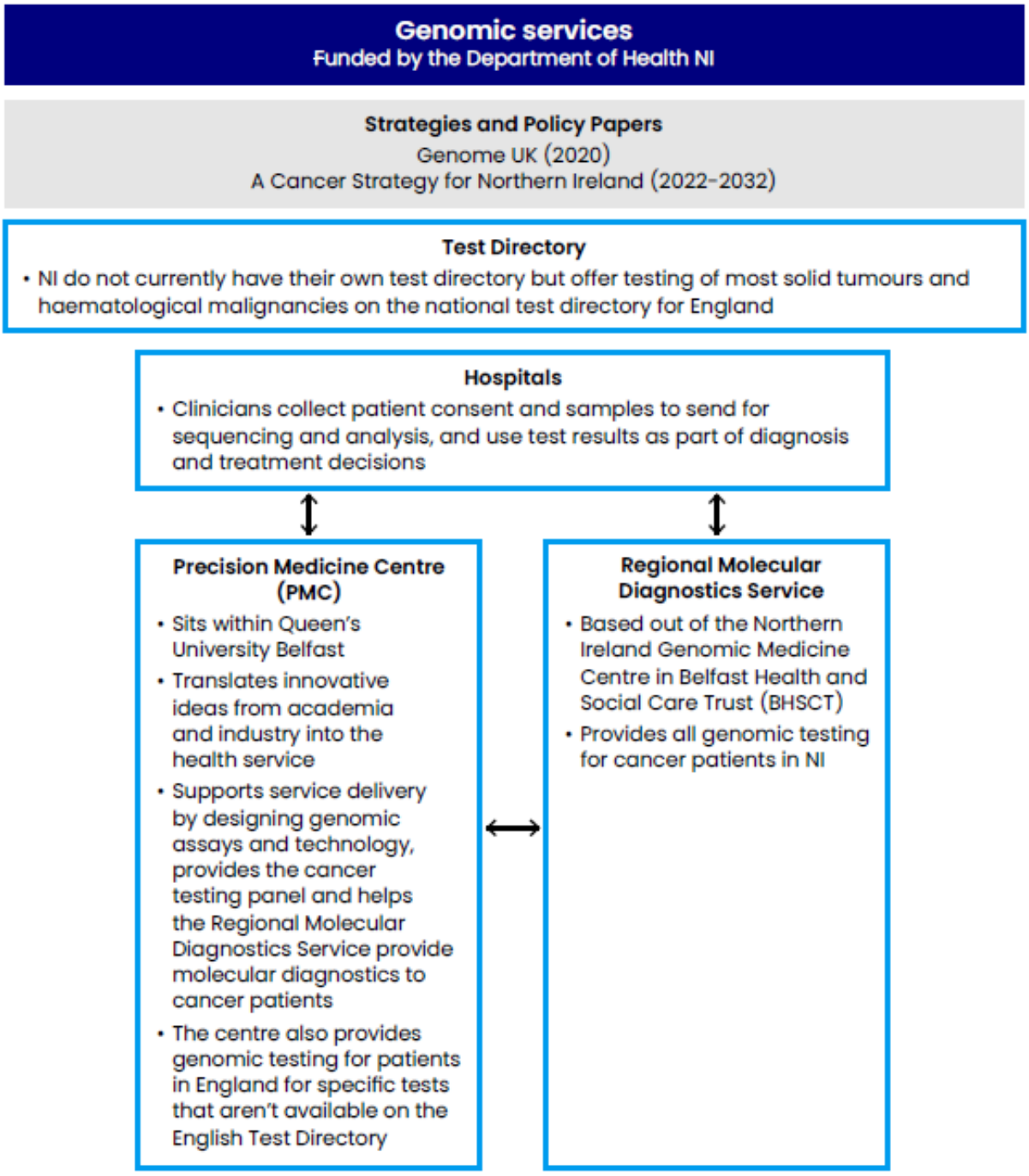


Figure D: Genomics in Northern Ireland



3. Mobilise genomics to detect and diagnose more cancers, earlier

"After [receiving a] colon cancer diagnosis I asked for testing as my father died from it. I was told this was not necessary. Four months later after being diagnosed with endometrial cancer I asked again and was referred. Later, I was diagnosed as having Lynch Syndrome."

CRUK Patient Involvement Network member

Beyond treatment, advancements in science present the possibility of using genomic insights to improve the early detection, and even prevention, of some cancers. As the front door to the NHS, and with a commitment to offering person-centred care, general practitioners will play an essential role in the future of genomic interventions delivered at the start of the patient pathway. But these exciting developments are often playing catch-up with a health system not yet prepared to implement these innovations. This chapter will explore how the governments and health services of each of the four nations can work to provide the early detection initiatives patients need today and prepare the system for the successful adoption of the next wave of genomic innovations. This will require:

1. **A more strategic approach to the testing, monitoring and, evaluation of current cancer genomic risk testing programmes** – improving our understanding of who needs to be tested and the effectiveness of surveillance programmes.
2. **Future-proofing general practice for genomic medicine** – providing the right education, mix of skills and systems so staff feel empowered to support patients.

3.1 Genetic testing for cancer risk

The potential of genomics for the early detection and prevention of cancer is already being seen through testing for germline genetic variants that substantially increase a person's risk of developing cancer. Most cancers are caused by gene faults that happen during our lifetime, but some people have an increased risk of particular types of cancer because they have inherited a version of a gene that does not function normally. Not everyone who inherits these genes will develop cancer, but they are much more likely to than other people and they are more likely to develop cancer at a younger age.

5 – 10 in every 100 cancers

Are linked to an inherited gene variant[3].

There are many pathogenic variants which increase someone's lifetime risk of developing cancer[i]. For this section we will focus on the two most common and view these as use cases. Firstly, *BRCA1* and *BRCA2* variants which increase the risk of developing some cancers including breast, ovarian, pancreatic, and prostate cancer. Secondly, a group of variants called Lynch syndrome. Up to 70% of people with Lynch syndrome go on to develop bowel cancer, most before the age of 50[4]. Women with Lynch syndrome will also have a lifetime cumulative risk of endometrial cancer of between 40 and 60%[5].

It is estimated that

1 in 400

People have Lynch Syndrome[6].

Around

1 in 400

People have a *BRCA1* or *BRCA2* mutation[7].

Genetic testing can help in the identification and management of individuals and families at high risk of developing cancer. This is either through testing those newly diagnosed with some cancers, those with a family history of some cancers, or by testing family members of those who have been found to have a pathogenic variant (known as cascade testing).

Good progress has been made in testing for variants that confer high risk. For example, in England, almost all people diagnosed with bowel and endometrial cancer receive the initial test to check for Lynch syndrome – 94% on average between 2021-2023 which is up from 47% in 2019[8]. Similarly, NHSE have recently rolled out *BRCA* testing for the Jewish population, who are more likely to have a *BRCA* gene variant[9,10]. There is still work to be done to ensure these programmes deliver for as many people as possible across the UK. For example, Bowel Cancer UK highlighted ongoing challenges across all four nations with proactively testing family members for Lynch syndrome before they develop symptoms and offering routine surveillance colonoscopies for those identified as having the condition. In Wales, Scotland, and Northern Ireland a lack of data collection on the uptake of surveillance colonoscopies makes it difficult to evaluate the success of these programmes.

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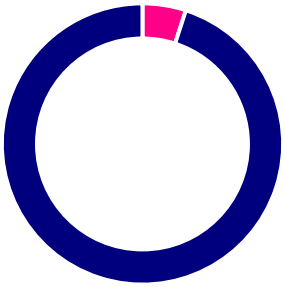
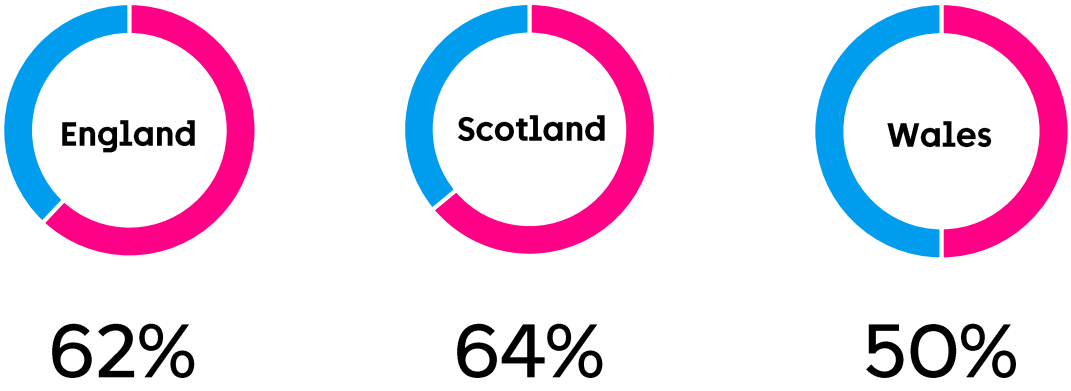
[i] Other common inherited cancer gene variants include: Li-Fraumeni syndrome (LFS), PTWN Harmartoma tumour syndrome, Familial adenomatous polyposis (FAP), MUTYH associated polyposis (MAP), Peutz Jeghers syndrome (PJS), Juvenile Polyposis syndrome (JPS), PALB2 gene, Von Hippel Lindau syndrome (VHL), Tuberous sclerosis (TS), Birt-Hogg-Dube syndrome (BHDS), Multiple endocrine neoplasia (MEN) type 1 and 2, RB1 gene, Familial atypical multiple mole melanoma syndrome (FAMMM), and Hereditary papillary cancer.



95% of people with Lynch syndrome have not yet been diagnosed[ii]

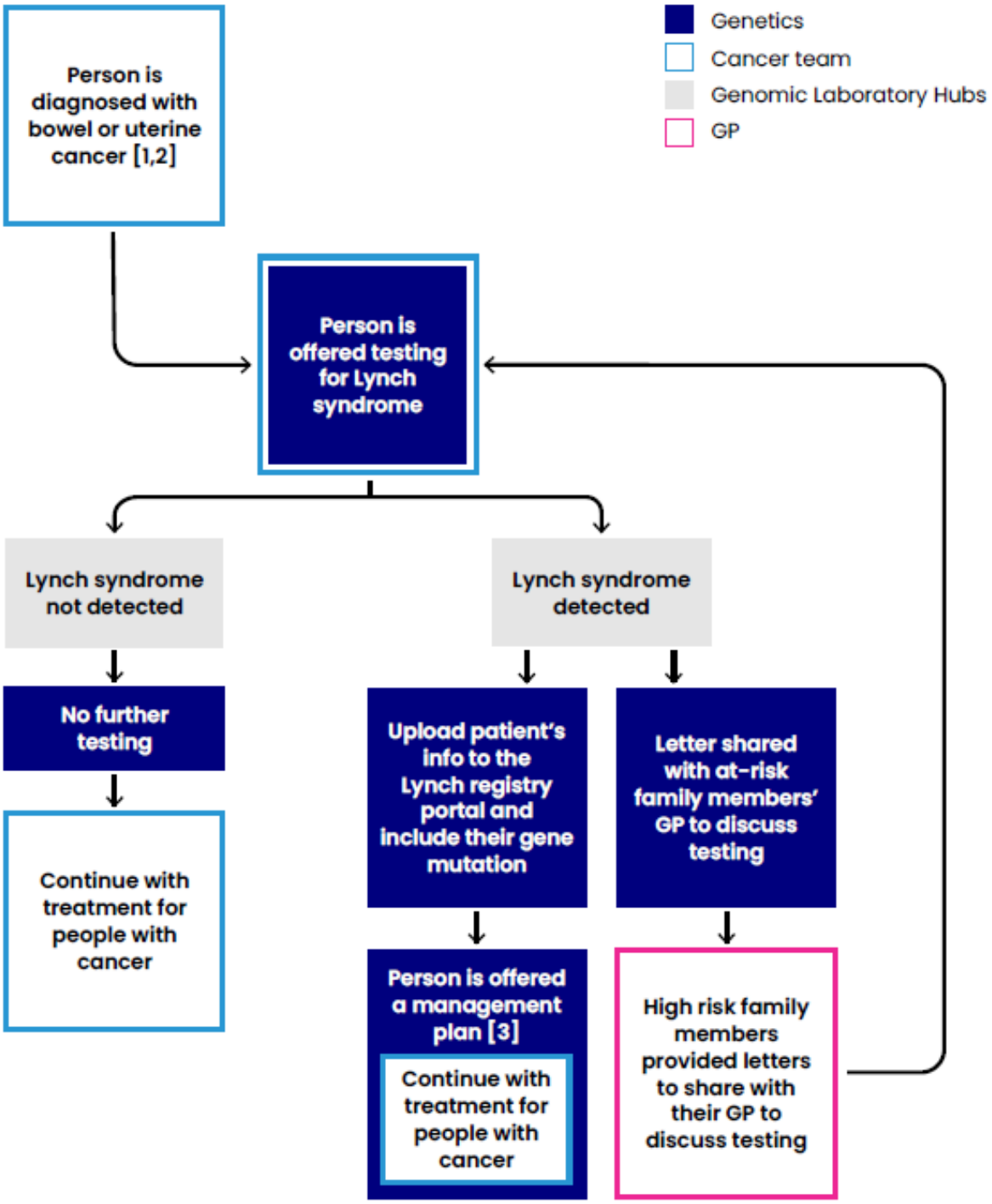
Perhaps most concerningly, even though 175,000 people are estimated to be affected by Lynch syndrome in England, only an estimated 5% of those people have been diagnosed, meaning 95% of people with Lynch syndrome have yet to be diagnosed or receive potentially life-saving risk reduction interventions such as surveillance colonoscopy[11].

Figure E: Percentage of UK health authorities providing close relatives with letters for their GP regarding testing for Lynch syndrome[12]



[ii] Bowel Cancer UK. 2024. Finding the missing 95%: Unlocking the potential of Lynch syndrome services

Figure F: Testing for Lynch Syndrome



1. Uterine cancers can be of two types: endometrial cancer (common) and uterine sarcoma (rare)
2. Patients with other Lynch-related cancers may also recieve testing as appropriate including epithelial ovarian cancer, ureteric cancer, transitional cell cancer of renal pelvis, cholangio carcinoma, small bowel cancer, glioblastoma, endocervical cancer, multiple tumours, prostate, gastric and pancreas.
3. Management could included surveillance colonoscopy, screening for Helicobacter pylori, daily aspirin, and risk reduction surgery such as hysterectomy with bilateral salpingo-oophrectomy.

To see real progress on catching as many people as possible with an inherited cancer risk we need to get the testing, monitoring and evaluation of all genetic cancer risk programmes right. This means ensuring everyone who is at risk is offered prompt testing and those confirmed to have variants are offered appropriate follow-up actions such as surveillance. Appropriate data must be recorded to monitor these programmes.

Not all the inherited genetic drivers of cancer are known, and even when they are, not all genes have an appropriate surveillance intervention. But investing time today in having a robust approach to testing, monitoring and evaluation will ensure a system well prepared to adopt future programmes as our knowledge and understanding continues to develop.

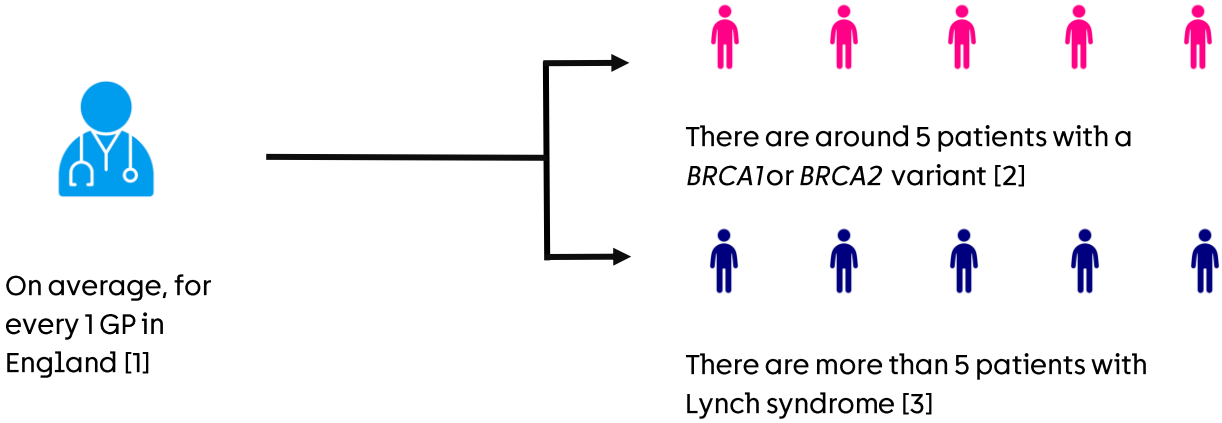
Policy Recommendations

1. Across all four nations, health services should commit to reporting on all cancer genetic risk testing and surveillance programmes in a similar way to reporting for screening programmes.
2. The health services in Scotland and Northern Ireland should set up a national directory of Lynch syndrome testing to improve data collection and monitoring.
3. The health services in Scotland, Wales and Northern Ireland should look to follow the example of NHS England and set out an implementation plan for Lynch testing and surveillance.

3.2 Genomics in general practice

Genomics is not only set to have a significant impact on general practice in the future, it already has a profound effect today.

Figure G: Patients with *BRCA* variants or Lynch syndrome per GP in England



1. Based on the total number of FTE GPs in England in April 2024, Key general practice statistics and insights, Royal College of General Practitioners.
2. Based on the estimated number of people living with a faulty BRCA1 or BRCA2 gene. Calculated by Cancer Research UK, https://www.cancerresearchuk.org/about-cancer/causes-of-cancer/inherited-cancer-genes-and-increased-cancer-risk/inherited-genes-and-cancer-types#Keyreferences0 , Accessed August 2024
3. Based on the estimated number of people living with Lynch syndrome, Bowel Cancer UK, Finding the missing 95%: Unlocking potential of Lynch syndrome, 2024

Patients with inherited cancer gene variants rely on their GP to recognise family history, refer them for testing and manage the care of those who are diagnosed. But GPs are already facing growing pressures, with rising demand and staffing issues meaning practices are under more strain than ever before. General practice will be essential to the delivery of genomic medicine, but to get the most from this workforce they need to be supported and well-resourced.

Alongside formal cancer risk programmes, growing public and commercial interest in genomics including the expansion of direct-to-consumer (DTC) testing is placing additional demands on an already stretched system. Most consumer genomics tests do not test for variants most likely to cause disease, known as high penetrance variants, raising questions over their clinical value to both patients and clinicians. In the Royal College of General Practitioner (RCGP) and British Society of Genetic Medicine (BSGM)'s genomic position statement, the organisations stated that healthcare professional should 'exercise caution when asked to offer, or provide, clinical expertise' about the results of these types of tests[13]. These tests present several concerns, relating to their clinical validity and utility, questions around quality control and data privacy, and test appropriateness[14,15,16]., because of this RCGP/BSGM's advice is to discount DTC results in favour of existing guidelines (e.g. family history and risk assessment, health lifestyle advice, or referral to specialist care).

Similarly, a lack of follow-on care and pathways for further testing and advice for

patients has been a frequent criticism of some DTC testing companies. Perhaps most challenging from a health system perspective is that the NHS, especially primary care, is unprepared in terms of capacity, pathways, or clinical expertise to adequately respond to demand that these tests might create for consultation, advice, and follow-on testing[17,18].

Alongside the potential impact of DTC testing, innovations like multi-cancer tests (MCTs) and polygenic risk scores could see a step change in the early detection of cancer. The ability to improve risk assessment and stratification is likely to drive more targeted screening programmes, as it will identify those at the highest risk of developing cancer. Primary care will be intrinsic to the success of many of these new genomic early detection initiatives.

General Practice remains the main point of access to the health service for most people, as well as having an important role in care coordination and prescribing. GPs also already possess many of the core skills related to genomic medicine, including performing risk assessments, managing complexity, and communicating information, including about risk, for example when discussing lifestyle changes or screening programmes. Genomics builds on these skills, but further education and advice will be needed to ensure the general practice workforce feel confident to take the most appropriate actions off the back of genomic information, particularly around risk, which can often be difficult to interpret.

GPs tend to be reactive in their learning and need quick, efficient, and easily accessible knowledge. GeNotes provides concise information to help healthcare professionals make the right genomic decisions at each stage of the pathway, it encompasses both information focused on the point of patient care as well as a knowledge hub to extend learning with an encyclopaedia of resources. GeNotes: Primary Care has recently been launched and aims to harness GPs' natural learning instinct by providing information on genomics 'just-in-time'. This programme must be continually monitored through the working group to ensure it is providing useful information in the most appropriate format for it to be taken up by practitioners.

A wide array of research has demonstrated the importance of clinical champions in ensuring successful implementation of innovation[19]. This was recognised back in 2008, when two GPs with a Special Interest (GPwSI) in Genetics roles were established following the UK Department of Health's white paper 'Our Future: Our Inheritance'[20]. Surveys of how GPs access information on genomics noted geographical clustering of awareness and response rates in areas where there are GPwSIs in genetics[21]. Since then the idea of leaders in primary care being able to champion the genomics agenda has been replicated, including primary care leads within GMSAs, and some Cancer Alliances now also recruiting primary care genomic leads. Creating more leadership roles in genomics, such as the expansion of GPs with an Extended Role (GPwER) in genomics should not only help drive change but also help to create career development opportunities for GPs, helping to boost retention.

As genomics continues to become routine in primary care, wider questions will need to

be answered on who should have accountability and responsibility for the integration of new genomic innovations and the continued upskilling of staff. With new local health structures in ICBs and ICSs, it will be important for genomics to be fully integrated into wider strategies for primary care at every level of system. General practice is facing considerable challenges, making the integration of genomic medicine more difficult. Increasing patient numbers, increasing consultations per GP, rising co-morbidity and a system lacking in resource and capacity making it difficult to prioritise innovation. Embedding genomics into primary care is essential, but without getting the basics right it won't have the impact it should. To truly future-proof general practice for genomic medicine, the governments and health systems across the UK should be thinking strategically about the incorporation of genomics into general practice, and primary care more broadly, formulating a plan of action on governance, workforce development and innovation adoption that understands the importance of the topic alongside the reality of the pressures health service is under today.

Policy Recommendations

4. The Genomics Medicine Service should continually monitor and evaluate the effectiveness of GeNotes: Primary Care, ensuring it's 'just-in-time' principles are delivering information, advice and guidance to healthcare professionals in the most appropriate and efficient way possible.

5. NHS England, alongside the Genomics Education Programme and Royal College of General Practitioners, should expand the number of GPs with an Extended Role (GPwER) in Genomics, aiming to have at least one per region.

6. To future-proof the system, governments and health services across the UK should be taking a strategic approach to the incorporation of genomics into general practice, seeking advice from key stakeholders to create a plan covering governance, workforce development and innovation adoption both today and into the future.

4. Design effective, efficient genomic systems with a strong research culture

As genomic healthcare services in each nation of the UK have developed and substantial investment has been put into genomic research, recognition of the value in genomic testing and demand for genomic services has grown in parallel. However, there are a variety of structural, strategic and cultural challenges that must be navigated to deliver an effective and integrated genomic service. This chapter looks at how DHSC, NHSE and their devolved equivalents can address these challenges by working to provide patients with opportunities to benefit from high-quality routine genomic care and seamlessly integrating genomics research into healthcare by increasing research accountability and improving the visibility of research's benefits.

This will require:

1. **Further integration of genomics and genomic research into routine care pathways** – developing clear guidance for clinicians that embeds genomic research and research in healthcare.
2. **Commitment to building resilient genomic systems**– ensuring delivery teams are adequately equipped to return patient results in a clinically useful timeframe, improving equity in access to services for patients across UK nations and on different cancer pathways.

4.1 Genomic testing in secondary care

Genomic testing holds significant opportunity to pinpoint disease risk, offer diagnostic insights, and determine eligibility for targeted treatments that positively influence outcomes for cancer patients. There are currently issues when it comes to delivering genomic testing comprehensively and effectively in cancer patient pathways across the UK, particularly now that genomic testing is being further rolled out as part of standard care.

National pathway guidance

Genomics has yet to be fully embedded into standard care across the UK, as the guidance around clinical pathways and turnaround time standards is still being developed for most cancer sites and solid tumour services. In England, for lung cancer and metastatic colorectal cancer respectively, the National Optimal Lung Cancer Programme[22] and Faster Diagnosis guidance[23] have defined turnaround times for genomic test results but as discussed above, capacity issues in the Genomics Laboratory Hubs (GLHs) mean these aren't currently being met.

Similarly, the provision of genomic healthcare services in Scotland is limited by the absence of clear and clinically meaningful pathway guidance. In Scotland, stakeholders

noted that there are several medicines approved by the SMC that Scottish patients can't access (although patients in England can) because the necessary companion diagnostic tests aren't yet available on the SSNGM Cancer Test Directory. It's positive to note in Wales that genomic testing has been added to the National Optimal Pathways for various cancer sites.

Testing capacity and backlogs

In each nation low pathology capacity, old and slow IT systems, skilled staffing gaps and inadequate infrastructure investment are contributing to a difficult environment where laboratories are struggling to process the increasing numbers of referrals in a clinically useful timeframe[24]. As highlighted in the Darzi review, in 2024, more than 35,000 genomic tests were being completed each month in England, but only around 60 per cent of these were reported on in time[25].

Box B
Types of testing

Single Panel Testing: Looks at a single gene, often when a clinician wishes to confirm or rule out a specific diagnosis or there is a family history of a certain genetic mutation.

Large Panel Testing: Looks at multiple genes at once. In the cancer context, relevant genes for a specific cancer type may be grouped together, or some laboratories use large panels with around 500 cancer genes as a standard.

Whole Genome Sequencing (WGS): DNA sequencing of close to your entire genome and can capture variants that might be missed with panel testing.

Whilst genomics is driving progress in cancer diagnosis and treatment, concerns remain around how equitable patient access to genomic testing is. Within and between each UK nation, patient experience differs based on location. The time it takes for genomics labs to return test reports to clinicians varies by geography[26,27,28,29], meaning not all patients receive their results back in time to be clinically useful. Also, the specific genes included on testing panels vary regionally and between the nations, with the development of panels at the discretion of the testing laboratory. It is important that where service delivery methods might vary, the service itself must remain equitably accessible to all patients across the UK where applicable.

Worryingly, stakeholders we spoke to said that in some areas of England it is still common practice for clinicians to send samples for single gene panel testing at their local pathology lab, instead of or before sending this to their local GLH. Reasons for doing these included concerns about turnaround times via the GLH route or sticking to pathways that existed before the GLHs were created. This causes several issues- as stakeholders identified, the NHS can end up double paying for testing, or in some instances pathology labs aren't paid, and there can be instances of 'sample ping pong' between local labs and GLHs, which can cause delays in turnaround times. Similar issues with pathology

capacity exist in Wales, which can lead to the 14-day laboratory turnaround time target[30] being missed and knock-on delays to the discussion of patient results at MDT meetings. Crucially, this can impact the care that patients receive, which we explore further below.

In Northern Ireland, it's positive to hear from stakeholders that for most patients, test results are reported and linked to hospital Electronic Care Records within 14 days. This means clinicians can assess and action findings quickly. However, patients with rarer cancers are more likely to experience delays as their samples typically have to be sent for testing in labs elsewhere in the UK. In addition, not all necessary biomarker tests or companion tests are available on the standard pan-cancer diagnostic test panel used. Therefore, in some cases samples must be sent to specialist labs outside of Northern Ireland. On top of the additional logistical hurdles of sending samples to labs elsewhere in the UK, stakeholders noted that on occasion these labs have to deprioritise processing and reporting on samples from Northern Ireland and Scotland as they address their own local backlogs, which can cause delays.

Large panel testing and WGS are particularly important in facilitating clinical trial recruitment, especially for patients with rare cancers. Patients typically do not receive genomic sequencing or profiling for clinical trials (either via new testing or additional reporting from existing tests) until after they have exhausted all the standard of care treatment options that are available. By that stage, most patients have deteriorated to the point they are not well enough to receive further treatment. Conducting multiple diagnostic investigations can also be stressful and uncomfortable experiences for patients. It also costs the health system more.

In contexts where Whole Genome Sequencing (WGS) testing is regularly used in England – primarily children's and young people's cancers and some rare cancers - there is a significant backlog in reporting, despite improvement described by stakeholders over the last year. Looking more comprehensively at your whole genome, WGS testing can capture variants that might be missed by more targeted approaches, but it isn't necessary in all cases; especially as this more detailed testing takes longer to interpret and report and can increase decision times for patients. WGS testing is also critical in informing research, and improving our understanding the nature of rarer cancers, such as brain cancer, and hard to treat tumours. As testing capacity is already stretched, NHSE's ambition to become the first national health service to offer whole genome sequencing as part of routine care, is unsustainable in the current environment. Whilst good progress is being made in developing a testing offer that includes more medium and larger panel testing alongside single gene panel and WGS, to fully meet this ambition, issues surrounding IT infrastructure and shortages in the workforce delivering genomic sequencing must be urgently addressed.

Service design

Some interviewees suggested that many of these delivery issues are rooted in the design of the genomic medicine systems of the UK. The genomic delivery structure in England and previous testing strategies were built from the foundations of the 100,000 Genomes

Project and are now struggling to meet the testing demands for both research and standard of care. In practice, differing approaches to testing by different labs has also led to inconsistent reporting templates within and between nations of the UK. We've heard from clinicians that test reports can often be difficult to interpret and that the clinically relevant information is hard to find, further complicating decision making on treatment. Alongside this, there is variation in the regional interpretation of the Genomics Unit's national strategy, meaning the testing practices used may also vary from patient to patient[31]. Stakeholders in the research community noted that large panel testing is preferable from a research perspective as more data is collected than with single panel and it facilitates matching patients to precision medicines as part of care delivery[32].

Problems caused by the inconsistent integration of genomics into health service cancer pathways are having a knock-on effect on patients' experience. When clinicians send off patient samples, long waits for test results can cause delays in starting treatment plans, choices made to start an alternative treatment during the waiting period[33,34,35], missed opportunities to participate in clinical trials and confusing messaging.

How can these issues be resolved?

It's encouraging to see work underway to improve genomics testing delivery, and recognition that a more conscious approach is required to address performance issues. For example, the rolling publication of GLH testing turnaround times and inclusion of turnaround times in GLH contracts for the 2025-2026 financial year will provide welcome accountability. As the GLHs have already been working at maximum capacity to meet service pressures, it is positive that linking turnaround times targets to commissioning will be accompanied by support and guidance from the Genomics Unit on achieving the targets.

This should be a consultative process with the GLHs, and not hold back those who are excelling. Support must be directed to those who need it most, with consideration of where additional resources will be needed to meet targets. There is also an opportunity for increased sharing of learning and best practice between GLHs, building on existing examples, such as sharing testing panels, to boost overall performance. Any future developments which further integrate commissioning and performance management should reflect the role of genomics across the whole cancer pathway, balancing testing requirements for routine care with those of research. It will be important for Integrated Care Systems, who sit on regional Genomics Medicine Boards, to be involved in this process.

It is positive to see that the plans outlined in the recently published 'Genomics in Scotland Year One Implementation Plan' to build genomic testing into end-to-end clinical pathways are in progress. It's encouraging that work is underway from the Scottish Strategic Network for Genomic Medicine (SSNGM) to drive the implementation of seven prioritised pathways in 2024, including for prostate and thyroid cancers, and that the SSNGM Transformation Team have identified an opportunity to improve service delivery models as part of a new Genomics Laboratories Transformation Delivery Plan for 2024-

2026[36].

We welcome news that the NHSE Genomics Unit is mapping and developing the genomic cancer patient pathways for 11 tumour groups to ensure that future turnaround times guidance aligns in a clinically meaningful way. As these pathways are implemented, a holistic approach is needed to make sure patients have access to relevant clinical trial opportunities. We also welcome the establishment of the new NHS Genomics Ethics, Equity and Legal Advisory Group and its scope to consider the full patient pathway, as well as developments in research and innovation.

These issues must be urgently addressed, as there is a risk that as some locations progress and others lag behind, unwarranted variation within and between the four nations of the UK could worsen, exacerbating inequalities.

Policy Recommendations

7. DHSC must increase support for the NHSE Genomics Unit and Genomics England to build pathology and laboratory testing capacity. The Genomics Unit should consider introducing a tiering system for Genomics Laboratory Hubs (GLHs) to provide additional targeted support for those struggling to meet turnaround times and facilitate increased shared learning between GLHs to help resolve issues.

8. Scottish Government must prioritise making the changes outlined in the Year One Implementation Plan for 2024-2025, so patients can begin seeing service benefits as soon as possible.

4.2 Integrating genomics research and care

"... our NHS is a unique environment that researchers and drug developers should want to conduct clinical trials in. We should be making the most of this massive opportunity, which would be good for patients, good for the NHS and good for the economy."

Labour, *Labour Party Manifesto 2024, 'Build an NHS fit for the future'*[37]

Integration of genomics research into healthcare delivery is crucial if we want to our patient experiences and outcomes. Despite significant investments in initiatives like the 100,000 genomes project, these pilot programmes have not effectively scaled into nationwide healthcare solutions in the UK. Instead, an artificial boundary has emerged between genomics research and healthcare, reflecting a broader issue within the health service where research is often seen as a 'nice-to-have' by health leaders[38]. This separation is particularly counterproductive in genomics, where interpreting genomic tests often involves both the exploratory nature of research and the direct benefits that clinical care aims to provide patients.

While it is understandable that service leaders prioritise immediate care needs and acute pressures, neglecting research can result in people missing out on potentially life-

saving treatments. The benefits of biomarker-based clinical trials are clear, including improved treatment response rates, patients living for longer without their cancer getting worse, and improved overall survival[39,40,41]. Targeting drugs to specific genomic profiles also reduces the side effects experienced by people with cancer and improves cost-effectiveness for the health system by minimising the use of ineffective treatments[42]. Rather than being viewed as competing with care delivery, genomics research should be embedded into clinical practice and regarded as a potential solution to the challenges facing the health service.

Clinical trials play a crucial role in evaluating the safety and efficacy of new diagnostics and treatments and have been instrumental in doubling cancer survival in the past 50 years[43]. By stratifying patients according to their genetic profiles, these trials enable more precise analysis of treatment responses and outcomes between patient subgroups. Research can also help us understand how different genetic variants affect an individual's response to existing treatments, which enables us to find new, and often improved, uses for existing medicines (see Box A).

Box A

Case Study: DETERMINE trial

Led by the University of Manchester and sponsored and managed by Cancer Research UK's Centre for Drug Development

DETERMINE is the first UK national precision medicine trial for rare cancers. The trial is exploring whether treatments that are approved for use in some cancer types could be used to treat other types in patients who have the same genetic alterations. The trial is open to adults, teenagers, and children. Drugs which show promising results will be accelerated for Cancer Drug Fund assessment and NHS approval.

Pharma companies can provide drugs off-label and free of charge to patients where there is a potential benefit through compassionate use requests. However, managing these requests is resource intensive, and outcome data is not routinely collected. DETERMINE offers a unique solution to these issues.

The DETERMINE trial is currently open to recruitment at sites in all four UK nations, with additional sites across the Experimental Cancer Medicine Centres network (ECMC) in setup. The ECMC network is jointly funded by Cancer Research UK, Little Princess Trust, NIHR, and the Departments of Health for Scotland, Wales and Northern Ireland.

Our survey of the UK clinical research workforce showed that the health service's current approach to research stems from a lack of accountability and prioritisation at senior levels. While interviewees highlighted the positive impact of local research champions, they also acknowledged that relying influential individuals is unscalable and leads to disparities in access to innovative diagnostics and treatments. Establishing systemic support for genomics research is crucial to ensuring equitable access and improving overall healthcare outcomes.

The enhanced duty to promote research in the Health and Care Act 2022 is a step in the right direction, but the absence of data on R&D activity, including genomics research, across NHS bodies at any level makes it difficult to monitor or enforce this duty. National performance standards, like Cancer Waiting Times targets, are a critical part of monitoring performance and motivating improvements in clinical service delivery.

The current lack of data also contributes to poor visibility and understanding of research's benefits, which in turn leads to a lack of perceived incentives to conduct research[44]. It is widely recognised that clinical trials offer patients a broader range of treatment options, especially those with rare or CYP cancers where market failures have resulted in fewer approved treatments. As well as providing a potential lifeline for patients with limited options, clinical trials also bring additional, often invisible, benefits to healthcare services. These include improved health outcomes, better staff recruitment and retention, and access to industry funded cancer medicines and treatment capacity.

To achieve successful integration, health leaders need a clearer understanding of the significant benefits that a well-supported research portfolio can bring. While there are pockets of innovation and best practice, a nationwide solution has not yet been found. In Scotland, a small number of Health Boards are utilising the finance tools available through EDGE – a clinical research management system used widely across the UK - to track and raise awareness of the savings made by taking part in trials, including capacity savings and drug cost avoidance[45]. Initiatives such as these should be treated as pilots, with built-in evaluation to determine if and how such a model could be scaled across all Trusts and Health Boards.

Policy Recommendations

9. NHS England, NHS Wales and NHS Scotland need to act to better embed research into the design of clinical pathways, with consideration of relevant clinical trial biomarkers as they are rolled out for more cancer types. This is needed to enable a more joined up route between genomic testing, reporting, implications for patient clinical care and, where relevant, enrolment in clinical trials and to promote timely, equitable pathway implementation across each nation.

10. NHS England, NHS Wales, NHS Scotland and Health and Social Care N.I. should develop a coordinated set of metrics to monitor and evaluate commercial and non-commercial research engagement. This should include metrics that capture the uptake of genomics-enabled medical research in the UK, such as the proportion of patients recruited into a clinical trial following a positive genomic test result. The latter would provide evidence on whether the genomic medicine services in each nation are facilitating recruitment into clinical trials for innovative precision medicines.

5. Strengthen the genomic environment for children and young people

"Anything that can help us to better understand what is happening within the cells and what's unique to the child would be invaluable."

CRUK Children and Young People Patient Involvement Network member

This chapter outlines the applications of genomic healthcare for children and young people (CYP), and why genomic research is so important in helping us to better understand cancers affecting children and young people. There are lots of differences in CYP cancers compared to those which affect adults – from the types of cancers that affect this age group to the long-term effects of treatment – requiring a different, dedicated approach to treatment.

1,900

Children get diagnosed with cancer in the UK each year[46].

2,300

Young people get diagnosed with cancer in the UK each year[47].

In the UK, children's and young people's cancers account for around 1% of all new cancers each year[48,49]. Cancer remains a leading cause of death for children and young people in the UK, which highlights the significant impact of this disease within these age groups[50]. Yet, the focus when developing treatments for children and young people has traditionally been on repurposing drugs that have been approved for adults. Consequently, there are only two drugs on the market approved exclusively for children with cancer by the European Medicines Agency and only five by the U.S. Food and Drug Administration[51]. Between 2008 and 2022, only 16 anti-cancer new molecular medicines with a paediatric indication were approved in the EU compared with 133 for adults[52].

Unlike in adult cancers, tumours in children and young people often have significantly fewer mutations and are instead driven by a few driver genes[53]. Increasing our understanding of the genome and these driver genes through research is therefore essential in finding new routes for diagnosis and treatment for children and young people with cancer.

CRUK welcomed the previous UK Government's creation of a Children and Young People Cancer Taskforce, particularly the focus on children's and young people's access to genomic testing and to high quality personalised therapies. It is important that this work is taken forward by the new Government. The Scottish Government's commitment to supporting children and young people with cancer, as outlined in their 2021-2026 strategy was positive- although there is much further to go on building a genomic healthcare system to fully realise these ambitions.

5.1 Diagnosis & Treatment

The NHS Long Term Plan[54] set out an ambitious 10-year goal of offering all children with rare diseases or cancer in England Whole Genome Sequencing (WGS) at point of diagnosis and the 2020 Genome UK strategy committed to expanding provision across the UK[55]. This goal was established to enable more comprehensive analysis at diagnosis due to the increased importance of genomics for children and young people with cancer, as well as to determine their eligibility for precision medicines/therapies and increase access to innovative treatments through clinical trials.

Rollout of WGS for children and young people with cancer across the UK began in 2021, with the service being implemented and delivered by the Genomics Medicine Service (GMS) and Genomics England. Currently, children and young people with cancer who are living in Northern Ireland, Scotland and Wales have their samples sent for genomic analysis to England, with the North Thames Genomics Laboratory Hub being commissioned by devolved nations' governments to conduct this work. Some stakeholders we spoke with felt this was the most cost-effective way of delivering CYP genomic testing across the UK given the small numbers of patients, and stakeholders who we interviewed reflected that the current setup is running smoothly. However, as with adult genomic testing, slow laboratory turnaround times is having a knock-on effect to patient care.

The need for this integrated approach is underpinned by the genomic delivery challenges seen at a whole-system level. CYP genomic testing provision operates in the same environment as wider cancer genomics, where delivery is restricted by tight workforce capacity, complex board sharing agreements and inefficient mechanisms for regularly updating the national test directory.

The process of WGS and interpretation can typically take four to six weeks to return clinically relevant results[56], depending on location. This process can therefore be too slow to initiate an effective change in a patient's treatment regime, coming too late for a multidisciplinary team (MDT) meeting where their treatment plan will be discussed. For children and young people with cancer, who are often diagnosed later than adults[57], returning results in a clinically meaningful timeframe is particularly important. WGS could play a role in understanding what's going on in a patient's tumour at the individual level, which in turn builds our wider understanding of CYP cancers. Expert stakeholders who we interviewed recommended that the GMS should aim for an expedited turnaround time of two weeks, in line with other cancer turnaround times in development in England.

Despite these issues, the use of WGS for in the diagnosis and treatment of children and young people with cancer is still having a positive impact. Over the last three years, analysis using WGS has provided additional information for over a quarter of CYP cancer patients in England[58] which further highlights the importance of utilising genomic data to improve patient outcomes.

Genomic testing for children and young people with cancer also plays a vital role in

determining their eligibility for precision medicines and therapies, which are targeted to a person's tumour type. These treatments can be kinder, more effective and lead to fewer short-term and long-term effects than some other forms of treatment, such as chemotherapy. Crucially, genomic testing can be used to make the stratification and personalisation of treatments more effective first-time round, minimising the need for multiple rounds of treatment and accompanying side effects.

There are a limited number of bespoke treatments for cancers affecting children and young people which means that many are treated with intense combinations of chemotherapy, radiotherapy and/or surgery as standard of care[59]. Consequently, most survivors of childhood, teenage or young adult cancers suffer long-term effects from treatment[60], which can have a notable impact on their mental and emotional wellbeing[61]. This was highlighted in our engagement with our panel of people affected by children's and young people's cancers. Given the distressing side-effects cancer treatment can have, those we spoke to saw value in genomic testing to treat cancer more precisely, and therefore reduce the negative side-effects. They also noted the role of clinical trials in providing access to promising new treatment options, which was one of the main reasons they would want to participate in genomic testing. We must harness the potential offered by genomic testing and genomic research to develop more effective and targeted treatments for young people with cancer.

5.2 Research

Genomically informed clinical trials can provide much needed new and improved treatment options for children and young people with cancer. But for children and young people to get the greatest benefit from genomic medicine, there is a need for more tailored and less toxic treatment options and improved clinical trial delivery and access.

Research and development of novel therapies for children's and young people's cancers is less likely to be funded by industry or other bodies, in part due to low patient numbers, issues with reimbursement and insufficient regulatory incentives[62]. To address this market failure, a sustainable route to discover and develop therapeutics specifically targeted at vulnerabilities of children's and young people's (CYP) cancers is needed. Cancer Research UK is already looking to address this through the recently launched *C-Further* CYP drug consortium, a partnership between Cancer Research Horizons and Life Arc.

To ensure that CYP patients can benefit from novel therapies, we need to see improved access to clinical research. Relatively small patient populations mean that virtually all CYP cancer clinical trials are international, which poses additional regulatory, finance and access barriers. Teenage and young adults (aged 15- to 24-years old) experience particular difficulties in accessing clinical trials, as they are often deemed too old for children's trials, but too young for adult trials. Uncertainty among clinicians about eligibility criteria means that young people are sometimes not referred by their clinician for whole genome sequencing or the full range of clinical trials. The ECMC Paediatric Network aims to improve access to clinical trials for this age group, but more must be

done at a national level to ensure this cohort receives equitable opportunities to take part in trials.

Cancer Research UK is the largest charitable funder of research into CYP cancers in the UK, and, in partnership with other funders and organisations, we are driving innovative approaches to genomic medicine[63]. For example, the StratMedPaediatrics (SMPaeds) programme, funded in partnership with Children with Cancer UK, is developing and driving processes, strategies, and innovations in cancer genomics for children and young people, with a focus on cancers that have either not responded or come back after treatment.

SMPaeds1 ran between 2019 and 2024, helping to develop a procedure for processing tissue-based next-generation (NGS) panel sequencing in relapsed/refractory CYP patients and improving our understanding of how CYP cancers develop and respond to treatments. The programme has already proven an effective system for analysing solid tumour and blood samples, with further discoveries expected as research continues. SMPaeds1 also established the pathway for entry into genomically driven clinical trials such as eSMART and DETERMINE (see Box A).

Once set up, SMPaeds2 aims to further research into different innovations for genomic diagnosis and treatment, including exploring the use of Next-Gen Sequencing, liquid biopsy to assess treatment response and emergence of resistance, and the role of epigenetics in CYP cancers.

Policy Recommendations

11. The UK Government must work with devolved governments, Genomics England, and the NHS Genomics Medicines Service to ensure that the genomic testing pathway is fit to meet the specific needs of children and young people with cancer, with link-up to clinical trial opportunities and adequate resourcing to support timely WGS as set out in the Genome UK strategy.

6. Build genomic testing pathways fit for research and care

"The four nations of the UK will work together to realise the potential of genomics for the benefit of patients and ensure that the genomics services thrive in each nation."

Genome UK, 2020[64]

In the four nations of the UK the ambitions for genomic healthcare are bold but service delivery is currently complicated by a genomic testing pathway that is not fit for research, and overall variation in the availability of genomic testing between and within nations. This chapter addresses these issues and sets out how the governments and health leaders in each nation can:

1. **Strengthen strategic guidance through collaboration with delivery partners** – sharing learnings and best practices between national teams, laboratories, and clinical workforce groups.
2. **Improve equity of access to services and research opportunities for all eligible patients** –expanding the list of genes that are reported on to include those relevant to research and trial recruitment opportunities and investing in infrastructure compatible with UK-wide clinical trials.

6.1 Commissioning in Northern Ireland, Scotland and Wales

As noted above, each nation in the UK has a distinct genomic service, with its own infrastructure and commissioning arrangements. There are opportunities for learning between nations, and for strengthening service delivery through improved and longer-term funding. This will be crucial in building genomic systems that can deliver for both routine cancer care and research, now and in the future.

In England there are some aspects of service delivery that are the responsibility of local actors and others that fall under national guidance from the Genomics Unit. For example, each GLH chooses the standard panel they report. Alongside commissioning, the Genomics Unit is responsible for national strategic oversight of genomic services in England, funding, and performance monitoring[65].

In Wales, the genomics service is commissioned by the Welsh Health Specialised Services Committee (WHSSC) with funding to deliver the tests that are listed on the NHSE test directory. Partly aided by the smaller population size and more streamlined structures, the close working relationships between the All-Wales Genomic Medicine Service, clinicians and government has created a constructive commissioning environment. If clinical service users request tests that aren't available on the test directory and an unmet need can be clearly outlined, then a business case can be taken

to WHSSC for discussion. It is positive to hear from a range of different stakeholders in Wales that these processes work effectively and constructively to meet patient and service user need.

In Northern Ireland, interviewees noted the constraints on commissioning arrangements, whereby funding is typically allocated in a singular envelope on a five-year basis and following business case approval. This system makes it hard to factor in the costs of new biomarker tests as they're developed, and add the relevant biomarkers to the panel, hampering their delivery within Northern Ireland, and limiting the tests that the service can offer. This in turn can limit patient access to new precision medicines which require a companion diagnostic test. It is clear from our interviewees that there is a strong political will in Northern Ireland to address these issues. Northern Ireland's forthcoming genomic strategy and implementation plan must consider and address this issue with the development of a financed genomic strategy as a priority for cancer patients.

The publication of the Scottish Genomic Medicine Strategy and 1-year implementation plan for 2024/5 is welcome. This recognised the need for a sustainable funding and commissioning model and noted that a comprehensive test approval pipeline is in development. To further expand Scotland's genomic healthcare offer, genomic estates, digital and testing infrastructure must be strengthened. This cannot be achieved without financial investment to accompany the strategy. Although this is challenging in the current economic climate, stakeholders noted that investment in building Scotland's genomic capabilities now could present cost saving opportunities in the future, by facilitating earlier diagnosis of cancer and making sure patients get the most appropriate treatment for their cancer first time round.

Adequate and sustained funding is needed to ensure that genomics testing infrastructure across the four nations is compatible and that no nation falls behind. The variation in how genomics services are structured and commissioned can make it hard to set up and carry out UK-wide genomically enabled clinical trials- a more coordinated and compatible approach between the four nations needs to be developed to facilitate research. A joined up, UK-wide approach to clinical trial access is also needed, so that cancer patients can enter clinical trials across the UK irrespective of where they live. Unwarranted variation between the four nations must be urgently addressed- all patients across the UK should have equitable and timely access to genomic tests and the opportunity to participate in genomically stratified clinical trials.

Policy Recommendations

12. The next genomic strategy for Northern Ireland should either provide full support for the genomic healthcare system to deliver directly against the National Test Directory in England, following the Welsh model, or funding should be provided to the genomics medicine service in Northern Ireland to develop their own test directory.

13. In Scotland, the Government must allocate funding to implement the full five-year genomic medicine strategy, to enable Scotland to provide the best genomic service to cancer patients. This should include publishing implementation plans for each year of the strategy, building on the 2024-5 implementation plan.

6.2 Commissioning for research

"We know that ensuring patient participation in clinical trials is key to success. That why we will give everyone the opportunity to participate in research if they want to."

Labour, *Life Sciences Sector Plan*[66]

One of the most significant barriers to research that emerged during our engagement is the current commissioning model for genomic tests and reporting, which is not aligned with the UK Government's ambition for everyone to have the opportunity to take part in research. Comprehensive and up-front reporting of research-relevant genes is essential to equip clinicians and patients with the information they need to plan patient care, including identifying potential trial opportunities.

In England, genomic testing is delivered through a network of seven regional Genomic Laboratory Hubs, commissioned by the NHSE Genomics Unit to perform tests listed in the National Genomic Test Directory. Wales and Northern Ireland follow England's directory, though Northern Ireland provides a more limited service. Scotland operates a different model, with separate test directories for rare and inherited diseases and for cancer.

These directories specify the gene results that genomic laboratories across the UK must include in their reports to treating clinicians. However, many vital research relevant genes are excluded from these lists on the grounds they do not have 'proven clinical utility'. We heard that the shortage of clinician scientists means that requests for additional reporting outside of these test directories are often rejected, even when there is a strong research case.

Stakeholders consistently highlighted how this operating model is at odds with research, given that a core aim of research is to determine the future clinical utility of various genes. By restricting genomic reporting to genes that already have proven actionability in standard of care, patient opportunities for trial participation are stuck in time. This is

especially frustrating for researchers and clinicians, as many of these genes are already analysed as part of large panel testing and whole genome sequencing (WGS) for standard of care (SoC).

There are understandable concerns that additional reporting could overwhelm clinicians and delay turnaround times for standard of care testing. But without these reports, it is often not possible to determine if a patient is eligible for a precision medicine trial. Instead of excluding research relevant genes from reporting, clinicians should be given the time and training needed to interpret these results more efficiently.

Interviewees also reported considerable variation in how different genomic laboratory hubs (GLHs) are responding to this issue, which further exacerbates inequities in access. Each hub independently develops its own testing solutions and analysis pipelines for next-generation sequencing (NGS) panel testing, with no central mechanism to coordinate how results should be interpreted for trial eligibility. Additionally, patients are more likely to undergo research analysis and interpretation if their treating clinician has strong research connections and influence within the GLHs, further deepening these inequities.

There is an opportunity for increased sharing of learning between GLHs, from sharing panels or ways of working, which could be facilitated by Genomics Medicine Service Alliances (GMSAs). Working in partnership, GMSAs and GLHs should ensure that lab governance combines both providing services for routine care and for supporting research, so that this tension is actively managed.

To prevent UK cancer patients from missing out on potentially life-saving treatment, some trials have been forced to recruit patients through molecular profiling studies, such as TARGET National. This approach reportedly introduces its own challenges, including increased costs, disputes over data ownership, and the further fragmentation of data that could be used for secondary research. Moreover, this strategy reinforces access inequities, as not all clinicians have the necessary knowledge, connections and influence to arrange this for their patients.

Policy Recommendations

Short-term:

14. NHS England and devolved health systems should urgently publish national guidance that clarifies the process for reporting on research relevant genes outside of the respective national test directories, including how to prioritise limited capacity within genomic laboratories to support research. This should include clearly defining the roles of clinical scientists and providing adequate resource for them to report on genes that are important for trials and research, as well as standard of care.
15. In England, Genomic Medicine Service Alliances should work with Genomics Laboratory Hubs to ensure that research requirements are more embedded within service provision, and that governance includes consideration of how to manage providing services for both standard of care and research, such as risk registers.
16. Working in partnership with the research community, NHS England and devolved health systems should also initiate a stepwise expansion of the genes that genomic laboratories are required to report on. This should be underpinned by a culture shift that promotes the reporting of research relevant genes.

Longer-term:

17. In the longer term, DHSC and devolved equivalents should work with key delivery partners – including the managers of UK trial registries - to establish a mechanism that automatically updates the national test directories to ensure genomic testing is aligned with current and upcoming clinical trials.

These changes must be underpinned by increased and longer-term investment into the delivery of genomic testing in the UK, and planning for future demand. This includes the expansion of analytical capacity within genomics laboratories in each nation.

Greater automation holds considerable potential. Effective digitisation and IT interoperability could reduce administrative burden when ordering tests, tracking samples, and increase efficiency in the reporting of results. This includes ensuring that systems used within and between hospitals, the genomic labs, as well as linkage to other services (particularly pathology) are interoperable. GMSAs could use their expertise in innovation to advise GLHs on innovations which would help improve efficiency, and drive adoption of new technologies.

As demand for genomic testing for both routine cancer care and cancer research continues to grow, forward planning for the strategic challenges this presents and how this will demand be met by genomics systems across the UK is needed now. If government and health system leaders across the four nations, working in collaboration with genomics services and labs, develop coordinated long-term strategies for genomics delivery, then systems can be adequately prepared, and investment targeted in a future-proof way.

7. Support a sustainable and research-informed workforce to deliver genomic medicine

"A patient's engagement is largely influenced by how the healthcare staff present and communicate the information, it doesn't have to be an overload."

CRUK Patient Involvement Network member

Genomics will play a significant role in the future delivery of cancer care and research, helping us to understand more about the drivers of cancer, how the disease progresses, and new approaches to treatment. To fully realise the promise of genomic medicine for cancer, we must first have the right workforce. This means undertaking comprehensive workforce planning to understand the workforce we have today, and where there are shortfalls. Planning must include a focus on meeting the needs of research, placing value on the skills and expertise needed to support clinical trials and recruit patients. Given the length of training for many genomic specialists, due consideration must also be given to increasing the number of university places, and training places on to meet future demand, alongside new workforce models that can use the staff we do have most efficiently.

The rate of change in genomics is staggering meaning that staff will need to be continuously learning to keep up with the latest knowledge and skills. Staff with a firm understanding of cancer genomics are better able to direct patients towards relevant clinical trials – enhancing the UK clinical research environment and, crucially, improving patient outcomes.

The workforce can be better equipped by:

1. **Robust workforce planning** – ensuring a sustainable supply of the specialist and highly skilled staff across clinical and scientific professions essential to the delivery of genomic medicine, including consideration of how the supply of certain professions will need to increase in line with patient need and the rate of innovation.
2. **Support staff to continually upskill in genomics** – providing sufficient time and funding to ensure staff are provided with adequate genomic literacy to support adoption, with materials tailored to the educational requirements of specific groups.
3. **Build a research-engaged workforce –** providing the training, time and tools to ensure all clinicians can connect patients with genomics-informed clinical trials.

7.1 Workforce planning

Genomics relies on a specialist and highly skilled workforce, which is facing acute shortfalls. For example, significant challenges remain in the pathology workforce where an ageing staff approaching retirement could represent significant issues in the near future[67]. Genomics also relies on a number of sub-specialisms within healthcare sciences such as clinical genetics and bioinformatics where we have heard there are particular shortages. However, workforce data on the healthcare sciences workforce is severely lacking making it difficult to fully understand the challenges some of these specialisms are facing.

The NHSE Long Term Workforce Plan made welcome recognition of the need to grow the healthcare sciences workforce, although this diverse and highly specialised set of professionals were grouped into one overarching category. This approach fails to recognise the unique skillsets and different training pipelines for specialisms. It is welcome that some of the issues may be addressed by the ongoing workforce modelling being undertaken by the GMS as part of commitments made in Genome UK. There are currently no plans for either this modelling, or the local workforce plans it will inform, to be publicly released. This means many of the challenges facing these workforce groups may remain hidden. This highlights the pressing need for a cancer-specific workforce plan as a companion to the NHSE Long Term Workforce Plan, which must consider the professions and skills necessary to meet patient needs now and, in the future, including in cancer genomics.

The Welsh Government are taking a different approach to Genome UK's direction on workforce planning, as they will publish a specific Strategic Workforce Plan for Genomics. Similarly, the recent Scotland Genomic Medicine Strategy 2024-2029 committed to developing a robust multi-disciplinary whole-system workforce model for genomics services. These plans are promising but they must be met with adequate funding to deliver on their ambitions. In Northern Ireland, long-term and strategic workforce planning for genomics must be included in the upcoming genomics implementation plan.

In England, the 2025 refresh of the Long-Term Workforce Plan must build on its previous healthcare sciences commitments and combine this data with insights from the GMS modelling to provide up-to-date and transparent analysis on the needs of the genomics workforce.

Whilst long-term workforce planning is essential, efforts to address immediate challenges are also needed. This is especially true given that it takes three years to become a Clinical Scientist on the NHS Scientists Training Programme. To become a Consultant Scientist takes a further five years of training via the Higher Specialist Scientist Training programme. Given this time lag, we need solutions that will ease workforce pressures today. Skill mix approaches, where members of the wider team with the skills to perform some duties previously undertaken only by doctors or other experienced professionals, offer a real opportunity to increase workforce productivity and allow the

most experienced staff to focus on the most complex cases.

With proper planning and quality assurances, skill mix approaches can deliver professional development opportunities for staff and ensure the best care for patients. The relevant medical royal colleges, professional bodies, regulators and the health service in each nation must actively work together to find the most appropriate skill mix approaches to support genomic medicine. The biomedical sciences workforce offers a useful pool of talent to do this, as places are not centrally set at undergraduate level and an accreditation model is already in place through the IMBS and Royal College of Pathologists. Specialist medics and healthcare scientists will always be needed in genomic medicine, but by focusing on skills rather than roles and using top-of-the-license thinking we can better utilise the capacity we have in the system helping to reduce pressures on an already stretched workforce and improve testing turnaround times.

Policy Recommendations

18. Teams across NHS England, the Genomics Medicine Service and DHSC must actively work together on healthcare workforce planning, ensuring a coordinated and robust approach that fully accounts for the needs of the future cancer workforce. This includes:
    - A fully transparent modelling framework detailing how NHS England will estimate staffing needs. NHS England should then publicly consult on the model to ensure it is independently verified.
    - Consideration of the impact of opportunities to engage in clinical research, including in genomics, on boosting staff retention.
    - Actions to support the adoption of 'skill mix' approaches in areas of workforce shortages.
19. The Strategic Workforce Plan for Genomics in Wales, and the workforce model for genomics services being developed as part of the Scotland Genomic Medicine Strategy 2024-2029, must both be met with adequate funding from their respective governments to deliver on ambitions to grow and develop the genomics workforce in these countries.
20. In Northern Ireland, the Department of Health and Health and Social Care NI must include long-term, funded workforce planning commitments in their upcoming genomics implementation plan.

7.2 Information and education

In Genome UK, the UK government recognised the importance of broad engagement in genomics across NHS staff, patients, and the public. Whilst the style of communication will differ for each audience, general themes come through from all groups around confidence in understanding and interpreting genomic information. Today's patients are

already benefiting from the explosion of genomic knowledge, with several genetic tests available to families with a history of specific types of cancer, along with targeted treatments based on the specific mutations in a tumour's DNA. But the public's understanding of genomics, and its inherent links with cancer remain low[68]. This presents issues when patients are given their cancer genomic testing results or are being asked to consent to sharing genomic data for research.

During our patient and public involvement work, we heard from many participants that they did not fully understand the concepts of genomics or genetic testing prior to the CRUK involvement session – even if they had received this testing as part of their cancer treatment. When asked about how they would like to receive information on genomics, it was clear that a variety of methods were favoured. Patients and family members expressed concern that genomic information or results could be given without appropriate context, leaving them to turn to the internet for clarification which could easily increase anxiety and worry. Patients also said that they would look for advice, clarification, and guidance from a variety of healthcare professionals on genomic testing or research, including their consultant, their clinical nurse specialist (CNS) and their GP. Some participants reflected that clinic appointments with a consultant can feel rushed and overwhelming, so being able to discuss results with nurses or GPs in a more relaxed environment would also be helpful. This demonstrates the need for appropriate expertise throughout the care pathway on genomics, so patients know who to go to with questions, and clinicians are prepared to provide guidance and direct patients to further tailored support as needed.

The new opportunities presented by genomics to understand and treat diseases like cancer are vital for patients but also create challenges for the workforce, who must adapt to this new landscape. Genome UK set out a commitment for Health Education England (HEE) to work with the Academy of Medical Royal Colleges to ensure genomic medicine is embedded in medical training. Since then, the Genomics Education Programme (GEP), now sitting within NHSE, has created a plethora of high-quality educational resources on genomics, set up the Clinical Pathways Initiative (CPI) and sought to actively engage the workforce in the topic. It is encouraging to see Scotland's Genomic Medicine Strategy commit to collaborating on and adapting GEP resources through NHS Education Scotland to improve genomic literacy and expertise across the wider workforce. Wales are also looking to provide further genomics education through flexible eLearning modules. Across the board, learnings from the rollout of GeNotes in England should be applied in the other UK nations, and GEP resources adapted and used where relevant.

Whilst the need for genomic literacy is clear, challenges remain in staff having protected time to engage in training and upskilling. Greater consideration is needed in local workforce planning to ensure that staff across the workforce, not just those already working in genomics, have allocated time for professional development, including training.

Policy Recommendations

21. Across the UK, NHS Trusts and Health Boards should support genomics staff by giving them dedicated time, and where necessary, funding for training and continued professional development. There must be increased recognition of the importance of continued learning and development outside of genomics-specific professions from senior leadership in local organisations. Training on genomics should be prioritised for staff at all levels and in a variety of professions, but with a particular focus on cancer nursing.

7.3 Supporting clinicians to connect patients with clinical trials

"There is good evidence that giving time for research to staff who are interested enhances recruitment and retention. We should be making most of what research can offer to contribute to staff development."

Labour, *Life Sciences Sector Plan*

Research must be integrated into genomics training and professional development, with join-up between providers of healthcare and research training. The advances in genomic technologies are an exciting development for research, but they will also place additional demands on an already stretched and under-supported workforce. Clinicians will need the expertise and time to accurately interpret genomic test results, stay up-to-date with open trial opportunities, navigate increasingly stringent eligibility criteria, and feel confident and incentivised to discuss complicated research information with patients during short, routine appointments.

These additional expectations are compounded by numerous existing barriers faced by clinicians. As set out in our Programme for UK Government, *Longer, better lives*, these include a lack of time, an overall lack of research awareness among clinicians and the public, and insufficient tools to help clinicians find open trials. These challenges deter clinicians from discussing research during patient appointments.

Only

45%

of surveyed cancer patients in England reported having conversations with their treating clinician about research in 2023[69].

Stakeholder interviews revealed concerning inequities, with patients more likely to gain privileged access to a potentially life-saving clinical trial if they are under the care of a senior clinical academic with an active research practice and network.

Normalising conversations about research as part of patient care is not solely the responsibility of clinicians, the health service, and educators. Medical research charities, royal colleges and unions, research delivery teams, industry, and government also play

a crucial role in connecting the public with research. This includes equipping clinicians and the public with the right tools and information.

Clinical trial registries, for example, are an essential recruitment tool used by clinicians, but UK registries, including the International Standard Randomised Controlled Trial Number (ISRCTN) registry, are not kept reliably up to date. This becomes particularly challenging when recruitment dates are extended, new sites are added, or studies close early.

It is vital that there is an equity of information about open trial opportunities provided to all clinicians across the UK, including easily accessible and up to date trial registries with a single point of entry. The UK Government and devolved administrations should consult medical research charities with expertise in registry building and maintenance to learn lessons from successful registries, such as the ECMC Experimental Cancer Trial Finder, and consider how to expand these further. The ECMC Trial finder is particularly innovative as it allows the identification of clinical trial opportunities for patients based on a search for the relevant gene or marker, rather than only by cancer site.

Policy Recommendations

22. The Health Research Authority, the International Standard Randomised Control Trial Number (ISRCTN) registry and National Institute for Health and Care Research (NIHR) Be Part of Research should make it easier for sponsors to update study records, while also strengthening accountability. BioMed Central, which manages the ISRCTN registry, will require the right level of dedicated resource to deliver these changes.
    - As recommended in the Lord O'Shaughnessy review of industry trials, we also believe UK trial registries should include gene and variant level inclusion and exclusion criteria, similar to the ECMC Experimental Cancer Trial Finder, and have a machine-readable interface that can be integrated within clinical decision support systems.

8. Develop data infrastructure that underpins the next generation of research

"We will make the UK the best place in the world to access genomic data for research."

Genome UK, 2020[70]

Excellent quality data is the lifeblood of the UK's Genomic landscape[71], bringing insights for improved patient care, and advancements in medical research. We know that data saves lives[72]. There are readily recognised issues for health data such as privacy, security, and efficient access. These issues manifest within genomic data but there are also specific distinct challenges that sit alongside and interact with these broader issues, complicating an already complex picture[73]. This chapter will explain how DHSC, NHSE and their devolved equivalents can work to improve the data landscape to provide more effective clinical care to patients, and deliver richer data to researchers. This will require:

1. **More uniform collection, curation, and accessibility of data –** improving the quality and quantity of data that is available to be accessed for research, whilst ensuring equity across the UK and addressing embedded inequalities.
2. **Improved data harmonisation, linkage, and flow –** harmonising genomic datasets with each other as well as ensuring that datasets are to be collected and curated as to ensure linkage to non-genomic data is easier to improve and hone the insights that can be gathered from data that already exists in the UK system.
3. **Continuous patient and public involvement with genomic data decisions** – ensuring continued public support, preventing potential public backlashes from undermining advances.

Getting genomic data systems correct now is essential. Sequencing the first human genome cost £4bn and took 13 years – it now takes less than a day and costs under £1000[74]. This is leading to a proliferation of genomic testing, and with that the generation of vast quantities of genetic data[75]. Historically, the primary focus of cancer research was relatively small-scale studies of essential pathways and genes. However, recent rapid advancements in technology has seen the growth of large-scale multi-omic data research[76]. This research is likely to produce some of the most cutting-edge advancements in our understanding of cancer. Whilst researchers develop innovations in data analysis, it is the responsibility of national health systems and departments of health across the four nations to develop innovations in the data ecosystem. Getting the collection, storage, flow and access governance and infrastructure correct now will mean our scientists can harness genomic data for its full, life-saving potential. The

longer that we wait, the more complex and difficult this process will be.

CRUK is pleased that health data has been central to recent strategies across the four nations, both genomic and otherwise. The new UK Government's promise to ensure the federation of genomic datasets with a single point of access has seen an explicit acknowledgement of the opportunity presented by NHS genomic health data – delivery of this is now critical. The evidence is clear, UK genomic data assets like UK biobank are central to our global standing into the research of cancer with over 26,000 researchers worldwide currently using the data[77].

The central tenet of health and genomic specific data strategies has been to harness these assets, harmonize across the system, and use genomics as an exemplar for the responsible use of health data[78]. The 2019 NHS Long Term Plan spoke of targeted investments in genomics, whilst 2020's Genome UK set three data pillars across research, including the ambition to see the UK as a global leader in data based genomic research. The UK nations have also set specific deliverables and data aims. In England, DHSC and NHSE's 2022 commitments to implementation further emphasised the need and ambition for federated access to genomic and non-genomic data, and increased collaboration across the UK – with the standardisation of data being critical. In Wales, the Genomics Delivery Plan committed to developing and implementing a digital roadmap that would allow the unhindered-yet-secure flow of genomic data. Whilst in Scotland, the most recent implementation update highlighted many actions including looking at solutions for a centralised data store, enabling sharing across health boundaries and with researchers. These ambitions have been mirrored in the Labour Party's Life Sciences Plan, with the view to create a single point of access for all genomic data[79].

Despite these laudable goals, implementation details have remained scarce. This is exemplified by the Genomics Information Implementation Plan remaining unpublished, despite being a commitment in the NHSE Genomics Strategy[80]. Public statements on NHSE's Research Secure Data Environment (SDE) Network have promised the integration of genomic data but without a delivery plan or timeline. It is critical that DHSC, NHSE and their devolved equivalents now follow through with the delivery of promised changes. The UK's genomic data infrastructure could be an exemplar for the effective use of health data both in the UK and abroad, but there is significant work to be done first.

Box C

Genomic Data Driven Cancer Research

Case Study: Professor Fitzgerald

Oesophageal cancer is poorly understood, and survival remains low; for 2017-2019, it is predicted that more than 1 in 10 people diagnosed with oesophageal cancer survive their disease for 10 years or more. OCCAMS (Oesophageal Cancer Clinical and Molecular Stratification) project, spearheaded by Professor Fitzgerald, collects tumour and blood samples from people with oesophageal cancer to decode the cancer's genetic profile to better understand it. Large amounts of high-quality genetic data are essential for making progress.

8.1 Collection, Consent and Access

"Labour will begin by... ensuring proper federation of data sets, with a single access point for researchers to use data from all our genomic resources ... to create a truly world-leading R&D platform."

Labour's plan for the life science sector, Feb 2024[81]

The collection and curation of genomic data is critical for both care and research – despite advances in the collection and access of genomic data, two critical issues remain.

1. **How collected data is made available for research**– Whole Genome Sequencing (WGS) unlocks a greater ability for driving data driven research, but only makes up a small percentage of testing. Some non-WGS data is left in disparate databanks unable to be used for research.
2. **What consent is given for the use of collected data -** Patients must be able to understand exactly what they are consenting to. Consenting practices must support innovation and research but current consenting practices for non-WGS mean that this doesn't always happen.

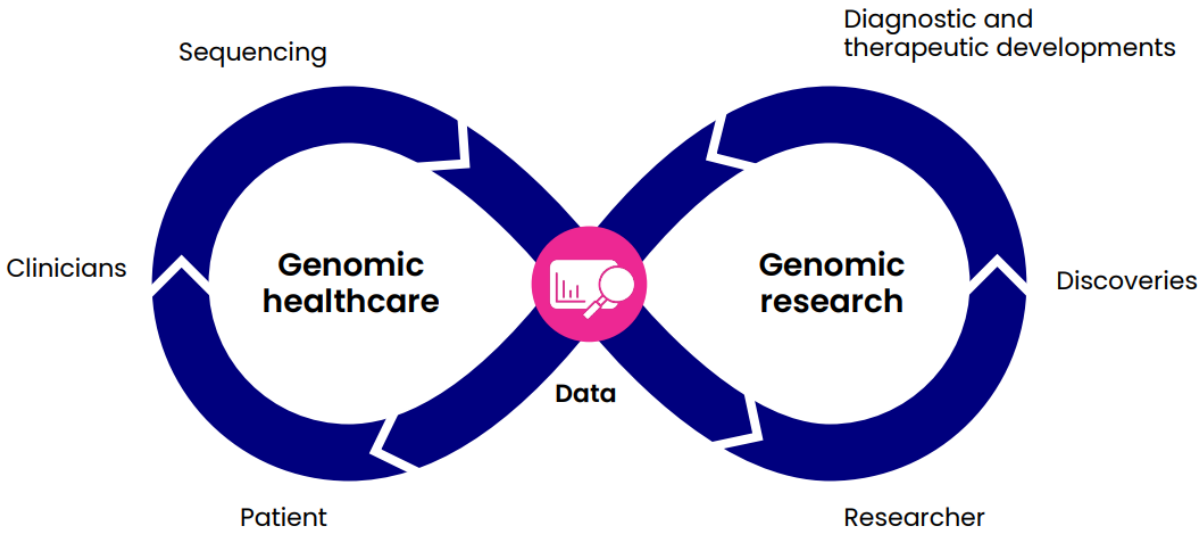
There are also further issues with inequalities, workforce and storage that should be addressed.

Collection

The NHS in England takes a multi-modal approach to the collection of genomic data. In England, targeted testing, panels and exome sequencing (i.e. testing for the presence of specific genes currently known to be associated with cancer) make up most of the current testing, whereas WGS (i.e. looking at *nearly* the whole genome) takes up the rest[82]. However, this is inverse to the quality of research that can be conducted. When patients undergo WGS this is sequenced by Genomics England and stored on Genomics England's Research Environment for researcher access[83]. This allows for quick and

efficient access to linked quality genomic data - producing life-saving data driven research. Genomics England describes this as an 'infinity loop', whereby the more genomic healthcare is delivered and data donated, the more powerful research becomes, improving healthcare delivery and bringing more data for research. It is welcome that embedding data driven research into the delivery of care is increasingly seen as critical and has been flagged across recent strategies.

Figure H: Infinity Loop



Within the UK, non-WGS data is processed by individual Genomics Laboratory Hubs (GLHs) and local labs. Despite lots of genomic cancer data being channelled effectively through the National Disease Registration Service (NDRS) and being able to be accessed via the Data Access Request Service (DARS), due to the complexity and fragmented nature of the GLH system, combined with capacity and IT issues timely, structured, and efficient data is not always delivered. This has meant that some non-WGS data produced by clinical care has been reported to us as being left untapped in disparate databases, unable to be fully harnessed. These databases are individually procured and collected data is stored differently on each one. Non-standardisation across different systems also means that even if collected data is submitted to the NDRS because of a lack of interoperability and harmonization it requires significant effort to curate the data into a usable, linkable format.

Box D

Unified Genomic Record

The north star of genomic data in England. This would mean that all genomic data is able to be accessed via a single front door, with datasets linked and federated. NHS England are currently exploring the possibility. We would like this approach to be considered in Scotland, Wales, and Northern Ireland too. If delivered effectively this would be fantastic for genomic healthcare and research. Establishing linkage with GP and primary care data is likely to be the largest stumbling block that NHSE must seek to address to ensure optimal data access for research.

Long-term, the solution should be to unify these disparate databases into one system in each nation. This would create a bank of federated genomic data and would put the UK at the forefront of a genomic data revolution. Optimising sharing between these national linked databases would then be the next challenge. CRUK is pleased that there has been some movement within NHSE towards a unified record, and increasing federated access has been highlighted in recent strategies, but details and timelines are yet to be publicly released. In Wales, the centralised system means that this is already in train but the Gene Park TRE must be delivered to allow efficient access to linked data. In Scotland options analysis has been initiated for a centralised data store, these options should be published as well as a timeline for delivery. In Northern Ireland, a similar unification across disparate datasets will need to take place. Delivery of all these initiatives is now needed.

CRUK appreciates the difficulty of progressing towards a unified centralised record. To achieve its full potential in England, this would require complex negotiations between GLHs, local trusts and NHSE. To accelerate this process, increased standardisation of the collection, curation, and storage of data across individual labs is critical, because even if federated access was achieved, it would be difficult for the data to be interoperable. Bringing GLHs together to agree set data standards and data access routes, could accelerate data access in the present, whilst making a potential unified genomic record a more viable prospect in the future. This should also happen in Scotland within each of the genomic laboratories.

Consent

Consent is a particularly difficult issue in genomics due to the many possible outcomes from a genetic test, including unanticipated issues with familial linkages, where results can give information about family members. As new uses of collected data emerge, there are questions over whether these new uses can be considered to fall under the consent that was originally given. Informed consent assumes knowledge of outcomes, but this is difficult or impossible to assure and has led to a lack of consensus of what constitutes genetic testing consent[84]. This has meant that many research organisations, like UK Biobank, rely on broad definitions of consent. We have heard that there is a need for further deliberation on the creation of a cross-nation consensus on how consent

should be offered and delivered for clinical care. Currently an appropriate discussion of genomic testing and the possible implications is required before any genomic testing takes place. (Aside from certain non-WGS tests on cancer tissue). It's important that patients feel empowered to make informed decisions about how their data is used whether that is WGS or non-WGS.

Consent for the use of clinical data in England for research varies significantly based on what test is occurring. During WGS, consent for research is offered alongside consenting for clinical tests. This 'hybrid-patient consent model' has been a large success with approximately 90% of patients giving consent for their data being used for research[85].

However, for other tests, these consent forms are not currently offered. Although NDRS has special permission to collect genomic cancer data, lack of consent forms means that they are constrained in the ways that this data can be used. It also means that patients cannot be linked with relevant clinical trials through routes other than clinician referral. This means that many patients must be tested twice, once for standard of care and then again within a trial. Patients are therefore required to undergo multiple investigations, some that may involve surgical biopsies which can be worrying, uncomfortable, as well as more costly for the health system. Creating and streamlining the necessary consenting forms to allow this data to flow readily though the system would also aid in the reduction of the size of stored genetic data.

To address this, NHSE should look to understand the viability of expanding consenting for research forms beyond WGS, to other types of tests. This was put forward, with specific mention of cancer, in Accelerating Genomic Medicine in the NHS[86]. This may require centralised forms and harmonized storage, as well as clear use cases. We are hopeful that NHSE can deliver on this promise to evolve patient choice over the next few years. It is also welcome that in the latest Scotland Genomic Implementation Strategy implementing a model of consent for research in testing pathways was suggested[87]. Now this must be delivered in Scotland as a priority.

These consent forms should be designed in consultation with the public and should be accompanied with videos that explain these difficult concepts in simple language. Effort should be made to ensure these explainers speak to the full range of communities that we have in the UK. Government could proactively partner with organisations such as Genetics Engage, who advocate for inclusion of all communities underserved and underrepresented in genetic and genomic service, to foster a truly inclusive approach.

Eventually, the NHS should set the goal of being able to consent and unconsent within the NHS app for the purpose of research, accompanied by other ways to consent to ensure that all patients are able to consent in the way that works best for them, digitally or manually. Standardised consent forms must include information on who will have access to the data and what the data will be used for. This was a critical theme of our public engagement with useMYdata.

Data Diversity

Representative data in genomic research is a known issue[88]. Diversity in the data is poor as flagship genomic research programmes like UK Biobank have predominantly recruited from certain demographics. 95% of participants in UK biobank were white[89].

Just

2%

of genome-wide association studies participants are of African ancestry[90].

This issue is not just academic; it has serious real-world consequences for the treatment of cancer. Research found that in a cohort of 6,500 diverse individuals from the UK, white respondents were approximately twice as likely to have had a genetic test than respondents from ethnic minority backgrounds[91]. Reasons for underrepresentation are complex, but language, cultural beliefs, as well as historic and more recent issues with trust in research programmes have all been cited[92].

CRUK is pleased to see the launch of the diverse data programme though Genomics England, and the focus on collecting the WGS of over 15,000 participants from minority communities. It is also positive that Scotland is working with the Anti-Racism Observatory to help address these issues. Building on these examples we must double down efforts to extend recruitment into clinical research programmes to traditionally marginalised groups and make sure that there is delivery of these commitments to diversity data.

To ensure that this goal is achieved, large scale research programmes should work to publish data on exactly how diverse their datasets are, and addressing inequalities in genetic datasets should underpin outreach campaigns. To monitor how equitable service provision is across the UK, there should be a consistent transparent measure of provision and uptake of testing by region and demographic groups. Making sure that collected data in non-WGS can be readily used for research and is linked to other datasets would also go some way to address these inequalities, as this data is more likely to be representative of the whole UK population.

However, it must be recognised that addressing inequalities in data driven research cannot be fixed purely by expanding datasets. This should be accompanied by sufficient governance and oversight to ensure that algorithmic based data analysis is not exacerbating inequalities.

Workforce

A highly trained data workforce is essential for the delivery of the most innovative data-driven research as well as more traditional clinical trial recruitment and health system examination. Innovations in data systems will not deliver on their promises unless they have well trained staff alongside them.

A data literate and diverse workforce is required to ensure that patients understand exactly what is happening with their health data. Healthcare staff need to be able to

explain and articulate the value of patients consenting to share their data with researchers, what exactly they are consenting to, as well as potential risks. This requires ground-up understanding and training in specific genomic data issues like de-identification. This is particularly important for front-line NHS staff who are the main point of contact for patients.

There also needs to be considerable investment in the back-office IT workforce within health systems to ensure that there are sufficient informaticians, data scientists and managers to effectively curate and process data. A variety of stakeholders suggested that salaries will need to be reviewed to be more closely aligned with industry standards to ensure that an adequate workforce is attracted and retained. Sufficient prospects for career progression as well as continuous training and development programs should be seen as essential to maximise investment in staff and drive forward change. It is positive to see that retaining high quality data scientists and developing an appealing career structure is mentioned in the genomic delivery plan for Wales, but this ambition must be realised.

Storage

Due to its significant size, the storage of genomic data is a challenge. At a clinical level the data requirements are huge, and this is only set to increase as genomic testing is increased. Keeping up with this demand is likely to be difficult and digital infrastructure can often be overlooked. Departments of Health in each nation must ensure that data storage requirements are met, resourced, and maintained at the highest possible standards. Forward planning is essential, so storage issues do not present a bottleneck to effective data use.

Current guidance on data storage is written for a pre-genomic age. There are strict data processing rules along with consenting practices that require care data to be deleted after several years- after eight years for generic health data and after 30 years for cancer data[93]. However, the detail and granularity of genomic datasets produced in care are likely to continue to produce an asset for researchers for generations to come. In our public engagement with useMYdata we found a broad consensus that with the correct consenting protocols and information, patients should have the option to consent for their data to be stored for an indefinite amount of time. Currently the infancy of most genetic datasets combined with a lack of guidance is creating confusion over what is the best course of action. To resolve this, DHSC and the devolved equivalents should urgently bring relevant organisations, including national health systems, researchers, charities, and clinicians together to discuss creating clear standardised guidance. This would ensure that valuable genomic data is not deleted prematurely.

Policy Recommendations

23. NHS England, NHS Scotland, NHS Wales and Health and Social Care N.I. should work to ensure optimal access to genomic data produced in the UK for research. This is particularly important for non-whole genome sequenced data where a more unified approach to collection, storage and access would aid in life saving research. Patient consent is critical, alongside establishing balanced consenting approvals that are not overly burdensome on patients whilst supporting research.

24. UK Government and devolved governments must prioritise workforce planning to ensure a highly trained data workforce for the future, with sufficient informaticians, data scientists and managers to effectively curate and process data. Creating the pay, working conditions, and progression incentives that more closely align with industry standards is essential for this.

Box E

Enabling data-driven recruitment to clinical trials

By delivering the recommendations in this chapter, the UK Government can unlock the growing potential of data-driven recruitment to clinical trials. While clinician-led recruitment will always be central, the NHS DigiTrials service has demonstrated the promise of a more data-driven approach. By using routinely collected patient data, a dedicated team can identify and follow-up with individuals who are likely to benefit from or meet the criteria for specific clinical trials. The NHS-Galleri trial benefited significantly from this approach, achieving a 12% success rate in recruiting participants – more than twice the industry standard[94].

To give more people the chance to participate in research, approved researchers should be able to access genomic healthcare data in a similar manner. This would allow them to identify and follow-up with patients who possess certain genetic variants or profiles relevant to a trial. This would ease the burden on time-poor clinicians and reduce the demand on GLHs for additional testing. Additionally, it would provide opportunities for horizon scanning by helping calculate the number of patients across the UK with different mutations. The provision of aggregated data for a fee could help industry make more informed decisions about where to place trials, thereby enhancing the UK's attractiveness as a research destination and potentially generating additional revenue for the health service.

In England, de-identified WGS data is already available to researchers through Genomics England's Trusted Research Environment. According to Genomics England, this data can be used for trial recruitment, with eligible patients being contacted by their clinical team or Genomics England[95]. During our engagement we were unable to confirm how this works in practice, what the uptake has been, or if researchers are

aware of this offer. Improved communication and clearer signposting from GEL are needed to clarify these questions.

By contrast, no centralised secure database with standardised consenting processes exists for non-WGS data. As a result, a large amount of genomic patient data remains locked within GLH databases, unable to be used to recruit patients to potentially life-saving trials or advance major discoveries. Several steps – detailed in this chapter – must be taken before non-WGS data can be effectively used to identify patients with specific genes that mean they are eligible for precision medicine clinical trials. These steps include ensuring more uniform data collection, curation, and accessibility; improving harmonisation, linkage and data flow; and maintaining continuous patient and public involvement to uphold public trust in the process.

8.2 Linkage and Sharing

"Oesophageal cancer is really complicated. We can't find the needle in the haystack by just using data from a few people."

Professor Fitzgerald, CRUK funded Genomic Researcher

Linkage

The UK has some of the most detailed genomic datasets in the world, but the data is spread, siloed, and fragmented across health systems in the UK, in multiple different banks and data stores. Genomic data is much more powerful when used in collaboration with other non-genomic data sets, such as clinical, but often this data is not interoperable, or it is locked behind difficult access protocols. Each UK nation must work to first centralise genomic datasets and de-fragment their disparate systems whilst working to link genetic data with other routine healthcare datasets.

The UK is a leader in genomics research but solving these two issues would allow it to fully harness its data assets. DHSC's Data Saves Lives strategy included commitments to ensure clinical care data is fed into patient records[96], and the 2022 to 2025 implementation plan for England saw the continuation of this with increased calls for linked genomic records though a federated platform.

Genetic data in the UK is produced in three major places, in clinical care, in research programmes such as UK Biobank or Our Future Health, and though direct-to-consumer genetic tests. These three strands are currently detached, both externally with each other but also internally within each strand, reducing the power of any one datastore. In England, the majority of NHS genomic testing happens in GLHs – all of which currently use a variety of different informatics systems. It is positive that a solution via interoperable informatics structure is at the heart of recent strategies, and there has been some movement towards introducing a unified health record. The exact method of linkage is less important than ensuring that the data does not remain siloed.

Even if all genomic data was linked together, linkage with other datasets would still be

essential. The largest potential barrier is primary care data. GPs are controllers of data within their surgeries. This makes accessing and linking this data to genomic records difficult, resource heavy, and often impossible.

Only

1/5th of GPs have allowed their patients' data to link with UK Biobank despite individuals having given their explicit consent[97].

In September 2023 NHSE, UK Biobank and the Royal College of GPs wrote to all GPs to urge them to approve the release of primary care data. The lack of linkage between secondary and primary care data is a huge issue for UK health data-based research, with solutions proving difficult. However, the current system is not sustainable, not only for research but also for respecting the wishes of patients regarding their own data. In October 2024 UK Health Secretary Wes Streeting announced plans to allow consented primary care data to be shared with researchers, paving the way for this issue to be addressed. While critical for all research, this would be indispensable for genomic research. It is important that timelines and details of plans are published soon and then delivered. But to fully realise GP data's potential, this should not be treated as an end point. By linking select GP data with other national health datasets researchers would be able to gain broader insights into the prevention and diagnosis of multiple conditions. This would significantly expand the value of the data and ensure wider-reaching benefits beyond specific consented research programmes.

Our interviews found that GPs were apprehensive about sharing data, not because they had fundamental disagreements with the programmes, but because they were time poor and were incentivised to be overly cautious with their patients' data. It is essential that Departments of Health in each nation work to not only allow data to be shared but also to ensure that GPs feel comfortable in data being shared for the purpose of research data. This should be achieved through clear centralised guidance and positive examples of the power of linked data. Online FAQ's as well a set of online resources for GPs developed by the Departments of Health in each nation in conjunction with national health systems should be created and updated to facilitate a positive atmosphere of sharing. The joint sharing model in Scotland, where both GPs and Health Boards share controllership of patient data, is widely viewed as a success, and has allowed data to be more readily available. NHSE and DHSC should investigate if this system would also work for England.

The efficient sharing of genomic data needs to work both ways- issues with data flows from secondary to primary care have been raised. Solving the interoperability of primary and secondary health data should be addressed so GPs can more easily see, and access genomic health record data produced in secondary care settings.

Sharing

There are specific challenges with genomic data, in that all data could be seen as potentially identifiable and therefore subject to additional data protection. There is currently little guidance on what can be shared, and what guidance there is often makes just cursory nods to genomic data. This causes researchers to take a cautious approach to data sharing for fear of legal issues. Researchers should be clear on what data they can share and when. There is an urgent need for cross-nation consensus and clarification on the definition of personal data. The government should work with regulators such as the Information Commissioner's Office (ICO) and equivalent Data Protection Authorities in the EU to build this. Any decision should take proportional approach, building on the work that GA4GH has done in setting guidance that weighs the risks of re-identification with the risks of data not being used for live-saving research, centred around common principles of trustworthiness[98]. The public should be actively brought into these conversations at the earliest opportunity.

Moreover, the four nations should work to ensure that it is as easy as possible to harmonise data across the entire UK. Exemplar programmes in each nation, such as the Wales Gene Park, and Genomics England, should work to make accessing data by researchers from across the UK as simple as possible and make sure that the data is as interoperable as possible between nations.

The larger the dataset, the greater potential for insights. Research into childhood and rare cancers often requires large populations that may not be found within the UK. Being able to share data across borders with non-UK countries is therefore critical for the success of some research programmes, making cross border collaboration vital. Current data sharing agreements can make this unnecessarily challenging. We heard from stakeholders that this is due to a combination of factors, such as agreements like the Nagoya protocol, that covers the sharing of genetic resources, and differences in interpretations of GDPR. Some cross-border friction will always occur, but DSIT should work to build a standardised consensus when it comes to data sharing protocols for genetic data, across and beyond the UK, working with partners abroad to ensure that international data transfers are as easy as possible. Ensuring that we retain data adequacy with the EU is critical for ensuring continued UK-EU data flows and so this should be a priority.

Policy Recommendations

25. The harmonisation of disparate genomic datasets should be seen as a critical next step in ensuring that genomic datasets across health systems in the four nations can be harnessed. NHS England, NHS Scotland, NHS Wales and Health and Social Care N.I. should work to ensure that genomic data is not siloed across multiple datastores.

26. For data to be optimally harnessed, there must be greater linkage between genomic data and other clinical data such as cancer status, stage, treatment, outcome, and lifestyle data (such as smoking status).
    - Linking GP data should be prioritised and DHSC should swiftly deliver on the promise to ensure that all data that is consented to being shared with a study, is shared.
    - To fully unlock its potential, DHSC needs to go further and ensure that select GP data are linked to other national health datasets, bringing wider-reaching benefits beyond specific consented research programmes.
    - DHSC and devolved equivalents should work to ensure that GPs and the public feel comfortable, supported, and informed about the responsible sharing of data.

Public Trust

"The NHS needs to demonstrate trustworthiness in the handling of genomic data."

CRUK-useMYdata Genomics Data Focus Group participant

Public trust is fundamental to any health data project. Without careful planning and engagement, mistakes whether real or perceived, threaten to override any good intentions. The public must view the health systems' use of data as fundamentally safe and secure, and driven by public benefit. Getting this right is vital for the success of any programme. This has been shown with the failed roll outs of GPDPR and care.data where lack of engagement and clear communication caused indefinite delays or eventual cancellations. Considering the potential importance of these programmes for advancing our understanding of cancer and the amount of public money that is spent, it is essential that health systems do further work to build public trust. Increased opt-outs threaten the validity of research, and because they are non-uniformly distributed there is very real risk of increasing bias in the data.[99] The fallout can also continue for years after a perceived mistake. Three years after GPDPR, misinformation is still causing opt-outs to increase[100].

It is welcome that NHSE has developed its public engagement campaigns in recent years and is increasingly bringing public representatives into conversations about how their data is being used. There is still a way to go though, with recent NHSE polling shows that

overall awareness about patient data is low. In our conversations with useMydata members there was a feeling that the NHS can do more to involve patients and the public in conversations about health data uses.

Only

20%

Of NHSE polled patients were aware of their data being used for research[101].

Although the whole of genomic healthcare and research cannot be viewed as one programme, like the SDE network, it's still important that when designing the underlying data system the same thinking is applied. This means that patients should always be aware of the potential uses for their identifiable data being shared beyond their care, be able to access clear information on what is needed to make an informed choice and be able to opt-out of secondary uses if applicable.

It is essential that the public are involved in the decision-making process when it comes to the creation and continued evolution of genomic data governance. There are excellent examples of where this is already happening, like Genomics England's participation panel. Genomic data brings with it ethical decisions that other types of health data may not, for example on how to procure consent for family linkages or what counts as anonymised genomic health data. The NHS should prioritise continuous engagement on these questions; building meaningful public trust is vital and should include continuous rigorous deliberation.

Policy Recommendations

27. Genomic data raises issues that are distinct from other types of health data - the public must continue to be meaningfully brought into conversations around what will happen to their genomic data when it is in the UK's health and research system.

28. Public outreach and education will ensure that opt-outs are kept to a minimum and don't threaten data driven research. Initiatives like the power of data campaign and Genomics England's participation panel should be expanded across the genomics system and properly resourced to make sure that the public feel confident in the ways that data can be used to help advance health outcomes.

9. Unlock the potential of future innovations

Genomic testing and analysis underpin several areas of emerging cancer innovations. There is also scope for innovation to support and streamline the provision of genomic testing and analysis across the UK. Here we explore key areas of innovation which could transform how genomic testing and analysis is delivered, such as AI and automation, as well as genomically enabled innovations which are, or could be, important for cancer, including liquid biopsy or circulating tumour DNA (ctDNA) testing, precision medicines and cancer vaccines.

To unlock the potential of innovations coming down the pathway, there are two key priorities:

1. **Ensuring that the right foundations and infrastructure are in place**. UK and Devolved Nations Governments, working closely with the NHS and Health and Social Care N. I., must act now to ensure that the genomics medicine service in each of the four nations has the capacity and capability needed to support innovation adoption.
2. **Longer term strategic thinking at national and local level** is needed to prepare the genomics system in each nation for the next decade and beyond, particularly as more personalised medicines come online.

9.1 Infrastructure for genomic innovation

Having the right infrastructure in place to support innovation, accompanied by sustainable funding for work driving innovation, is essential. In England, the Genomics Medicine Service (GMS) was established with innovation at its core, with Genomics Medicine Service Alliances (GMSAs) playing a vital role in embedding personalised medicines and new diagnostic tests, such as ctDNA tests, into mainstream care. The establishment of eight NHS Genomic Networks of Excellence in 2022 further reinforced this infrastructure, providing welcome national level leadership and coordination around priority areas such as ctDNA testing and AI, bringing together resources and expertise from across the NHS, industry, third sector and academia[102].

Sustainable funding

Whilst stakeholders we spoke to applaud the ambition and vision of Genomics England, NHS England (NHSE) and the UK Government and the flagship innovation projects in train, such as the Cancer Vaccines Launchpad, they stressed the need to further cement the supporting infrastructure for innovation within genomics. The importance of additional funding for these new initiatives was noted so they can be adequately resourced and not draw too heavily on existing capacity within Genomics Laboratory Hubs. This would help realise NHSE's ambition to develop a sustainable infrastructure across testing,

clinical services and research and innovation[103].

The GMS in England needs predictable and consistent funding from NHSE, to enable GLHs and GMSAs to prepare for anticipated service changes and invest in new equipment which could improve efficiency and lay the foundation for more innovative delivery. GMS stakeholders emphasised that they welcome the level of funding committed by the UK Government and NHSE, but as this is often only allocated towards the end of the financial year, there are time pressures attached to spending. This is reflective of the wider NHS funding environment in England, and annual budget allocations received from the Treasury. The current model of investing capital has a profound impact on our ability to scale and develop genomic healthcare, presenting challenges for planning when to buy costly new equipment, such as a sequencer, which could increase efficiency.

A lack of clarity on funding levels and funding allocation patterns for each financial year also makes conducting workforce planning harder as contracts for staff within GLHs must frequently be awarded on a six month or annual basis. Stakeholder conversations revealed that this was also often true for roles dedicated to innovation, such as Innovation Director roles within GMSAs. Stakeholders noted that this made it harder to recruit and limited post holders' ability to put in place longer term strategies and cement coordinated ways of working between Alliances, Hubs, and other relevant innovation partners. To truly embed innovation in how the genomics service is delivered in England, and fully prepare it for future innovations, service delivery posts within GLHs, and those driving innovation within GMSAs, need to be funded longer term.

Investment in the future

Two key enablers of service improvement, which will also boost genomic medicine services' ability to cope with increasing demand, are high throughput sequencing machines and increased automation. In Wales, government investment of £15 million in creating a purpose-built facility for All Wales Medical Genomics Service, Pathogen Genomics Unit and Wales Gene Park and in increasing genomic sequencing capacity through investment in equipment has been greatly welcomed[104]. The creation of a brand new, unified site for genomics delivery has laid a solid foundation for future genomics delivery and innovation, facilitating closer working between research and standard of care delivery, and provides much needed physical space for modern labs and sequencers.

In England, GLHs' ability to invest in the latest sequencers varies depending on their location and intersection with local hospital Trusts. The most up to date sequencers, which enable greater automation and thus quicker processing, require significant physical space as well as up front capital investment. As noted above, the current system of central funding allocation for GLHs often prohibits this, leaving areas with hospital Trusts who can provide investment better equipped. High demand for genomic testing and analysis is already placing significant pressure on GLHs, and demand is only set to grow. High throughput machines will play an essential role in meeting this demand and enabling delivery of certain innovative cancer treatments. The speed of test and

sequencing turnaround will need to increase as precision medicines become more commonplace in cancer treatment.

Policy Recommendations

29. To reduce regional variation in England and equip all GLHs to speed up turnaround times and increase capacity, DHSC should support NHS England and the Genomics Unit to build more stable funding mechanisms. Improved annual and multi-year budgeting is needed to deliver a sustainable GMS for the future. Any new strategic ambitions need to have funding attached to avoid GLHs and GMSAs being further overburdened.

9.2 Liquid biopsies

A promising area of innovation within genomic or biomarker testing is liquid biopsy. Genomic or biomarker tests that use blood, urine, breath, or other 'liquid' samples are being investigated for their use in detection, diagnosis, and guiding treatment options. These tests look at cancer cells from a tumour or small pieces of DNA (ctDNA), RNA, or other molecules released by tumour cells into a person's body fluids[105]. Liquid biopsy allows multiple samples to be taken over time, which may help clinicians to monitor molecular changes more easily, which could indicate response/ resistance to treatment[106]. Crucially, liquid biopsy tests could help reduce the number of patients who need an invasive tissue biopsy and provide quicker and more accurate diagnosis, as well as speeding up cancer patients' access to personalised treatments. Here we focus on liquid biopsy tests for use in symptomatic patients, rather than those in development for use in screening.

Exciting progress in ctDNA testing is being made in both England and Wales. NHSE has funded a transformation project through the NHS GMS Alliance, which is exploring the implementation of ctDNA tests in the NHS, starting in stage 3 and 4 non-small cell lung cancer patients. With the pilot underway, NHSE hopes to offer the test to 10,000 patients who have received CT scan results showing suspected lung cancer from next March[107]. Since 2017, Wales has offered a ctDNA testing service for Non-Small Cell Lung Cancer (NSCLC) patients and for patients with metastatic colorectal cancer[108]. In Wales, the QuicDNA project is evaluating the use of liquid biopsies within the lung cancer diagnostic pathway, exploring their use in improving and shortening the current pathway[109].

Liquid biopsies could also offer potential for children and young people with cancer. Gathering samples via solid tissue biopsies or surgery is invasive and can be more challenging in young patients, and repeated sampling is not always possible. Research through the CRUK-funded Stratified Medicine Paediatrics programme explored the potential utility of liquid biopsy tests for diagnosis of children and young people (CYP) with cancer[110]. It also highlighted how the use of liquid biopsy could aid rapid enrolment onto clinical trials in the future. Liquid biopsy testing could also be used to monitor CYP cancer patients' response to therapies, which could give clinicians the opportunity to

explore other treatment options sooner if patients are developing treatment resistance.

This is a fast-moving area, with many products in development, which holds significant potential for people affected by cancer. Agile processes must be established to assess these new tests, and that health systems support the development of use cases to understand where liquid biopsy tests can contribute most impactfully to cancer pathways. To ensure the successful implementation of new liquid biopsy tests, there must be adequate workforce training, the development of sample pathways and the collection of data regarding test uptake and performance that can be included in national datasets linked to patient outcomes. The UK and Devolved Nations Governments must work closely with health systems in each nation to develop robust mechanisms, underpinned by health economics models, to inform decisions around adoption and reimbursement of biomarker tests across the pathway.

9.3 Precision medicines and cancer vaccines

Advances in technology and our understanding of cancer are leading to the development of increasingly specific and personalised cancer treatments. As genomic testing of tumours has become more common, we can more accurately target drugs to the molecular biology of an individual tumour, improving the efficacy of a treatment and patient outcomes, minimising short and long-term adverse side effects. We've heard from people affected by cancer that the side effects of their treatment often cause significant trauma- it's vital that we continue to develop kinder and more targeted treatments. We also need more treatment options for people with rare types of cancer, including children and young people, for whom better and less toxic treatments are desperately needed. Examples of genomically enabled precision treatments include:

Pharmacogenomics - a treatment or dose tailored to an individual based on their genetics.

Targeted therapies - such as drugs that target specific proteins or pathways in cancer cells based on tumour mutations.

Over the next decade, greater numbers of precision medicines will become available.

Access to precision medicines

There are several issues within the genomics system affecting patients' access to precision medicines across the UK. Companion diagnostic biomarker tests help to determine whether there is a personalised medicine that might be suitable for a patient based on the profile of their tumour. Currently, there is a disconnect between the precision cancer medicines that have been approved by NICE, the Scottish Medicines Consortium (SMC) and the All-Wales Medicines Group (AWMSG) and the companion diagnostic tests required for certain drugs which have been added to national test directories[111]. As it takes time to assess, evaluate and add a new test to a test directory,

genomics medicine services must be given adequate notice of drugs that are being assessed to avoid delays in these reaching patients. Genomics medicines services and laboratories also need notice so that they can prepare to report on the relevant biomarkers to assess eligibility for new drugs. Stakeholders in England, Scotland and Wales noted that they would like to receive more notice from NICE, the SMC and the AWMSG of when a precision medicine might be approved, and that there was scope for improved join up in Health Technology Assessment (HTA) processes and the process for updating the test directories. In Scotland, stakeholders noted that this lack of join up creates time pressure to validate tests and have them added to the directory to prevent delays to precision cancer medicines delivery. As noted above, stakeholders we spoke to highlighted that this is currently delaying Scottish patients' access to precision cancer medicines. There is also a clear need for enhanced horizon scanning for genomic advances and technologies from the MHRA and HTA bodies across the UK, accompanied by earlier communication to health systems.

As noted above, testing directories are updated on an annual basis, with the option to apply for fast-track approval for tests that meet certain criteria[112,113]. As more precision medicines are being developed and approved, greater agility will be needed to ensure that test directories can be updated in line with medicines approvals, and to ensure patients have timely access to proven treatments.

Policy Recommendations

30. In all four UK nations, the MHRA and health technology assessment bodies must work with healthcare systems, genomics medicine services, and industry, academic and charity partners to ensure alignment between companion diagnostic HTA processes, test directories and new precision medicine approvals. This will be vital in speeding up patients' access to precision medicines.

31. UK Government and devolved governments must prioritise bolstering the MHRA and HTA bodies' capacity for horizon scanning for genomic advances and technologies, to ensure equitable rollout of new innovations and system preparedness for adults, children and young people with cancer.

32. The government departments responsible for health in each of the four nations should work with the National Institute for Health and Care Excellence (NICE), Scottish Medicines Consortium (SMC), All Wales Medicines Strategy Group (AWMSG) and internal partners in the Northern Ireland Department of Health, Social Services and Public Safety (DHSSPS) respectively to ensure the same diagnostic tests are approved across the UK, so patients have equal access to precision medicines as they are approved.

A whole pathway approach is necessary for the delivery of precision medicines, including holistic funding for their delivery, and for accompanying diagnostic tests where needed, not just funding for the medicine itself. HTA processes across the UK do not

capture the true cost of delivering precision medicines, such as staff time. Stakeholders also noted a similar issue when new biomarker tests are added to genomics test directories - the lack of support for implementing new testing pathways can delay their use in practice. We've heard that across the UK the omission of pathway requirements and capacity building for precision medicine delivery is placing significant pressure on services along the delivery pathway, including pathology, pharmacy, clinical genomics services and non-surgical oncology. This is leading to delays in patients being able to access new precision medicines and benefiting from these cutting-edge treatments.

It's positive to hear that the assessment of whole pathway costs is under review in Scotland. It will be important for the SMC to work closely with Health and Social Care Directorates to fully capture these costs and consider how they will be sufficiently resourced by government. In Wales work is being led by the All-Wales Genomic Service to address this challenge by mapping a whole pathway approach for precision medicine, building on the Wales Precision Medicine Strategy. In England, it is welcome that the Genomics Unit has introduced an impact assessment process to ensure that GLHs are building the required capacity to manage the workload from new genomic tests that it is anticipated may need to be implemented. This must be accompanied by support from DHSC in funding delivery costs for new genomic tests and precision medicines for the rest of the pathway.

In Northern Ireland, Action 27 of the 2022-2032 cancer strategy committed to delivering precision medicine and genomic testing in line with NICE recommendations[114]. The strategy noted that Northern Ireland lags behind other UK nations in building its genomic capabilities[115]. Stakeholders felt that a notable reason for this was funding constraints, and the way that funding for the genomics service is structured, as noted above, which makes it hard to keep pace with commissioning any new companion diagnostic tests required by new precision medicines. However, it's promising to hear that the Department of Health will be funding expanded provision of genomics testing and precision medicine for cancer patients via the Precision Medicine Centre and Genomics Medicine Centre. Governmental consideration of how sustainable this funding model is for long term genomics delivery is needed as part of the expected genomics implementation plan for Northern Ireland.

Box F

Case Study: Northern Ireland Precision Medicine Centre

The Northern Ireland Precision Medicine Centre helps translate innovations from research into clinical application. Pathology and genomics teams are integrated in the centre's laboratory, which enables teams to collaborate closely. The lab also performs innovation validation and tech transfer and holds a unique position as the only UK lab to be accredited for molecular diagnostics. The team play a vital role in horizon scanning for genomic advances and technologies, looking ahead to what's likely to be approved in the next five to ten years. This enables them to prepare to report on the relevant biomarkers, and to communicate to the regional genomics service any service changes that may be required.

Cancer Vaccines

Another form of targeted cancer treatment of significant interest are personalised vaccines. Currently in early-stage trials, it's hoped that vaccines can be used to stimulate the immune system to recognise and eliminate cancer cells and provide a foundation for other types of immunotherapies. Cancer vaccines could be transformative for patients, reducing the need for invasive treatment and offering improved outcomes. NHSE's Cancer Vaccines Launchpad, designed to support and expedite clinical research on vaccines, is a welcome initiative. But this programme – and any further transition of cancer vaccines to routine care – must not place additional strain on already overstretched pathology and genomics services. Whilst the introduction of Genomic Pathology Hubs will go some way to alleviate pressure on pathology labs and Genomic Laboratory Hubs, more planning needs to go into managing demand for the future. Mapping workforce requirements against the predicted increase in precision medicines and therapies for cancer, including how diagnostic pathways might change with the introduction of more companion diagnostic tests, will also be necessary.

Stakeholders we interviewed praised the vision behind the Cancer Vaccine launchpad, particularly the greater integration of pathology and genomics through the creation of Genomics Pathology Hubs. However, it was felt that more work was needed to unpick which responsibilities will lie with which part of the system (GMS/GMSA/GLH) and how they should work together to deliver the launchpad and future provision of cancer vaccines post-trial phase.

9.4 AI and automation in genomic healthcare

Innovative technologies have the potential to support genomics delivery and speed up testing turnaround times. There is potential to use automation throughout the processing and analysis of genomic samples, from sample loading and DNA extraction to sequencing preparation and reporting. The increased use of automation will be essential to handle growing demand for genomic testing and analysis and could free up

staff time to dedicate to other tasks, such as supporting research.

Increased automation must be accompanied by improved linkages between different platforms and systems across genomics laboratories and pathology laboratories. Stakeholders we spoke to in England identified this as a current issue hampering the effective use of automation, with a disconnect in systems in some areas necessitating staff to manually input information. This would require an uplift in IT capability and improved digital infrastructure. Another clear theme from stakeholder engagement in England was a lack of time and headspace across GLHs to plan for future developments and think about how they could optimise current working to be as efficient as possible, which has been caused by current delivery pressures. Further support from the Genomics Unit to boost GLH capacity, freeing up time and headspace for teams to identify promising innovations for service delivery will also be needed.

Longer term, AI could hold great potential in helping us to better, predict, detect, diagnose, and treat cancer. AI could be used to identify patterns from large and complex datasets, such as genomic datasets. This technology is currently in the early stages of development. Much more work is required to understand the ethical implications, build more representative genomic datasets to train AI algorithms to mitigate algorithmic bias, and to determine how these algorithms should be regulated[116]. Patients and the public must be meaningfully consulted at an early stage so that they can be involved in shaping considerations and plans for the use of AI algorithms in genomic healthcare.

Work to implement bioinformatics algorithms/decision support for genomic variant filtering and analysis, and digital reporting is underway, though implementation of these innovations is not universal. Genomics England and NHSE are already driving progress, such as partnering with the National Pathology Imaging Co-operative to deliver a £26 million innovative cancer programme which will evaluate cutting-edge genomic sequencing technology[117]. NHSE's strategic commitment to collaborate with Genomics England to evaluate the potential for the clinical implementation of artificial intelligence (AI) and machine learning in genomics, through a series of pilot schemes in the NHS GMS, is also welcome[118].

When we spoke to people affected by cancer, and participants in the involvement workshop that we ran with useMYdata, about potential future uses of AI in genomic healthcare, they said that they would want to be informed if AI was involved their care, and the specifics of how it was being used. They also noted the need to give patients information about the use of AI in genomic healthcare before it becomes a widely established part of delivery to increase comfort levels and assuage any concerns. The people we consulted recommended that information be made available in a variety of formats to suit different people's needs, including those who are time poor, as well as those who are less digitally fluent.

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