Sequencing Success: Genomics for cancer research and care

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

November 2024

Executive Summary

CANCER RESEARCH UK

About this report

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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. In 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.

Bold
Act with ambition, courage and determination

Credible
Act with rigour and professionalism

Human
Act to have a positive impact on people

Together
Act inclusively and collaboratively

Registered with FUNDRAISING REGULATOR

Executive summary

What is genomics and why does it matter for cancer research and care?

Cancer is a disease where genetic changes lead to uncontrolled cell growth that can spread through the body. Genes are sections of your DNA, and your entire set of DNA makes up your genome. Genomics is the study of someone's genome, so it is key to furthering our understanding of the fundamental nature of cancer and ways to prevent, detect and treat the disease.

Genomics offers the opportunity to shift from 'one size fits all' cancer management to a more personalised approach. For people affected by cancer, a healthcare system that fully harnesses genomic healthcare, as well as supporting lifesaving genomic research, can be game changing. This gradual integration of genomics into routine care offers an exciting opportunity to better prevent, detect, and treat cancer by:

Identifying increased hereditary risk of developing cancer

Diagnosing cancer more accurately

Getting cancer patients into the right clinical trials based on their specific disease

Helping match patients to targeted treatments

Gathering data from genomic testing that can be used to speed up research into cancer biology

CRUK has spearheaded pioneering genomic and genomically enabled research, funding programmes such as the Stratified Medicines Programme (SMP1) and the DETERMINE trial. The CRUK Cambridge Institute has driven the employment of cutting-edge genomic tools into clinical practice in the NHS, including furthering the use of personalised medicines and streamlining processes to identify where further clinical research exploration is needed[1].

Challenges and opportunities across the UK

The UK initially made great strides towards being a world-leader in genomics research, including establishing Genomics England and delivering the associated 100,000 genomes project. These built on the UK's existing research strengths, with our world-renowned universities and research institutions, health systems, and strong industry partnerships. In 2020, the four nations of the UK set bold ambitions to embed genomics in routine health and social care in the Genome UK strategy.

Now, alongside innovative digital and data advancements, genomic testing is becoming faster and cheaper, and consequently more common in cancer diagnosis and treatment. 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 could be of particular benefit to children and young people affected by cancer. Genomic research offers the potential to better understand how children and young people's cancers differ from those that affect adults, and to develop kinder, more targeted treatments.

Nevertheless, the UK's genomic systems – the tests, procedures, protocols and organisations put in place to deliver genomic tests to patients and gather insights on the use of genomics in patient care – are far from realising their full potential. The system's infrastructure was primarily designed to deliver research and rare disease testing and is struggling to support rising demand for genomic testing as a more routine diagnostic and decision-making tool. Genomic services are stretched beyond an effective working capacity.

There is significant variation in genomic infrastructure and in patient access to genomic services within and between the nations of the UK. This variation applies to:

- The specific tests available
- Turnaround times for test results and the precision medicines that rely on those tests
- Applications of testing methods and cases when testing is used to inform care decisions
- Opportunities to participate in genomically stratified clinical trials

This can impact cancer patients' care, potentially resulting in delays to a patient receiving their diagnosis and impacting decision making around their treatment.

Research and data

Variation in services available to patients can also make it hard to carry out UK-wide genomically enabled clinical trials. A more coordinated and compatible approach between the four nations is urgently needed to better support clinical research, UK-wide trial access, and address unwarranted variation in access to clinical trials. This must

include effective curation and collection of health data and improved linkage and flow with non-genomic assets so this data can be harnessed for lifesaving research.

Across the UK, a lack of integration between routine care and research in healthcare settings is making genomically-enabled research more difficult:

- Research-relevant genes aren't always reported on, impacting cancer patients' access to genomically enabled trials.
- UK trial registries often don't keep up to date with live genomically-enabled oncology trials, making it hard for clinicians to know what opportunities are available to their patients.

By addressing these structural issues, patient access to clinical trials could be improved and goals to better support clinical research can be achieved. As well as the benefits for research, evidence shows that greater research activity is associated with an overall higher quality of care, lower levels of patient mortality and improved staff recruitment and retention[2,3,4,5]. Investment will also further the UK's strengths in the life sciences industry[6,7].

Staffing

Challenges with understaffing and workforce training, retention and planning persist across the genomics delivery pathway. In each nation this poses a risk to progress in delivering genomic medicine if not addressed quickly.

There are specific professions, such as specialisms within pathology and healthcare sciences, which will require increases due to growing patient need. At the same time, competition is increasing for many scientists with skills in genomics as industry is also looking to recruit these professionals. Greater consideration of new workforce models, including the utilisation of skill mix approaches, will help meet growing demand for genomic medicine.

To keep up with the science and ensure the workforce is prepared to offer the latest advances there is a need to build a culture of continuous learning and development in the workforce. It will need to include rapidly delivering personalised treatments to embracing cutting-edge tools such as AI solutions. Initially, this work should prioritise professions that encounter genomics most frequently in their work, such as pathologists, bioinformaticians, biomedical scientists, oncologists and cancer nurses.

Time for change

In England and Wales, as the current implementation strategies come to an end in 2025 meaning governments must act to develop the genomics system. In Scotland, investment in the new genomic medicine strategy must be urgently prioritised to deliver the longer-term change needed. In Northern Ireland, significant steps must be taken to develop, fund and implement a genomic strategy that fully embeds genomics into pathways and facilitates closer collaboration between health and research organisations. We expect demand for genomic testing to keep growing and its role in

supporting cancer diagnosis, treatment and research to increase exponentially, so this planning is crucial to supporting future success.

The UK Government has a unique opportunity to use genomics as a lever to achieve its growth and health missions, and to build an NHS fit for the future. The innovative nature of genomic healthcare and research exemplifies the UK Government's position that the "revolution in data, medical technology and life sciences can be game changers for our health service if harnessed properly"[8].

The UK Government has highlighted the impact that the life sciences sector has on growth and that supporting innovation is a key driver of increased productivity in the NHS. If our genomic services are resourced appropriately, they could support faster and more accurate decision making across NHS pathways for a range of diseases.

Acting on this report's recommendations would help prepare health systems across the UK for future innovation. The UK Government's proposed NHS innovation and adoption strategy[9] and the upcoming NHS 10 Year Plan, as well as commitments in the Life Science plan[10], offer vehicles for much needed changes in England.

A similar opportunity for progress must be grasped in Wales, Scotland and Northern Ireland. Genomics should be a priority for all political parties, with elections upcoming over the next few years. We have examined the genomic structures in each part of the UK and our recommendations reflect the differing contexts in each system. Leaders must be forward looking and resolute to keep pace with advances in genomic healthcare and research and ensure that the genomic systems in each nation are delivering for people affected by cancer.

Our recommendations outline how the genomic system can be optimised for cancer diagnosis, treatment and research, to address current issues and lay solid foundations for the future. They are framed around seven overarching ambitions for genomics and cancer (see below) that must be realised across the UK if we are to move towards a genomic future. This should be coupled with longer term strategic thinking at national and local level, from governments, health systems and genomics services across the UK, to prepare the genomics system in each nation for the next decade and beyond. If national leaders act quickly to close the gap between the commendable ambition of Genome UK and the current reality, then the UK could realise its aim of becoming a genomic superpower.

Overarching ambitions

1. Mobilise genomics to detect and diagnose more cancers, earlier
2. Design effective, efficient genomic systems with a strong research culture
3. Strengthen the genomic environment for children and young people
4. Build genomic testing pathways fit for research and care
5. Support a sustainable and research-informed workforce to deliver genomic medicine
6. Develop data infrastructure that underpins the next generation of research
7. Unlock the potential of future genomic innovations

Policy recommendations

Mobilise genomics to detect and diagnose more cancers, earlier

Recommendation 1: Across all four nations, health services should commit to reporting on cancer genetic risk testing and follow-up actions like surveillance colonoscopies in a similar way to reporting for screening programmes.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Design effective, efficient genomic systems with a strong research culture

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Strengthen the genomic environment for children and young people

Recommendation 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.

Build genomic testing pathways fit for research and care

Recommendation 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.

Recommendation 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.

Recommendation 14: In the short term, 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.

Recommendation 15: In the short term 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.

***Recommendation 16*:** In the short term, 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.

***Recommendation 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 precision medicine clinical trials.

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

***Recommendation 18*:** Teams across NHS England, the Genomics Medicine Service and DHSC must actively work together on various 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.

***Recommendation 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.

***Recommendation 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.

***Recommendation 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 nursing.

Recommendation 22: The Health Research Authority, the International Standard Randomised Controlled 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.

Develop data infrastructure that underpins the next generation of research

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

Unlock the potential of future genomic innovations

Recommendation 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.

Recommendation 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.

Recommendation 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.

Recommendation 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.

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