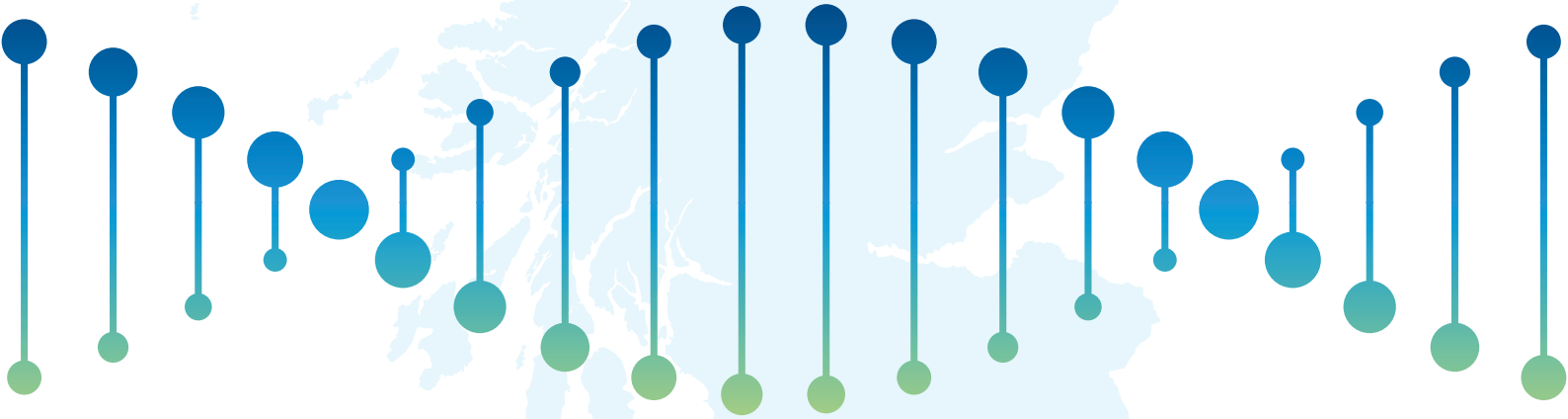
Genomics in Scotland: Building our Future

Delivering an equitable, person-centred, population-based genomic medicine service for Scotland

A five-year national strategy



April 2024

1. Ministerial Foreword

This is Scotland's first human genomic medicine strategy. In recognition of the scope for advancing services for people in Scotland, this strategy gives us a platform on which to build the infrastructure necessary for a modern and fit-for-purpose genomic medicine service, agile enough to grasp the opportunities presented by this exciting and rapidly developing discipline.

This strategy is the first step towards developing a world class genomic medicine service which will be based on the principles of person-centred care and focus on providing systems that can improve health outcomes for people in Scotland and better enable us to use genomic information to support disease prevention and early detection.

In recognition of the challenging fiscal position and immense pressure across our services, our initial implementation plan will focus on the foundational and preparatory work needed to ensure that investment and partnership working is targeted to benefit those that need it most and delivers maximum value as part of the ongoing recovery and renewal of our services.

This is one strategy, but it has clear links with many other strategy and policy areas, for example the Cancer Strategy for Scotland 2023-2033, the Rare Disease Action Plan, the Public Health Scotland Pathogen Genomic Strategy and the Data Strategy for Health and Social Care to name a few. We also remain committed to working with our counterparts elsewhere in the UK on the shared commitments under 'Genome UK: The Future of Healthcare' published in September 2020.

We will be working to make progress on all elements of this strategy. By the end of the five-year period it covers, our genomic medicine service will have the staff, skills and equipment necessary to support people across Scotland to access to the right treatment and management, by using the right genomic test at the right time.

Neil Gray MSP
Cabinet Secretary for NHS Recovery, Health and Social Care

2. Executive Summary

Genomic medicine is the use of genetic information (the instructions within our cells that shape a person's health, growth and development) to diagnose disease, guide the use of different treatments or predict the risk of disease.

In March 2023 the Scottish Government published Genomics in Scotland: Building our Future which outlined our commitment to the development of a robust national genomic medicine service. The creation of the Scottish Strategic Network for Genomic Medicine (SSNGM) is already working to bring together the genomics community across Scotland and build on existing expertise and knowledge with partnership working across the NHS, academia, industry, the third sector and our citizens.

This document is Scotland's first genomic medicine strategy and complements Scotland's Pathogen Genomic Strategy. Our vision is to establish an equitable, person-centred and rights-based genomic medicine service that can improve health outcomes for people in Scotland and better enable us to use genomic information to support disease prevention and early detection. In collaboration with the SSNGM Patient Involvement Advisory Group and service users we will work to improve people's experiences of genomic testing in Scotland, and increase public understanding of genomic medicine and the support available to them. To achieve this, we must put in place solid foundations, our building blocks, that can pave the way for an innovative and integrated genomics ecosystem that is fit for the future.

3. Strategic Aims

1. We will develop collaborative multi-disciplinary genomic testing services that include diagnostics, research, innovation, horizon scanning and funding processes. This approach will ensure the timely implementation of new genomic tests and technologies to help improve our understanding of disease and support clinical decision making.

2. We will deliver comprehensive national genomic test directories that enable healthcare professionals to provide the best possible care and treatment for people in Scotland as part of an accredited end-to-end cost-effective clinical service. By working with healthcare professionals we will support the development of national guidance and signposting to support the right test for the right person at the right time.

3. We will offer people in Scotland the best available genomic testing to support early decision making and patient care before and during pregnancy, whilst minimising risk to the mother and baby. Testing will be available, where relevant, to couples at risk of having children affected with severe genetic conditions. Urgent genomic testing in neonatal and paediatric care will be available where it will influence patient management.

4. We will explore and develop streamlined delivery models for genomic testing in partnership with healthcare professionals and service users. We will implement and expand a variety of technologies including, but not limited to, the analysis of circulating tumour DNA (ctDNA), large panel Next Generation Sequencing (NGS), Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS) where relevant within both cancer and rare and inherited condition test directories.

5. We will improve clinical management pathways for people in Scotland with cancer and rare and inherited conditions by integrating information about genomic testing and resources, and supporting new clinical practice models across Scotland.

6. We will develop a robust multi-disciplinary whole-system workforce model in collaboration with key stakeholders and service users that informs the investment, capacity, staffing levels and skills required to meet the needs of current and future genomics services.

7. We will support the design of a flexible career framework including opportunities for career development, advancement and collaboration on research, development and innovation. This will be supported by training infrastructure and underpinned by robust education programmes and educational resources to enable a sustainable, flexible and future-proofed healthcare genomic workforce.

8. We will ensure that genomics data is within scope for wider work within the National Digital Strategy to facilitate aligning this data with other healthcare data e.g. digital pathology, radiomics.

9. We will build a secure, scalable digital infrastructure to allow the analysis and storage of large-scale genomic data which will include a genomics variant repository. This will enable the sharing of genomic data across health board boundaries and with researchers. Genomic test results should be reported comprehensively in NHS electronic health records and applications, which should also be available to both primary and secondary care to inform diagnosis, prognosis and treatment planning.

10. We will develop genomics data storage and sharing solutions that are CHI linked, have 'Once for Scotland' information governance and standard data models, with appropriate consent including for teaching, audit, research and innovation collaboration.

11. We will develop a long-term genomics data and IT ecosystem that aligns with the National Data Strategy, NHS service, Public Health Scotland (PHS) and research partner requirements and identifies the required workforce to support the efficient management and analysis of NHS genomic data.

12. We will work with key stakeholders to actively promote genomics in order to increase public awareness and understanding of genomic information and raise awareness of the choices and support available. We will also support the provision of information, resources and training packages that improve access to and use of genomic testing by non-specialist clinical staff.

13. We will maximise the use of existing innovation pathways to align genomic research and innovation priorities with horizon scanning and future service requirements and improve opportunities for people to participate in genome-enabled research and clinical trials. By working with academic, NHS and industrial partners we will use an evidence-based approach to help bridge the translational gap for new innovations.

4. Acronyms

Acronym	Definition
AI	Artificial Intelligence
ANIA	Accelerated National Innovation Adoption
CHI	Community Health Index
CMO	Chief Medical Officer
CNO	Chief Nursing Officer
CPO	Chief Pharmaceutical Officer
CSO	Chief Scientific Officer
ctDNA	Circulating tumour DNA
DNA	Deoxyribonucleic acid
HCS	Healthcare Science
HCP	Healthcare Professional
HTA	Health Technology Assessment
IDA	Innovation Design Authority
LIMS	Laboratory Information Management System
NSD	National Services Division
NES	NHS Education for Scotland
NDP	National Digital Platform
NGS	Next-Generation Sequencing
NRS	NHS Research Scotland
NHS	National Health Service
NSS	NHS National Services Scotland
PGD	Pre-implantation genetic diagnosis
PHM	Public Health Microbiology
PHS	Public Health Scotland
RDS	Research Data Scotland
RNA	Ribonucleic acid
SG-TAG	Scottish Genomics Test Advisory Group
SMC	Scottish Medicines Consortium
SSND	Scottish Strategic Network for Diagnostics
SSNGM	Scottish Strategic Network for Genomic Medicine
WES	Whole Exome Sequencing
WGS	Whole Genome Sequencing

5. Contents

6. Introduction

Genomic medicine, the use of genetic information (the instructions within our cells that shape a person's health, growth and development) to inform medical care or predict the risk of disease, is transforming healthcare systems across the world.

As outlined in our Genomic Strategic Intent document published in March 2023, Scotland has a wealth of expertise and knowledge across our NHS laboratories and Regional Genetics Centres, academic centres and our thriving life sciences sector.[1] The purpose of this five-year strategy is to set out our ambition to build on these foundations and develop a comprehensive, robust and scalable national genomic medicine service and associated infrastructure, and for this to support, as well as be improved by, research and innovation.

Genomic medicine: the use of genetic information to inform and shape medical care or predict the risk of disease

Genetics: the study of individual genes, genetic variation, and inheritance in living organisms

Genomics: the study of all of a person's genes (the genome) and how they interact with each other and the person's environment

Genomic medicine is already used to determine diagnosis, prognosis and treatment within NHS Scotland, helping to guide clinical management and save people unnecessary and potentially harmful investigations and treatments in favour of therapies tailored to them as individuals. It has been used effectively for many years, particularly for people with rare and inherited conditions, to guide decision-making for them, their families and healthcare professionals (HCPs). Genomics is also transforming how we study, diagnose and treat cancer with cancer-related testing now the largest area of growth within Scotland's genomic medicine laboratories.

We also know that genomic medicine has enormous value at a population level, when integrated with national clinical data, to help guide public health, service planning and policy decision making.

In 2024, Public Health Scotland (PHS) publish a Scottish Government endorsed Pathogen Genomic Strategy to consolidate and strengthen Scotland's pathogen genomic services (concerned with the genetics and genomics of viruses, bacteria and other infectious organisms) as an integral component of public health.[2]

This current strategy is focused on positioning genomic medicine, in terms of human genetic disease, to improve health outcomes for the people of Scotland by better characterising disease, improving how symptoms are managed, guiding the choice and use of different therapies and informing the risk of disease. It is complimentary to the PHS strategy in recognising the need to work as a cohesive community around key requirements, such as workforce, and in support of efforts to improve individual and population health outcomes.

We publish this strategy in the context of an NHS in recovery from the COVID-19 pandemic and an extremely challenging financial climate. Our approach throughout this document is guided by the principles of precision medicine and the need to ensure that genomic technologies can support diagnosis and access to the right treatment

and management, at the right time, for the right person (as illustrated by Figure 1). In doing so, we need to ensure that Scotland's genomic medicine service and infrastructure can support, by design, a person-centred and data-driven approach which enables people to make informed decisions about their health.

Precision medicine (PM) aims to deliver prevention and treatment tailored to individuals' molecular characteristics. Effective implementation of PM requires seamless integration of laboratory, healthcare data, and decision support systems.[3]

Genomic medicine is a fast-moving and dynamic discipline; to build and sustain the workforce that we need in five and ten years, to develop public awareness and the infrastructure required will necessitate both a very different approach and greater partnership working across the NHS, academia, industry and the third sector. This strategy outlines the direction needed to meet our immediate and most pressing needs, and is structured around the core building blocks required to allow the population of Scotland to benefit from advances in genomic medicine going forward. Within each section we will outline our current position and context before laying out where we want to get to. Throughout, we have endeavoured to explain what these aims mean for people in Scotland more broadly.

Our initial implementation plan will focus on the foundational and preparatory work needed, in collaboration with stakeholders across Scotland, to ensure that investment and partnership working is targeted to benefit those that need it most, and delivers maximum value as part of the ongoing recovery and renewal of our health services as a whole. Thereafter we will publish subsequent implementation and delivery plans, including the performance measures that we will use to monitor and assess our progress across the term of this current strategy.

"Healthcare is rapidly moving towards precision medicine, incorporating early diagnosis and prevention as a means of improving outcomes with access to more effective treatments earlier. It presents opportunities to adopt transformative innovations at scale to the NHS, improve care for patients and communities, create high value jobs and economic growth for Scotland.

Genomics is an important component of precision medicine and, as emphasised in this strategy, we realise the full potential of precision medicine only through the integration of laboratory and healthcare data into clinical decision making, and close collaboration across clinicians scientists, patients, healthcare providers and industry."

Professor Dame Anna Dominiczak
Chief Scientist for Health

The UK strategy Genome UK: The Future of Healthcare, published in 2020, and the UK Shared Commitments, published in 2022, set the direction of travel for the UK as a whole and we continue to learn from, and share with, counterparts from other parts of the UK.[4,5] We also look to the European and international genomics community to ensure that our national genomic information meets international data standards and can be integrated into wider collaborations, increasing the number of people in Scotland who benefit through enhanced diagnostic, prognostic and preventative capabilities.

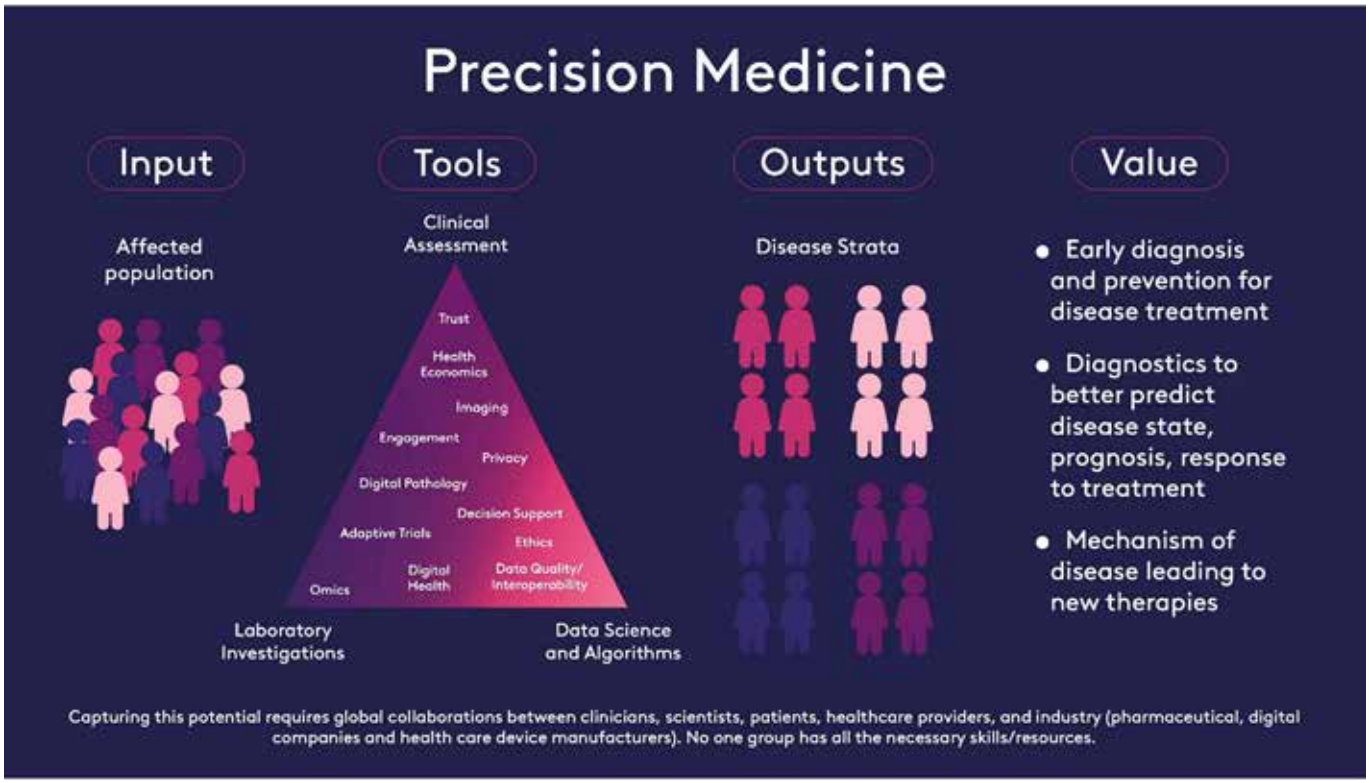


Figure 1. Visualisation of the definition of precision medicine (courtesy of Cambridge Prisms' Precision Medicine).[3]

7. Where we are now

Across the NHS in Scotland, a growing number of clinical specialties seek to use genomic information to guide diagnosis, prognosis and treatment with genomic medicine as a key component of precision medicine. Much of the demand for tests and technologies is driven by the development or licensing of new therapies, which require a genomic test, or improved knowledge about the best use of existing therapies for an individual's genetic characteristics. Advances in genomic medicine worldwide are also driving changes within clinical guidelines and the classification of disease which can lead to clinically significant diagnoses and impact on clinical decision-making. There is also a corresponding demand from clinical teams, patients and their families for interpretation and guidance about the genomic information produced by testing to help guide decision-making.

In terms of laboratory capacity, Scotland currently has four human genomic medicine laboratories located in Aberdeen, Dundee, Edinburgh and Glasgow. These are commissioned as national specialist services and delivered through a multi-site distributed service model on behalf of all Health Boards by NHS Scotland National Services Division (NSS NSD). Each Health Board contributes to the funding of this service with additional funding provided by the Scottish Government.

In 2018, the Scottish Science Advisory Council was asked by the Scottish Government Chief Scientist Office to assess the development needs for genomic medicine in Scotland. Their report recommended a national strategic approach to core issues around infrastructure, workforce and research.[6] In March 2022, NSD published a Major Service Review of the four genomic laboratories with a set of recommendations, including the need for an over-arching national strategic approach to genomic medicine and a common data environment for the NHS laboratories to connect with each other, with clinical services and key national bodies such as Public Health Scotland (PHS).[7] The outputs from both reports have informed the development of this strategy and will continue to do so throughout its implementation.

The recommendations from both reports have implications not just for the laboratories but for the clinical services that they interact with and support. In addition to the commissioned laboratories model, there are four Regional Genetic Centres across Scotland, encompassing clinical geneticists and genetic counsellors, to help HCPs, patients and their families understand genomic information in the context of rare and inherited conditions.

The volume and breadth of work conducted within the genomic laboratories and genetic centres, however, has changed significantly over the past 10 years. Rapid advances in genomic research have identified more genes that cause rare genetic conditions, requiring genomic laboratories to move towards technologies that allow the testing and analysis of either larger numbers of genes through large gene panels, or clinical and whole exomes.

The workload of the laboratories is also increasingly dominated by cancer, with referrals for testing from pathology, oncology and haematology services increasing sharply. This increase in testing has resulted in an increased workload for our pathology, oncology and haematology workforce who need to help patients understand the genomic information generated, and accommodate the changes to service delivery that precision therapies demand.

"The number of new cases of cancer is predicted to rise by 33% between 2008-2012 and 2023-2027"

NHS Information Services Division Cancer Incidence Projects for Scotland 2013-2027 report.[8]

There is now an urgent need to develop, at pace, the testing capacity and workforce to manage not just the increasing demand for genomic testing, but the associated interpretation and analysis of genomic information with and alongside healthcare scientists, clinical geneticists, genetic counsellors and increasingly other HCPs from across a wide range of clinical specialties. This trend is part of a global shift in genomic medicine from a specialist service to a 'mainstreamed' component within the wider health and social care system with many more clinical specialties seeking to utilise genomic information.

Genomic medicine in Scotland has benefited hugely from partnerships between academia and the NHS, notably the Scottish Genomes Partnership which saw collaboration particularly around rare and inherited conditions across the Universities of Edinburgh, Aberdeen, Dundee and Glasgow, the NHS, Edinburgh Genomics and on the 100,000 Genomes project with Genomics England Ltd. These projects have resulted in important legacies in terms of knowledge generation and collaboration between NHS teams and academic partners, and we need to build on these and ensure that the benefits are not only available across Scotland, but are also adaptable to different clinical indications.

Scottish Strategic Network for Genomics Medicine

At the heart of this strategy is the Scottish Strategic Network for Genomic Medicine (SSNGM). The SSNGM was established in August 2022 to provide strategic direction and develop a 'Once for Scotland' approach to the planning, design and delivery of genomic medicine services and infrastructure across the whole patient pathway. The SSNGM is commissioned by Scottish Government and led by a consultant clinical scientist and lead clinicians both for cancer and rare and inherited conditions drawing together expertise from multi-disciplinary teams of HCPs across Scotland, with accountability to both the Scottish Government and NHS Board Chief Executives. Figure 2 shows the organisational structure of the SSNGM. As part of the SSNGM, the Scottish Government has also funded a team at NSD to accelerate the transformation of the delivery of genomics services across Scotland and action the recommendations of the Major Services Review concluded in 2022. The SSNGM will play a crucial role in implementing this strategy and working with stakeholders from across Scotland to ensure that our policies can become a reality.

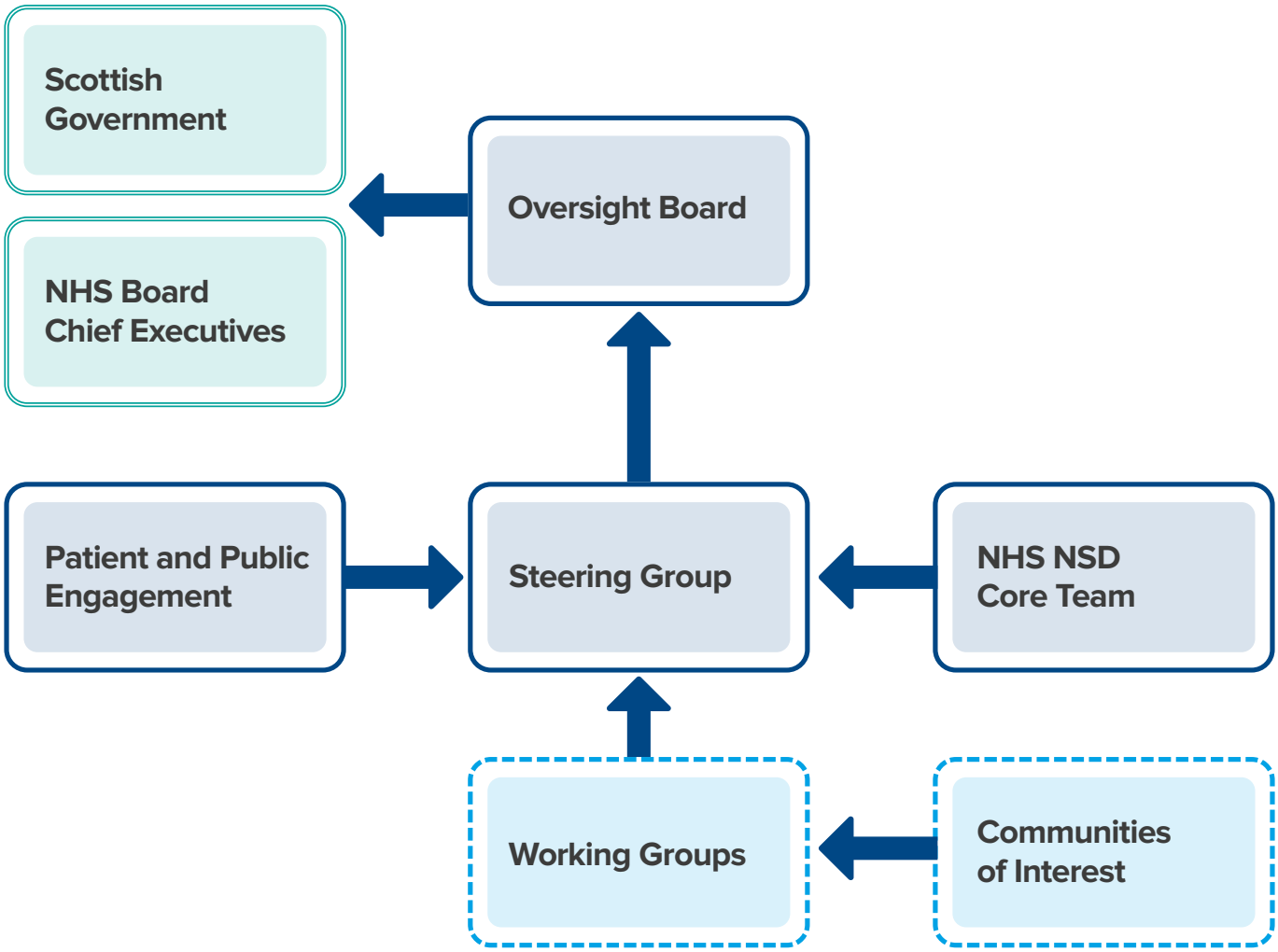


Figure 2. Organisational structure of the Scottish Strategic Network for Genomic Medicine (SSNGM).

8. Approach to Strategy Development

The SSNGM played a major role in the development of this strategy and, in doing so, developed meaningful engagement and connections with stakeholders across Scotland. Three key cross-cutting themes were identified as priorities for this strategy: 1) workforce and education, 2) data and digital infrastructure, and 3) research, development and innovation. These themes run throughout our strategy and are amongst the core building blocks identified as our primary focus and outlined below. Four specialty areas were also identified as representing urgent priorities within genomic medicine in Scotland: 1) rare and inherited conditions, 2) solid tumours, 3) haematological malignancies and 4) pharmacogenomics.

In total, seven working groups were formed covering each of these themes and specialty areas, bringing in multidisciplinary expertise and representation, and tasked with identifying drivers for change, challenges and opportunities within their field. Each group was then empowered to identify key deliverables that were required to support service improvements.

These deliverables were ranked in terms of priority, including an assessment of the critical dependencies and enablers required to support their implementation. In doing so, each group considered evidence from their own professional networks including specialist survey feedback, speakers and papers from experts across the UK and assessments of national genomic strategies both within and outwith the UK.

Figure 3: Activities and workshops informing the development of our strategic aims.

The deliverables were then aligned into strategic aims as set out within the Executive Summary. Throughout the drafting of this strategy, input was also sought from a range of stakeholders including working group members, policy makers across the UK, NHS Boards, patient advocacy and support groups, PHS, third sector organisations, academic partners and industry association groups. This approach, and the commitment from those involved across the working groups, have allowed us to develop an informed and ambitious strategy which will serve the people of Scotland for many years to come.

Acknowledgments

We want to take this opportunity to thank everyone who generously gave their time, energy and expertise to help with the development of this strategy by participating in working groups, providing expert opinion and reviewing drafts as part of the consultation process.

9. Clinical priorities

While the potential for genomic medicine is vast, the focus of this strategy is on cancer and rare and inherited conditions. These represent both areas of immediate need and provide opportunities to develop partnerships and delivery models that can be expanded and built upon beyond the terms of this strategy.

Rare conditions are defined by the UK Rare Diseases Framework as those affecting no more than 1 person per 2,000.[9] Although individually rare they are collectively common, with an estimated 1 in 17 people in the UK affected by a rare disease at some point in their lives.[9] While not all rare conditions are genetic, the vast majority (an estimated 80%) have a genetic origin.[10] Cancer, a disease of the genome, is caused when changes in a person's DNA cause cells to grow and divide uncontrollably. These changes can be inherited through families (known as 'germline' variants) which account for around 5-10% of cancer, or they can be acquired during a person's lifetime (known as 'somatic variants'). Both germline and somatic variants may influence how a person's cancer behaves or responds to treatment. Cancer can be further split into solid tumour (an abnormal clump of cells that does not contain any liquid or cysts) and haematological malignancies (cancers of the blood, bone marrow and lymph nodes). While cancer is very common in Scotland, there are many different types of cancer which on their own are considered to be rare (defined as an incidence of 6 per 100,000 people). As with rare conditions, rare cancers collectively account for more than 22% of all cancer diagnoses.[11] There are, therefore, many interdependencies and interactions across the testing, clinical pathways and services for people with cancer and with rare and inherited conditions, and many opportunities for shared learning and knowledge exchange.

Cancer genomics

As set out in the recent Cancer Strategy for Scotland 2023 to 2033, cancer is one of Scotland's biggest health challenges. Improved understanding of cancer genomics has resulted in a requirement for genomic testing to help inform diagnosis and determine the most effective clinical management.[12]

In a number of cancers, specific genetic alterations are indicators of response or resistance to cancer therapies, as well as being useful for disease monitoring and risk assessment of disease recurrence. Due to the growth in development of targeted therapies (those where a genetic marker determines benefit) there is a consequential requirement for genomic information to determine eligibility and patient selection for many clinical trials.

Based on the current trajectory, genomic testing and the use of genomic information is expected to become central to cancer diagnostics within the next 10-20 years.[13] For people with rare cancers and cancers that are more difficult to diagnose and treat, access to genomic information and expanded access to biomarker testing can be transformative in enabling people to benefit from precision therapies and management. The current use of genomic testing within teenage and young cancer (TYA) cancers and paediatric cancer as set out in Case Study 9.1. illustrates the value that genomic medicine already has, and how it is predicted to transform cancer care and management.

9.1. Case study: The application of genomic medicine in Teenage and Young Adult (TYA) and Paediatric cancer care

Diagnostics in haematology and oncology is an ever-expanding field, with new technologies being developed and implemented in clinical decision making at an extraordinary rate. Within the paediatric and young adult populations, the addition of NGS, RNA sequencing and whole genome sequencing (WGS) is changing how we predict the course of a disease (prognosis) and the likely outcomes for people affected by different types of cancer and shape decisions about how to best manage care and treatment.

A good example of this is a 9-year-old male who presented to his GP with a 1-week history of easy bruising and intermittent fevers. Blood tests demonstrated a high white cell count and he was diagnosed with acute myeloid leukaemia (AML). AML is a cancer of the blood and bone marrow that requires urgent treatment, or the patient will die. Genomics is used routinely within AML to determine disease risk and outcome upfront, as well as guide treatment decisions depending on different genetic abnormalities. This 9-year-old patient had a specific genetic abnormality associated with a more low-risk disease and he was treated in accordance with best practice. As he was diagnosed in England, WGS was performed at diagnosis and this demonstrated that, along with the genetic abnormality, he had a mutation (a variation) within a specific gene, termed cKIT. Mutations of cKIT are found in up to 50% of patients with specific genetic abnormalities and are thought to be associated with poorer patient outcomes and often a poorer response to treatment. In keeping with this, our patient had detectable disease after initial treatment and required a stem cell transplant. Following transplant, clinicians looked at his bloods and were concerned that there was a risk of disease relapse (the disease coming back). The clinicians used NGS to track the genetic abnormality and the cKIT mutation to ensure that this was not the case and he continues to be followed up by paediatric haematology services. If there had been a disease relapse with a cKIT mutation identified, a targeted treatment would have been used to give him the best possible chance of recovery. This highlights the use of genomics in assessing disease development and treatment response, and allows the potential use of new targeted, personalised treatments.

Rare and Inherited Disease conditions
Scotland's first Rare Disease Action Plan, published in December 2022, set out the priorities for improvement identified by engagement with the rare disease community in Scotland. We will work closely with Scotland's Rare Disease Implementation Board (RDIB) to ensure alignment between their aims and actions and those set out in this strategy.[14]

An accurate diagnosis from genomic testing can lead to better management of a rare and inherited condition, access to therapies or avoidance of unnecessary therapies and improved quality of life. The 'diagnostic odyssey', the time that people with rare genetic conditions can wait for a diagnosis, can vary from months to decades.

This can have a considerable impact in terms of the time and expense involved in extensive rounds of investigation, the available options in terms of care and management and emotional distress for both patients and their families.[15] Understanding the underlying genetic cause, and whether a condition can be inherited, allows people to consider their options around family planning, while being able to name a condition opens up potential access to support communities and resources. Within maternal and foetal medicine, genomic testing is also increasingly used to guide the diagnosis and assessment of a range of different congenital conditions, helping to guide decision-making on pregnancy, antenatal and postnatal management and family planning.

Beyond the benefit for expectant families and unborn children, genomic testing is an essential part of providing a diagnosis for children with developmental disorders and other serious genetic conditions, providing important information for their parents as to causation and prognosis. In some situations, this is required urgently in order to deliver the best possible care and improve people's health outcomes. Genomic information increasingly now leads to changes in treatment. Many adults also have genetic conditions where correct diagnosis and management has a key impact on health, including in inherited heart disease, cancer risk and metabolic disorders.

10. Timely and equitable access to genetic testing

We want to ensure that genomic medicine in Scotland supports diagnosis and access to the right treatment and management, at the right time, for the right person with a national genomic test directory that is harmonised with clinical management pathways.

Background
Test directories serve as a backbone to any genomic medicine service providing information to clinical teams about tests available, and where they can provide clinically meaningful information. They represent a dynamic catalogue and require continuous review to ensure that older testing methods are phased out, or supplemented, and that new tests are developed and validated for adoption in a timely manner to meet clinical need.

Where we are now
As service commissioners, NSD currently maintain two genomic test directories: the Scottish Cancer Test Directory and the Scottish Rare and Inherited Disease Test Directory.[16,17] These online directories detail the tests available to clinicians in Scotland as well as the referral criteria and turn-around times (TATs). The Rare and Inherited Disease Test Directory has benefited from over £8 million in investment since 2017, as part of the Bridge to a Scottish Strategy for Genomics project, including support for clinical exome sequencing across all four Scottish genomic laboratories and a whole exome sequencing (WES) service which is delivered in collaboration with the University of Edinburgh.

Within the Scottish Cancer Test Directory it is recognised that there are significant gaps, with some tests recommended as part of clinical guidelines or to accompany medicines accepted for use by the Scottish Medicines Consortium (SMC), which are not yet available. Across Scotland, a number of genomic tests are sent externally to NHS or commercial laboratories elsewhere in the UK or abroad because the testing technology and capacity does not yet exist either in Scotland or in the UK as a whole.

There are, however, gaps remaining in the availability of genomic testing in Scotland that we need to address urgently. As part of this strategy, we want to ensure that people in Scotland have access to the required genomic testing. In doing so we will continue to collaborate with our counterparts across the UK to maintain flexibility to send extremely rare tests to specialised centres where necessary.

Where we want to be

We will ensure that the Scottish test directories are comprehensive, taking account of developments in genomic medicine and staying responsive to clinical need. To support our longer-term service development these directories will be aligned as far as possible with both the other nations of the UK and relevant international standards to ensure that people in Scotland have access to the same medicines and standards of care.

Horizon scanning and the identification of future trends, technologies and tests

There are a wealth of resources, organisations and professional bodies that provide advance information about new tests and targets: from drug discovery, pre-clinical development and clinical trials to changes in clinical management guidelines around diagnosis, prognosis or prevention.

Robust horizon scanning for the SSNGM should not replicate this work but resource, expertise and funding are required to consolidate advance information on new tests and targets with sufficient notice to ensure engagement with HCPs, service users and the genomic laboratories. In doing so, we will work to strengthen the SSNGM as the 'front door' for genomic testing in Scotland to encourage early engagement and a collaborative approach encompassing academia, industry and the third sector.

This process will be further supported by the Access to New Medicines Horizon Scanning Advisory Board (HSAB) which has been established to identify and analyse new medicines currently in licensing and Health Technology Assessment (HTA) pipelines that are due to be considered by the SMC for routine access within the following 18-24 months.

Robust test assessment

Test directories require a robust review framework in place to evaluate and optimise existing tests as well as assess novel tests, tools and technologies identified through horizon scanning. The SSNGM has established a Scottish Genomics – Test Advisory Group (SG-TAG) for both cancer and rare and inherited conditions.

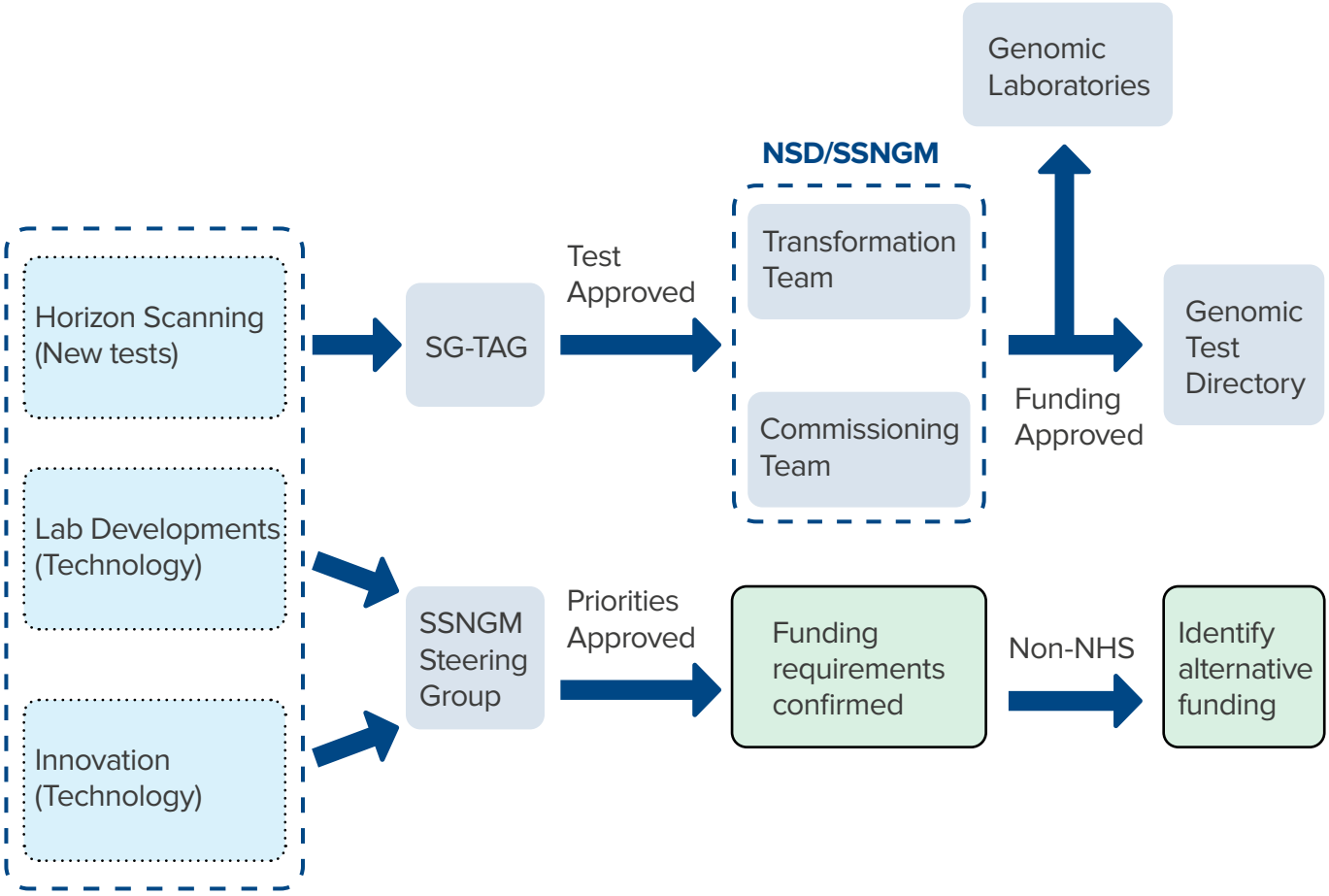


Figure 4. The SSNGM process from horizon scanning to implementation of new tests and technologies as part of the Scottish genomic test directories.

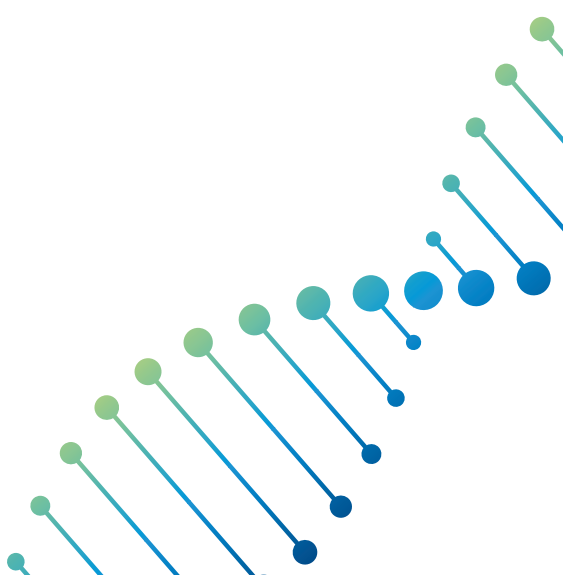
The purpose of this process is to ensure that decisions relating to the approval of new tests added to the test directories are based on the best available evidence regarding efficiency and efficacy. SG-TAG will make recommendations to NSD and the SSNGM on the clinical and analytical validity, meaningful clinical impact, patient outcomes and best delivery model for tests.

For the assessment of more complex tests and technologies, particularly in relation to rare conditions or cancers with more complex genomic profiles, we will look for additional support and health economics input via the Scottish Health Technology Group (SHTG) as well as our academic partners, recognising holistic health economic assessments as key to understanding and evidencing the impact genomic medicine has on the recovery, renewal and transformation of our NHS services.

Presently, existing funding models are a major factor in the delay between identification of new tests and their adoption into the national test directories. We will develop a sustainable funding model which is responsive and supports the timely and national adoption of tests and technologies approved under SG-TAG, including real-time delivery in line with SMC approval of medicines with associated genomic testing in collaboration with different partners.

Ensuring equitable access

As part of the SSNGM transformational programme, the Scottish test directories will be mapped against those offered in England, Wales and Northern Ireland to identify gaps and divergence. It is important to ensure that people in Scotland receive as equitable a standard of care and access to testing as the rest of the UK and that available testing is aligned as far as possible but is also responsive to clinical needs in Scotland. Where the test directories do differ in terms of test methods, turn-around times or referral criteria, there should be a clinical and scientific rationale as to why this is the case, and clear communication with HCPs, clinical networks and service users. Beyond our close neighbours in the UK we will also continue to follow the advancement of genomic medicine around the world, including engaging with life sciences companies of all sizes to ensure that the Scottish test directories remain responsive and fit for purpose. We also recognise the importance of genomic information and tumour profiling not only in supporting better care but to support clinical pathways where access to UK-wide and international clinical trials and experimental therapies are embedded within standard of care.



Expansion of genomic testing
We will work with the Scottish Cancer Network, regional cancer networks and the managed clinical networks to identify testing gaps within the cancer test directory, ensure SG-TAG review and plan for timely implementation of tests approved for adoption. We will also deliver a broader range of testing technologies across cancer and rare and inherited conditions and determine the most cost effective way of doing so. This will include the analysis of circulating tumour DNA (ctDNA) and large panel Next-Generation Sequencing (NGS) for cancer and Whole Genome Sequencing (WGS) where these technologies will deliver clinical benefit. Key to the delivery of these technologies will be collaborative working with clinical teams, service users and academic, industry and third sector partnerships.

Integration with clinical pathways
Alongside the development of the Scottish test directories we need to maintain close collaboration and ongoing dialogue between the SSNGM and genomic laboratories with the clinical teams and networks responsible for maintaining clinical pathway resources. This is to ensure that information within the test directories is aligned and up-to-date and is clearly signposted to both clinical teams and service users.

Clinical pathway resources currently exist in different formats and across a range of platforms, depending on the teams responsible for their development and maintenance. The Right Decision Service, which started out as an innovative new approach trialled by the Digital Health and Care Innovation Centre, is evolving as a go-to point for national clinical pathways and clinical decision support.[18] The service has now transitioned into business as usual activity with Healthcare Improvement Scotland, with a clear vision to scale up its use.

The Scottish Cancer Network has developed a series of clinical management pathways for breast cancer, neurological and lung cancer on the platform. Additional pathways will be added in due course. The SSNGM will look to integrate, where possible, with the Right Decision Service to signpost relevant test information, test directories and support resources within these clinical pathways. As part of the Right Decision Service, there is also scope to develop clinical decision support tools with targeted information, linked to the test directories and test requesting in a way that is consistent with wider national efforts.

The SSNGM recognises the time and effort involved in developing clinical pathways for inclusion within the Right Decision Service and will also collaborate with clinical teams, networks and service users across Scotland to integrate links or signposting to the genomic test directories into clinical pathway guidance where appropriate. We will also look to strengthen the Scottish Clinical Genetics Forum to allow them to advise on required changes in practice and improve clinical pathways in response to rapid changes in genomic knowledge and, particularly, on areas of cross-over around the management of germline (inherited) variants.

Delivery models
While genomic testing for clinical use must be delivered within accredited laboratories, there is scope for partnership with other organisations around the analysis of clinical genomic data and the reanalysis of data as new knowledge develops both nationally and internationally. The Whole Exome Service (WES) Service for developmental delay currently offered by NHS Lothian, the University of Edinburgh and the Edinburgh Parallel Computing Centre (EPCC) currently sits within the research environment (see Case Study 19.1) but has demonstrated considerable value and resulted in new diagnoses that, for affected patients and their families, have been life-changing. We are committed to translating work such as this into accredited services and exploring their potential for other clinical indications and testing technologies.

What will this mean for patients and the people of Scotland?

Timely access to the right test will shorten the time people have to wait for their genomic results. This can allow an appropriate treatment plan to be started sooner that can improve people's lives, giving better outcomes from quicker intervention.

11. Building Blocks

The previous chapter focused on the immediate need to develop testing capacity and capability. The focus of this five-year strategy must also, by necessity, be on the development of the core building blocks needed to underpin the further expansion of genomic medicine in Scotland. This includes the development of additional genomic technologies and their integration into wider health and care systems.

Without national strategic effort and investment in these core building blocks, Scotland will not be able to leverage the benefits of this rapidly developing field. The efficient delivery and expansion of technologies such as large Next-Generation Sequencing (NGS) panels, WES and WGS, and the associated need for bioinformatic support and high-performance computing, are entirely dependent upon this infrastructure.

Running across each of the building blocks set out below is the need to develop, and cultivate, partnerships across the NHS, academia, industry, the third sector and the people of Scotland, recognising within a field such as genomics that research, development and innovation, secure data access and service delivery are inextricably entwined.

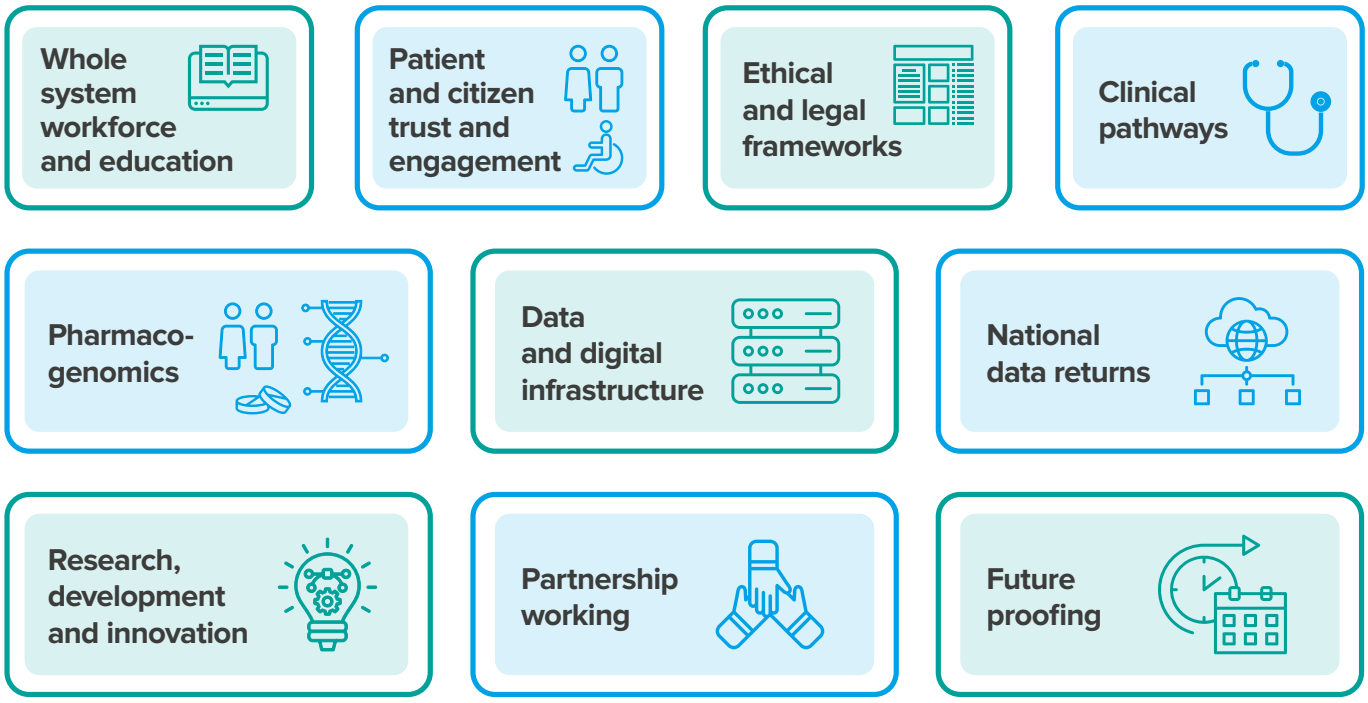
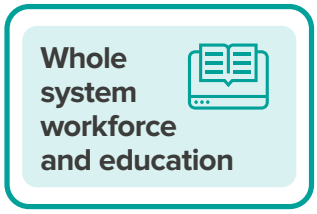


Figure 5: Our building blocks showing main themes of work set out within this strategy.

12. Whole system workforce and education



We recognise that the knowledge and expertise of our workforce is our greatest asset. As genomic medicine becomes an integral part of the health and social care system we must ensure that we retain this knowledge and expertise to support the changes needed through service redesign. We must develop a fit-for-purpose sustainable genomic workforce for the future which is supported by robust education that also spans the wider healthcare profession to better help them support patients and families.

Background

The genomic workforce is diverse encompassing healthcare scientists; including bioinformaticians, data scientists and genetic counsellors, specialist pharmacists, clinical geneticists and genetic nurses. In addition, there are numerous professional groups that interact with, and increasingly overlap and support, genomic medicine. These are outlined in Figure 6 and described in more detail in Annex 1.

In addition to the core groups described above, there are a wide range of healthcare professional groups and disciplines who need to engage with genomic medicine but require defined career pathways that are supported by access to educational resources to equip them with the knowledge and skills to enable them to undertake their roles within the genomic medicine landscape. We are aware that in some areas roles are being expanded to include genomics, and training has been developed and we need to ensure that these are brought together as part of a comprehensive workforce model for Scotland's genomic medicine service.

Where we are now

Current workforce data for healthcare science disciplines across NHS Scotland indicates that there is a developing crisis in both recruitment and retention. Scotland is not alone in this: international data indicates shortages of trained professionals with the necessary expertise and skills and there is a very real risk not only to the expansion of genomic medicine but also to the sustainability of existing activity. The number of new staff entering services through existing training and professional registration routes is not on track to keep pace with the rate at which staff are leaving through retirement, ill health or for alternative sectors, and this comes as demand for these services is projected to expand significantly. We also need to acknowledge that our workforce, both in and outwith the NHS, has also changed with more people opting for part-time or flexible working patterns, or roles that accommodate remote or hybrid working.

Where we want to be

For genomic medicine to embed into and across the wider health system we require a fit for purpose sustainable workforce which is supported by transparent career frameworks underpinned by robust education and training provision.

The Genomic Workforce
To be able to achieve such an ambition requires a whole system workforce model which will inform the skills and expertise[2] needed to develop and sustain genomic medicine in Scotland. Our future model must align with the wider healthcare science strategy; NHS Scotland workforce plans; the PHS Pathogen Genomic Strategy; and must be developed in collaboration with the Scottish Deanery and service users.

An important element within workforce modelling and resource planning will be factoring in the upstream and downstream impact of an expansion in genomic medicine service activity and technologies. We know, for example, that increased requirements around solid tumour testing will impact on pathology teams particularly around the time-intensive sample preparation favoured for sequencing technologies. We will engage with pathologists through the Scottish Pathology Network (SPAN) and the Scottish Strategic Network for Diagnostics (SSND) to ensure that requirements for additional resource are communicated and planned.

We also recognise the downstream impact of genomic testing in terms of the identification of germline variants (inherited changes) and incidental findings in patients that have implications for other family members and for the workload of clinical genetics services and oncology teams. Although the development of targeted screening and screening based on germline findings is outwith the scope of this strategy, we are committed to exploring with partners such as Public Health Scotland how to support this activity in terms of data infrastructure, data collection and information governance.

The 'mainstreaming' of genomic testing and the delivery of targeted precision therapies in cancer also requires a larger oncology and haematology workforce with more specialist genomic training. We are committed to working with the Scottish cancer networks, and the Haematology and Transfusion Scotland Network (HaTS), the Royal Pharmaceutical Society and NES to develop, signpost and support access to targeted training.

Career Framework
We want to promote genomic medicine as a rewarding career option with clear progression and prospects within a Scottish genomic medicine service. Given the pressures on existing staff and services, and universal concerns around staff recruitment and retention, there is a need to ensure that there is a requirement to address these challenges whilst ensuring that we have a robust career framework in place to attract our future workforce.

As part of this strategy and in line with the healthcare science strategy, we will develop a career pathway that is underpinned by accredited training programmes, competency models and educational support. This will be done in conjunction with the NHS Scotland Academy, NES, the Scottish Deanery and professional bodies.

To strengthen the position of genomic medicine in the wider healthcare system we will explore opportunities and models for partnership working across different sectors outwith the NHS, particularly with Scottish universities on joint clinical academic research positions as a mechanism for embedding research and innovation into service whilst providing opportunities to support staff recruitment and retention.

We will also look to explore joint roles that cross over disciplines as a mechanism for mainstreaming genomic medicine, and develop new roles and training routes, such as genetic nurses and genomic champions, to better support genomic medicine going forward.

Integration of genomics into education programmes
Ensuring that genomic medicine is well represented at undergraduate level in particular and that curricula for medical doctors, nurses and pharmacists is regularly updated to incorporate new technologies, tools and knowledge is vitally important not only for our long-term workforce planning but also to improve the genetic literacy of the wider workforce and preparing HCPs for the expansion of genomic medicine and precision medicine.

We will, in collaboration with NES and partners across UK policy teams, academia and the third sector, engage with higher education and professional bodies to ensure that appropriate training is incorporated into undergraduate, graduate and specialist professional training for medicine, nursing, biomedical scientists and pharmacists. In doing so we will seek to learn from examples of best practice across other organisations including the NHS England Genomics Education Programme and GeNotes resources, the European Reference Networks and the US National Institutes of Health (NIH).

Improving genomic literacy and expertise across the wider workforce
As well as developing innovative ways to train and develop new staff, we also need initiatives for existing health and care staff to improve their knowledge of genomics in healthcare. In terms of the clinical workforce, this is crucial to improve the confidence of staff in understanding and explaining genomic information and adapting to changes in service delivery and, in doing so, improve the experiences of service users.

We will work with NES, the academic sector, and colleagues within the Pathogen Genomic Service to develop and adapt educational resources and training materials, working in collaboration with NHS England Genomics Education Programme, the outputs of the Beyond 1 Million Genomes (B1MG) project, the European Society for Human Genetics and professional bodies both in the UK and internationally. We will also explore other methods including the use of cross-disciplinary genomics competency frameworks, for example Health Education England's Facilitating Genomic Testing: A Competency Framework and clinical decision support tools.

Signposting to information and resource
Aligned to our building block on patient and citizen engagement, we will work closely with third sector parties to maximise the availability and access to resources and educational materials for HCPs, in both primary and secondary care, and service users. We will also engage closely with existing initiatives such as The Health Literacy Place and NHS Inform to better signpost genomic medicine service resources and support, and encourage conversations about what it means for people both within the NHS workforce and across the wider population in Scotland.

Communication and Collaboration across Scotland

As part of the development of this strategy, the SSNGM is building relationships with stakeholders across Scotland. These connections will be strengthened through a range of activities including newsletters tailored for professional networks and services, engagement activities in collaboration with NES and forums to bring together professionals across the NHS, academia, industry and the third sector.

It is also important that we connect with PHS and NHS colleagues on the Pathogen Genomics Strategy and the Pathogen Genomic Oversight Group (PaGOG), as well as the Rare Disease Implementation Board (RDIB) recognising the common aims of their respective strategies around workforce and education and the collaborative approach needed to develop sustainable and vibrant pathogen genomics and human genomic medicine services in Scotland.

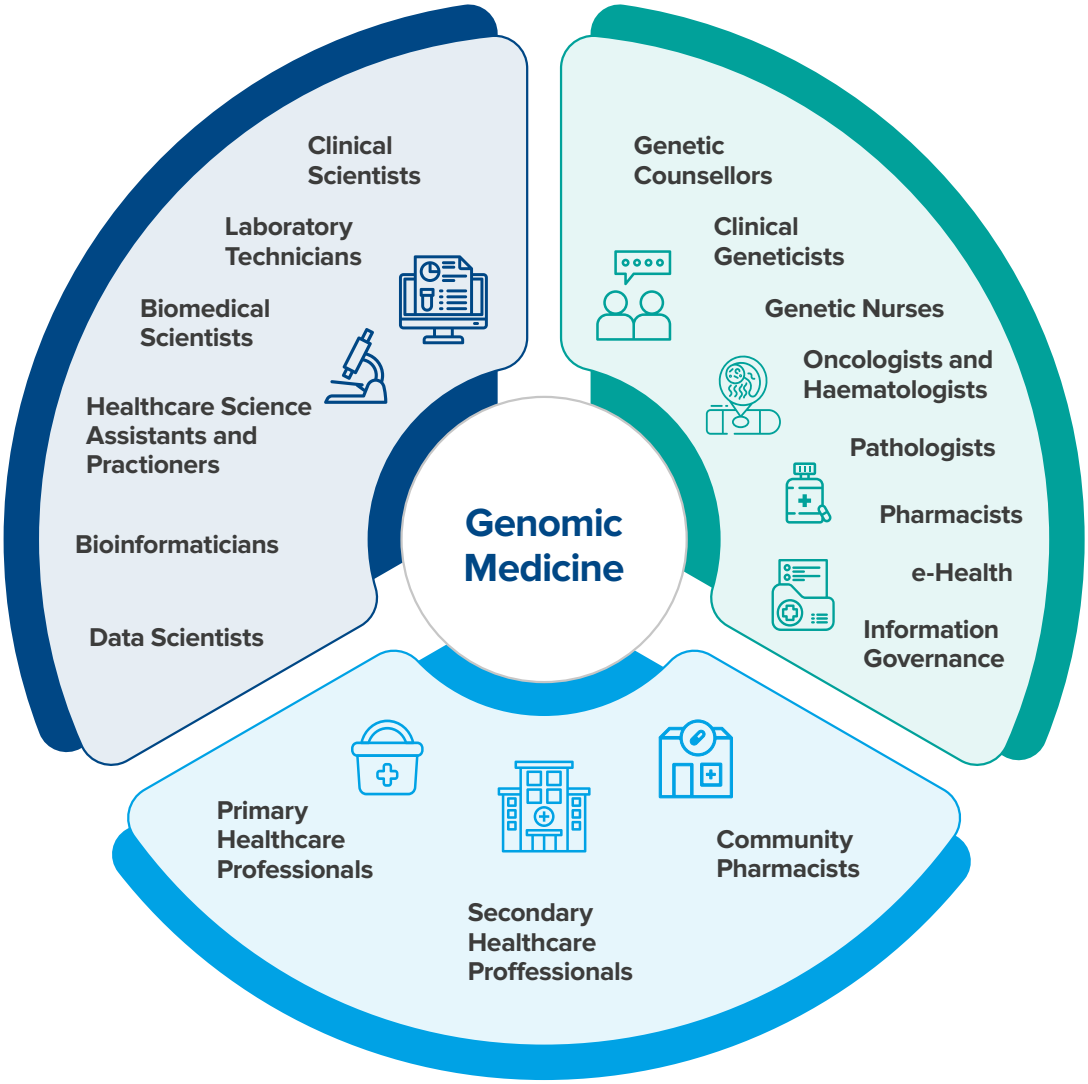


Figure 6. Professional groups working in and around genomic medicine services.

What will this mean for people of Scotland?

By establishing a workforce that has the right skills and competencies, this will ensure that the potential set out in this strategy will be translated into practice providing a high-quality service to the people who require it.

13. Patient and Citizen Trust and Engagement

We want to ensure that the people of Scotland are involved and at the centre of their care by empowering individuals and their families to make informed choices around their own genomic information. As genomic medicine develops, we are also committed to exploring how people wish to access genomic services and genomic information.

Background

A recent review of diagnostic programmes identified public support and patient education as important enablers for the take up of genomic testing, emphasising the importance of raising overall awareness of genomics and helping to support its wider use within health services with clear and accessible information and resources.[19] There is also already a demonstrable public appetite for greater information and insights from genomic data: from the rapidly expanding market for direct-to-consumer (DTC) tests promising predictive or diagnostic information, to recruitment to established research studies across Scotland such as Generation Scotland and the NHS Research Scotland initiative the Scottish Health Register and Biobank (SHARE) which now has over 300,000 people collectively helping to support a range of research projects.[20]

Where we are now

Working with representatives from patient advocacy groups and charities across Scotland, a Patient Involvement Advisory Group (PIAG) was included within the SSNGM governance structure to guide and advise on patient and public engagement activity and inform the SSNGM's work going forward.

Where we want to be

We want genomic information to be provided to people in a way that is meaningful, accessible and easy to understand, and helps them to make the right decision for them. We want people in Scotland to have an understanding of the use of genomic medicine and how it might impact them and we are committed to exploring how people in Scotland wish to access genomic services and information, recognising the need for targeted and sustained engagement with different groups.

Development of the Patient Involvement Advisory Group (PIAG)

The PIAG was formed as part of the SSNGM with representatives from professionals from across the genomic medicine community and third sector patient advocacy groups which guide our engagement with people across Scotland on the subject of genomic medicine. Developing this group, and working with them as a key component of the SSNGM, will be key to the delivery of this strategy.

Promoting person-centred care and supporting informed decisions
By working with the SSNGM PIAG, patient advocacy groups, service users and academic centres we will identify examples of best practice care that provide support for individuals and their families to make informed choices. We will explore, where appropriate, the use of novel educational resources using a range of different formats to accommodate different communication needs and preferences.

Recognising the findings of the Good Diagnosis report and the SGP project (Case Study 13.1) we will work to signpost resources developed by the third sector to help patients better understand their options, both before and after testing, and support shared decision making as part of clinical pathways.[15] In doing so we will ensure that this activity is collaborative and linked with the work of the Rare Disease Implementation Board, the 2023 Mental Health and Wellbeing Strategy and Cancer Strategy action plans and the ongoing NES Psychological Therapies 'Matrix' project to highlight the importance of people's mental and emotional wellbeing as they interact with genomic medicine services and learn about genomic test results.[12,21]

Capturing patient experience
As part of our work to improve health outcomes (detailed under Data Returns) we will assess how best to capture and measure people's experiences to define a baseline understanding for genomic medicine in Scotland that progress can be benchmarked against. As part of this work we will also assess the points within the care and reference pathways for genetic testing and counselling where inequalities are apparent and what the impact of these are.

Identifying barriers to equity of access
We will identify barriers to equity of access to genomic testing on health and racialised inequalities, particularly the use of health data and the recommendations from the Expert Reference Group on COVID-19 and Ethnicity and the findings of the NHS Race and Health Observatory Rapid Evidence Review. [22,23]

We recognise the multiple factors contributing to inequity, including location of services, the cultural acceptability of genomics, socio-economic factors, race and ethnicity, the potential for discrimination or perceptions of discrimination, and evaluation of delivery of genomic services themselves for accessibility. We will also work to develop appropriate information and educational resources for specific groups identified as having inequitable access or with racialised inequalities.

Engaging both patients and citizens in genomic medicine
We will identify best practice examples from across Scotland and from other nations engaged in educational efforts around genomic medicine and develop our engagement in collaboration with different communities. As part of this process we will seek to engage with national initiatives already underway within Scotland to empower citizens around their health and social care data, such as the Digital Front Door project, and work with partners to see how genomic medicine and genomic data could fit within these models.[24] We will also seek to link genomic medicine with wider national conversations regarding the use of health data in research, new technologies including Artificial Intelligence (AI) and Scotland's wider Data Strategy for Health and Social Care.[25]

13.1. Case study: The Scottish Genome Partnership and participant experiences of Whole Genome Sequencing (WGS) in undiagnosed rare conditions

The Scottish Genomes Partnership (SGP) was a pan-Scotland initiative to explore the application of genomics to clinical problems through a collaboration between NHS, academic and industry partners. This included an investigation, in conjunction with Genomic England Ltd's 100,000 Genomes project, of the extent to which WGS could improve genetic testing and clinical follow-up for people with rare conditions. As part of this project interviews were conducted with a subset of participants to explore what aspects of WGS were most valued for those with undiagnosed rare conditions.[26]

All aspects of WGS, including the chance of a diagnosis, waiting times for results, impact on reproductive choice, information from tests and contribution to research, were thought to be important with 'peace of mind' and 'closure' consistently cited with the value of WGS associated with reducing the stress and uncertainty of the diagnostic odyssey. One mother of a patient who had awaited a diagnosis for 29 years said "It was very difficult for people to understand how to relate to Christopher as even we didn't know ourselves what was wrong. And I, like many mothers – albeit irrationally – always blamed myself, thinking I'd done something wrong during my pregnancy. I'd carried that guilt with me, to get a diagnosis was just wonderful. I was so delighted – I was walking on air."

What will this mean for people of Scotland?

Engaging with people who are using our services ensures that their lived experience, and what matters most to them, is reflected in our priorities. Promoting person-centred care and resources will help people know what options are available, what questions to ask and how to get the most out of services that are available.

14. Ethical and Legal Frameworks

Genomic information raises a number of ethical issues and, under the UK implementation of the General Data Protection Regulation (GDPR), is classed as identifiable information. This raises particular considerations both for NHS organisations producing genomic information, and for those working with it. The implications and insights generated from genomic information, however, are difficult to predict and this impacts both on informed patient consent and consent processes. Furthermore, the value of genomic information lies in its comparison and linkage with other sources of genomic information and wider clinical datasets, which can have unintended consequences around health data inequalities and genetic discrimination.

Background

To date in Scotland, there has been a high level of public trust in the use of healthcare data for clinical services, research and population-level health screening. This is evidenced by recent publications from the Office for National Statistics (Trust in government, UK) and reports such as Data Dialogues 2 and Public attitudes to data and AI: Tracker survey (Wave 2).[27,28,29] Attitudes of participants within a Scottish Genome Partnership study on the acceptability of data access for the clinical reuse and reanalysis of samples and data within a rare disease setting was positive, with the potential risks seen to be outweighed by the benefits of new diagnostic information.[26] A Citizen's Jury convened around the potential for newborn bloodspot cards (known as Guthrie cards) to be reused for health data science and predictive medicine concluded that the benefits would be of potential value but wanted assurances around how potential research would be conducted and monitored.[30]

Where we are now

We operate within ethical and legal frameworks that govern the way in which genomic data can be used, but there are numerous areas of ambiguity that can be, and have, been interpreted differently across Scotland. Health Boards, safeguarding data as part of their legal responsibilities, approach data sharing differently and sharing genomic information for clinical service, audit and research is currently associated with significant costs in terms of staff time and resource.

A further complication is the fine line within genomic medicine between what constitutes a clinical use and a research activity, particularly in clinical pathways where research has now become an element of standard care. This was highlighted by the recommendations of the Equity of Access to Cancer Clinical Trials Short Life Working Group which described the value in cancer care of linking genomic testing with engagement in research.[31] The O'Shaughnessy review, commissioned by the UK Government into the UK commercial clinical trials landscape, also recommended that patients receiving genomic sequencing of any kind be offered a standard consent for engaging in research.[32] It is important

that any models adopted to increase opportunities for access to cancer research are, from the outset, considered more broadly to extend the same opportunities to people with rare conditions and other specialties with genomic targets as far as possible. There are a number of initiatives underway that we need to engage with and key publications regarding the ethical use of digital information (Ethical Digital Nation, Unlocking the Value of Data, A changing nation: how Scotland will thrive in a digital world).[33]

Safeguards against genetic discrimination
There are existing UK-wide safeguards around genetic discrimination, including the Association of British Insurers Code on Genetic Testing and Insurance, an agreement between the UK Government and the insurance industry stipulating that no one can require or pressure someone to have a predictive or diagnostic test under any circumstances, or ask for results of genetic testing to be disclosed, with the exception of Huntington's disease in applications for life insurance cover above £500,000.[34] There are also overarching frameworks such as the UK Government Data Ethics Framework.[35]

Where we want to be

We want to develop consistent consent models for genomic medicine services, underpinned by Scotland-wide information governance, that are transparent and easy to understand for patients and their families, and that facilitate coordinated clinical care and support research and innovation activity. We want to work towards agreed and transparent nation-wide information governance that supports robust links between the genomic medicine service, NHS Research Scotland research infrastructure, national registries and PHS.

Fit-for-purpose consent models
We will identify and develop standard models of informed consent for genomic testing that are consistent with the British Society for Genetic Medicine guidance on consent and confidentiality, and that support participation and use of genomic data within research and innovation, recognising this activity as a key driver of genomic medicine.[36] In doing so, we will ensure that consent processes are not overly burdensome for staff, patients and families, recognising the existing knowledge and expertise of NHS staff of both clinical and research consent processes. We will also work with service users to ensure that consent models are supported by accessible and appropriate information resources and support person-centred, coordinated care pathways.

Information Governance
As we develop national data solutions and digital infrastructure for genomic medicine in Scotland we will work with Information Governance (IG) personnel within the NHS and with the Information Commissioner's Office (ICO) as early as possible to ensure that governance frameworks are fair and lawful, enable wider, appropriate access to genomic data and protect against possible harms. We will work in partnership with different stakeholders around IG and data requirements to ensure that genomic data can be shared, received and integrated lawfully with NHS laboratory data, and to ensure that different models of partnership working with patients and families and other sectors including academia, industry and the third sector are considered from the beginning. We will also ensure that we develop an IG framework that is in line with our wider health and social care data strategies and define clearly what a 'Once for Scotland' approach means for individual Health Boards and organisations around genomic medicine services.

Research governance
There are well established governance controls in place to ensure that research involving patients, and the use of patient data and tissue is ethical and protects the rights, safety, dignity, privacy and wellbeing of research participants. This includes independent review of research and approval by NHS Research Ethics Committee, and the Public Benefit & Privacy Panel for Health & Social Care (PBPP). We will work with key stakeholders to support appropriate use of genomic information in research and also to ensure that they are involved in the development of consent models to identify and minimise obstacles to the use of genomic information for audit, research, development and innovation.

What will this mean for people of Scotland?

People using genomic services or tests can be confident that their data is being used safely, securely and in a legal, ethical manner appropriate to its purpose.

15. Clinical Pathways

We want to ensure that the development of genomic medicine and infrastructure within Scotland is person-centred by design to support precision medicine and coordinated care. This involves providing support and guidance to HCPs around the best use of genomic testing and information but also ensuring that this information is readily and securely available to those who need it. In conjunction with our building block around patient and citizen engagement, we will work to ensure that clinical teams, patients and families have genomic information that is appropriate and accessible, to allow them to make informed decisions about their health.

Background
Genomic information, encompassing the tests carried out, results and any interpretation, needs to be securely and readily available to those who need it across all health and social care systems. As genetic and genomic medicine impacts on ever more clinical areas, this becomes more complex as information must move across territorial Health Board jurisdictions, clinical specialties and sectors to support care personalisation and coordination. At the same time, we recognise that genomic information is increasingly complex: we need to ensure that systems are in place to allow the reinterrogation and appropriate reuse of data over time, by different groups with systems in place to provide interpretation and context where needed, linking to the wider education of clinical communities, patients and families.

Where we are now
As genomic medicine develops, it will impact upon an increasingly wide range of people, specialties and sectors across the health and social care system. Although many groups have already developed multi-disciplinary care pathways and collaborative groups to deliver services and integrate genomic insights into patient care, some variation in expertise and pathways continues to exist across Scotland.

Where we want to be
We want to build a genomic medicine service and infrastructure that supports person-centred care pathways and coordinated clinical services in which patients and their families can access support and treatment in a timely manner, and supports collaboration between professionals and different service areas to make best use of resource across a system under immense pressure.

Coordinated care
We need to ensure that genomic information and results are available to those who need them across health and care systems to help coordinate care efficiently. This is particularly acute for those who access specialist secondary care services which may be available only in a minority of Health Boards in Scotland. This can mean patients travelling outwith their home Health Board area to access services, care and support. We will work with Scotland's Rare Disease Implementation Board in support of the Rare Disease Action Plan, as well as the Scottish Cancer Strategic Board in support of the Cancer Strategy, around their plans for improving the coordination of care for patients across Scotland.[12, 14]

Wherever possible, we will work with existing groups to understand where and how they need to access genomic medicine reports and information, as well as exploring the development of new roles to support HCPs. The need for reports, and underlying data, to be accessed nation-wide and to be reanalysed or reused to support patient care will also form a core component of our initial work around information governance and data management.

National Genomic Multidisciplinary Teams (MDT)

There are strong examples of multidisciplinary team (MDT) working and molecular tumour boards (MTB) in Scotland for specific indications and, where these exist, they have played a crucial role in supporting HCPs to make sense of genomic information and helping shape patient care and shared decision-making. We also recognise that these play a vital role in educating and upskilling clinical teams about genomics in their field and its practical implications. We will work with existing MDTs and MTBs to identify areas of best practice and assess the potential for different models to be scaled up, understanding the staff time and resource needed to support these. We will also work with clinicians across different clinical specialties to help support the co-ordination of services for complex testing pathways to ensure that information governance and digital solutions, developed alongside the national Laboratory Information Management System (LIMS), can support comprehensive multidisciplinary reporting for patient diagnosis and care.

Support for patients and families

Within our workforce building block, we identified a need for clinical geneticists, genetic counsellors, oncologists and haematologists to be able to support patients and their families to understand their genomic information. We also identified a need for wider workforce upskilling and the use of novel resources to both educate and communicate with clinical groups, patients and families about what genomic information and reports mean for their condition, and their choices in terms of care and treatment. Working with the Rare Disease Implementation Board we will support the development of the clinical genetics forum, so that it can progress the clinical pathway elements of this policy and improve harmonisation of practice across Scotland. We will also work with the different cancer networks to identify where clinical pathways require clinical genetics and genetic counsellors to be involved and in what capacity, and where other professional groups and roles should be identified, signposted and developed.

Supporting precision medicine within cancer

As described above in relation to expanded testing we know that an important area of activity will be enhancing cancer precision medicine and access to SMC approved medicines. Delivery will rely on interaction between pharmacists, nurses, clinicians, laboratory services, primary care and digital teams. To contribute to this we will work closely, through the SSNGM, with partners within the SSND, Cancer Research UK Scotland Centre, the Experimental Cancer Medicine Centres, the Scottish Cancer Network, Scottish Pathology Network (SPAN) and the regional cancer networks to explore how best to embed testing within clinical pathways. Critical within this area is also engagement with pharmacy colleagues around the use and optimisation of different medicines and the interpretation of genomic information within these settings.

Prevention and early detection within and outwith screening programmes

An important area of expansion within genomic medicine is its application to the early detection of genetic conditions from pre-conception (before pregnancy) and antenatal (during pregnancy), through early childhood and beyond. Pre-implantation genetic diagnosis (PGD) is a technique used to remove a small number of cells from a fertilised embryo following in-vitro fertilisation (IVF) to test for a known genetic condition in the family. This methodology is a form of pre-conception genetic testing used before pregnancy to prevent families passing on genetic conditions to their children. Non-invasive prenatal testing (NIPT) and non-invasive prenatal diagnosis (NIPD) use a blood sample from a pregnant mother to test for a suspected (NIPT) or a known (NIPD) genetic condition in her baby during pregnancy. NIPT for the most common chromosomal abnormalities is currently offered as a second-line screening test to those women who have received a higher-chance result that their baby may have Down's syndrome, Edwards' syndrome or Patau's syndrome. NIPT is delivered across Scotland through the National Pregnancy Screening Programme. NIPD is used as a diagnostic test rather than a screening test to identify known genetic conditions which are inherited within a family and can be used for both common and rare inherited conditions.

In Scotland we want to expand our diagnostic testing that is offered both pre-conception (PGD) and antenatally (NIPD) to more couples who would benefit. There is also a need to offer a rapid comprehensive genomic test, such as WES or WGS, to try to identify genetic abnormalities in babies who present with a potential serious genetic condition either during pregnancy (antenatal), shortly after birth (postnatal) or for critically ill children. We recognise the importance of making testing available as early and as rapidly as possible to help optimise the management of these challenging cases and support appropriate treatment where possible.

In adulthood, the early detection of inherited risk of disease particularly for those families at risk of developing inherited cancers can also drive preventative efforts. We also want to evaluate the expanded use of technologies such as ctDNA as a potential test to help with the detection of some cancers.

In terms of our wider preventative efforts, national screening programmes in Scotland are overseen by the Scottish Screening Committee, which is aligned to the UK National Screening Committee (NSC). While this is out of scope for the current strategy, we will work with stakeholders as the NSC pivots to include targeted population screening and the inclusion of genomic information. We will also seek to learn from the findings of the Genomics England Ltd research study on expanded childhood genomic screening.

Infectious diseases and point-of-care (POC) testing

During the COVID-19 pandemic, genomic sequencing was used within the diagnosis and monitoring of cases, tracking outbreaks and new variants and supporting the development of therapies and vaccines. The PHS Pathogen Genomic Strategy has outlined the plan for the further expansion of genomic sequencing to identified or suspected pathogens beyond SARS-COV-2 and an important area of expansion is point-of-care testing, particularly for infections and pathogens which are resistant to anti-microbial drugs. We are committed to working closely with PHS colleagues to ensure that systems and data collection developed as part of the genomic medicine service are compatible and interoperable with POC testing for potential pharmacogenomic targets identified as a growing area of demand.

15.1. Case study: Care coordination for rare conditions

People with Friedreich's ataxia are at increased risk of cardiomyopathy, cardiac arrhythmias and diabetes. They are usually diagnosed in childhood following a genomic test and will later transition to adult health services on leaving school. The NHS Tayside Genetic Care Coordinator supports those affected by supporting them through the transition process and ensuring that they are linked into the correct healthcare team. This also ensures that they receive screening investigations such as annual blood tests and ECGs (heart rhythm recordings), as well as 2 yearly echocardiograms (heart scans) in a timely manner and that these investigations are reviewed by appropriate specialists and the results fed back to them along with a follow-up plan or care plan. The care coordinator also acts as a point of contact so that if issues or new concerns arise in between planned events, the person has access to ongoing support which can include referral onto other specialist services depending on need, such as speech and language therapy or physiotherapy. This is just one example that shows the importance of ensuring that genomic information is available across the multidisciplinary teams that need it to ensure that appropriate support is in place to help people, both before and after genomic testing and to understand genomic information. Putting in place this support and coordinated care will require collaborative working across multiple NHS services, disciplines and policy teams.

What will this mean for people of Scotland?

Co-ordinating care helps everyone. Healthcare professionals working in multi-disciplinary teams will be able to quickly share expertise and take the whole picture of a person's condition into account. People attending appointments may have less of a travel burden, and can make more out of appointments, when care is well-co-ordinated. Having clear pathways in place will also increase people's knowledge about their data or information and how to access it.

16. Pharmacogenomics

We recognise the enormous potential of pharmacogenomics for the health and social care system in Scotland with genomic information used to tailor treatment and guide the choice and use of different medicines to ensure the best clinical outcome for patients and minimise side effects and adverse reactions. The infrastructure and expertise needed to support robust pharmacogenomics is considerable and we recognise the role that Scotland's genomic medicine service will need to play as a key collaborator.

Background

The introduction of pharmacogenomics into routine clinical practice has the potential to transform the use of medicines.

Pharmacogenomics: using an individual's genetic information to maximise the safety and effectiveness of medicines

In 2022, a joint publication by the Royal College of Physicians and British Pharmacological Society called for pharmacogenomic testing, and pharmacogenomic-based prescribing, to be made available in the NHS to improve patient outcomes.[37] There is also growing clinical evidence of the effectiveness of pharmacogenomics based on testing for a wide range of medicines, with incorporation of pharmacogenomic testing included within best practice clinical guidelines for a number of common conditions affecting patients in Scotland. Pharmacogenomics is required to support prescribing decisions, optimise existing medicines and contribute to the safer use of medicines in future, regardless of who the prescriber is and where they are based across the health service.[37,38]

Where we are now

Pharmacogenomic testing is already available in Scotland and Case Study 16.1. on DPYD testing for people with cancer prior to the use of medicines known as fluoropyrimidines, outlines one application. There is growing evidence to support testing for an increasing number of widely used medications but, to date, there are no nation-wide infrastructure or mechanisms to efficiently link individual pharmacogenetic (single gene tests for individual medicines) or pharmacogenomic information (genome-wide panels supporting the use of multiple medicines) with prescribing across health and social care systems.

Multi-disciplinary collaboration across Scotland is key to the successful implementation of both pharmacogenetic testing and the more comprehensive pharmacogenomic panels into clinical practice to improve patient care. Pharmacists have a pivotal role to play, working with other professions, in the delivery of many aspects of pharmacogenomics. They are well placed to contribute in a leadership capacity to the effective and equitable local implementation of pharmacogenomics into clinical practice to improve patient care.

Where we want to be

We will support the development of a scalable pharmacogenomics service with delivery models that are both person-centred and evidence-based, with consistent Scotland-wide information governance, consent models and agreed assessment criteria to use for additional targets.

Developing an evaluation framework for new targets

We recognise the need to ensure that any new pharmacogenomic targets are evidence-based as tests are commissioned, drawing on resources available through PharmGKB and the Clinical Pharmacogenetics Implementation Consortium (CPIC) and working in collaboration with partners across the UK and Europe. There is also a need for clear structures in place at implementation to assess patient outcomes and the health economic impact, and the SSNGM will support the development of a framework to support these assessments and integrate this into the existing SG-TAG process.

Development of delivery models

The development and delivery of pharmacogenomics as an integral part of precision medicine on a national basis will require collaborative working and interaction across a range of different disciplines and organisations across Scotland. We will work closely with the Innovation Design Authority (IDA)'s Accelerated National Innovation Adoption (ANIA) pathway to support the development of a national pharmacogenetic panel (Case study 16.2). We will also continue to work with European partners to optimise the role of pharmacogenomics in routinely used medicines for non-communicable diseases (NCD), enabling a person-centred approach, so that outcomes are optimised and harm from medicines minimised.

16.1. Case study: Pharmacogenomics testing for DPYD for people with cancer who require fluoropyrimidine chemotherapy medicines

Fluoropyrimidines (including fluorouracil or 5FU, capecitabine and tegafur) are widely used chemotherapy medicines. We know that some people with cancer treated with these medicines (around 15-30%) may experience severe or life-threatening side effects because of a genetic variation in the dihydropyrimidine dehydrogenase (DPYD) gene. This gene provides the body with the instructions it needs to produce the dihydropyrimidine dehydrogenase enzyme that helps break down one of the compounds in the fluoropyrimidine class of medicines. If a person has low or no levels of this enzyme then these medicines build up in the body and can cause more side effects than normal, some of which can be life threatening. In Scotland a pilot programme for DPYD genetic testing was initiated for colorectal cancer patients in 2019 and was extended during the COVID-19 pandemic to cover all cancers eligible for treatment with fluoropyrimidines. In April 2020 the European Medicines Agency (EMA) recommended that all patients eligible for treatment with fluorouracil, capecitabine or tegafur should have DPYD genetic testing before starting treatment. Health Improvement Scotland and the Scottish Health Technologies Group have published their findings justifying the economic case for DPYD genetic testing before patients are prescribed fluoropyrimidine-based chemotherapy in October 2020. DPYD testing has been incorporated into the Scottish test directory and has been available to all eligible cancer patients[39] since July 2020.

16.2. Case study: The Tayside P4Me Clopidogrel pharmacogenomics initiative

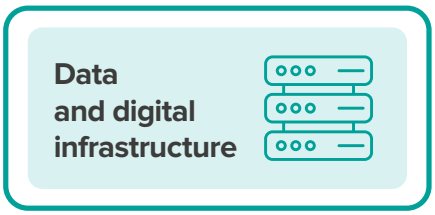
Patients prescribed a medicine called clopidogrel who are unable to metabolise or process it effectively have a significantly increased risk of stroke. An NHS Tayside pilot study, P4Me – Clopidogrel (from P4 Medicine: predictive, personalised, preventative and participatory), in the Acute Stroke Unit ran from April 2022 to March 2023, in which a genetic test specific for clopidogrel metabolism was developed to NHS laboratory standards. The test could be requested through routine hospital blood testing procedures and the ward pharmacist ensured it was carried out for all patients with an indication for antiplatelet therapy. Clinically interpretable results were returned through a service-specific email and directly integrated into electronic patient records. For people identified with impaired metabolisms, a standardised clinical decision support communication was sent to GPs with a clinically appropriate, regionally approved recommendation for alternative therapy, specified in the Local Area Formulary. The NHS Tayside Stroke Liaison Nurse team also helped ensure appropriate long term antiplatelet prescribing in the community. During the project 723 genetic tests were reported, 204 (28.2%) people with impaired metabolisms were identified and 168 (23.2%) prescribed an effective alternative. The project's success led to long-term adoption by NHS Tayside, creating a necessary foundation of knowledge and experience for more general and wider implementation of pharmacogenomics in the NHS.

Gene testing for clopidogrel is now being considered for adoption by the IDA's ANIA pathway as a potential pathfinder project for the development of a wider national pharmacogenomics panel.

What will this mean for people of Scotland?

Targeted treatment that suits the individual and their condition has the potential to be transformative for many people. Increasing the understanding of pharmacogenomics will allow more people to benefit from treatment that can improve their lives, and promote the safer and more effective use of medicines.

17. Data and Digital Infrastructure



Developing national solutions for the management, storage and analysis of genomic data and the implementation of secure and scalable infrastructure is the biggest challenge faced within genomic medicine in Scotland, and one that underpins our ability to deliver on almost every one of our strategic aims. A national, federated model is needed to digitally connect the genomic laboratories, ensure genomic data and reports are available across the health and care system and to support population-level healthcare screening and decision-making.

Background

The success of a genomic medicine service is grounded in the ability to securely collect, test, store, process and analyse samples and data for diagnostic, prognostic and predictive testing. It also needs to support reanalysis as new knowledge develops, to allow reuse across different specialties throughout a patient's life and be accessible for research as a key driver for innovation. Genomic datasets in humans can be large and, as the national test directories expand and include new testing technologies, the volume and complexity of this data will increase significantly, and at pace.

Where we are now

In keeping with health and social care systems across Scotland, the genomic medicine laboratories have developed and implemented different IT and eHealth solutions over time. The NHS genomic laboratories exist within virtual silos: unable to connect digitally with one another, reliant on local variant repositories and with no national data return or integration with national registries.

Scotland has well-established research centres with high-performance computing capacity but there is an urgent need for national-level storage and services capable of supporting high volume data and analysis. The Scottish Government has committed, through the Digital Health and Care Strategy and the Health and Social Care Data Strategy to improve technology and infrastructure across health and social care services to support recovery and reform.[25,40] It is vital that the digital infrastructure and data solutions developed for genomic medicine are harmonised with the wider digital infrastructure in Scotland.

A pilot project within the National Digital Platform (NDP) MediaStore is in development with an Application Programming Interface (API) to test the transfer of genomic data between laboratories. This has intersected with a wider transformational project around data standardisation across the genomic laboratories, as part of the groundwork for the national Laboratory Information Management System (LIMS) currently under development.

Recognising the need for more than just a genomic data repository, the Scottish Government supported the development of the WES service which, using a novel partnership between NHS Lothian, the University of Edinburgh and the EPCC, has shown the benefits for a clinical service of interacting with an academic core of bioinformaticians and access to a software-rich, high-performance computing environment but this service remains within the research space. We know that the value of genomic medicine to individual patients, and for the population as a whole, lies not only in its ability to link with other sources of data at a national level, but in the ability to share genomic intelligence across the UK, Europe and internationally to support clinical care and research and innovation.

Where we want to be

We want to develop a secure, scalable infrastructure to allow large-scale genomic data storage and analysis with the ability to use data across all Health Boards, laboratories and academic centres that is compatible with the wider Scottish Government Digital Strategy and Data Strategy. We want to support coordinated care by ensuring that results are available both across NHS services and a federated data model which allows data to be shared securely across the UK and internationally where appropriate.

Data standardisation and adoption of core data standards

We will continue to support the work of the SSNGM transformation team in standardising the data generated across our genomic laboratories to ensure that they can work effectively as part of a joined-up service, and to support national-level data returns. We will work with the National Data Standards Board and in alignment with international data standards, such as the Mondo Disease Ontology and those developed by the Global Alliance for Genomics and Health (GA4GH) to support data sharing across the UK and beyond. The use of common data models and standards across all data systems which operate across the genomic laboratories are fundamental to the gathering of genomic intelligence and translation into datasets that can be used for research and support policy-making.

Laboratory Information Management System (LIMS)

A nation-wide LIMS is technically complex but the advantages are significant: supporting sample receipt and tracking, standardised test ordering and reporting as well as enforcing data standards by design. A consortium of Health Boards have commissioned a national LIMS and are developing a bespoke genomics module with input across the genomics laboratories, and in collaboration with pathology laboratories.

Development of a secure genomic data repository within the NDP

The National Digital Platform (NDP) is a central component of Scotland's national digital infrastructure which will allow real-time data and information from health and care records, and the necessary tools and services needed, to be accessed securely and safely. Long-term, the potential offered by the NDP and the applications it can support are promising. A pilot NDP MediaStore project is intended as an initial step and we will work across Scottish Government and with NES to explore the development of a genomics data repository architecture. We will also identify solutions for known technical challenges around the need for high-capacity networks to support data transfer across Health Boards and large-volume data storage capacity. As work in this area progresses, opportunities to share lessons learned should be taken to the mutual benefit of human and pathogen genomic workstreams.

Genomic Variant Repository
The identification of medically-significant variants and the use of genotype-phenotype resources are important tools in making sense of the huge volumes of information that genomic testing generates. Within the genomic laboratories, in keeping with most clinical laboratories, clinical scientists use large genomic data sets, their own local laboratory and clinical data and variant libraries and *ad hoc* data mining. This can result, however, in differences across laboratories in interpretation of similar genomic abnormalities since complete data sets are not centralised or available across Scotland. Furthermore, our understanding of the relationships between a target and a disease are not always fixed and interpretations can change over time for some genomic abnormalities as genomic intelligence increases. We want to explore the need for a Scottish NHS variant repository.

Software-rich high-performance computing environment
Central to a genomic medicine service infrastructure will be the ability not only to store and share genomic data but the ability to analyse it within a high-performance computing environment and employ different software solutions for data analysis. The 'last mile' of analysis is dependent on the identification of raw genomic data, generation of sequencing reads (primary analysis) and alignment (secondary analysis). This is vital for the expansion of testing technologies such as WGS, WES and large NGS panels which generate complex genomic information in greater volumes. We recognise that this component is needed as a matter of urgency and may need to involve commercial applications or novel solutions such as AI/machine learning interacting with, or alongside the NDP structure.

To this end, we will conduct an options appraisal to scope the requirements, delivery timescales and potential for integration as part of wider digital infrastructure. In doing so we need to build on the capability and expertise gained from the work outlined in Case Study 19.1 and the world-class High Performance Computing (HPC) environment managed by the EPCC. Genomic data file formats (for example BAM and VCF file types) are standardised and well-supported across both academic and industry software which will help mitigate the difficulties in moving data between commercial and open-source systems.

Data federation: decentralised data model with data governance standards defined centrally and a shared data infrastructure layer to support data sharing and interoperability across organisations and disciplines

Genomic reports and interpretation
While the components outlined above focus on the genomic information generated by laboratories and subsequent analysis, there is also a pressing need to consider access to genomic diagnostic reports and interpretation. Reports are currently stored within individual Health Board Scottish Care Information (SCI) Store systems, frequently in PDF formats that require complicated efforts to share or data-mine the vital clinical information that they contain or which follow reporting standards that are not designed with patients as the intended end users. We need to ensure, in conjunction with the wider SSND, a whole-system approach is taken so that diagnostic information is securely accessible to those who need it across health and care systems, including primary care. We need to consider, as a whole, how these reports and the reporting standards used fit with the wider ambitions to provide access for patients to their own health records.

Data interoperability
Underlying these components is the importance of data interoperability; the ability to integrate genomic data with other health and care data in Scotland at a local, regional and national level, and to link up across the UK and internationally to share genomic intelligence. In Scotland, we have the Community Health Index (CHI) which is used for health care purposes and uniquely identifies a person. The CHI acts as a principal means of recording demographic information for patients within the NHS. A technical change programme is underway to revise the CHI and make it more flexible and functional across the wider health and social care system. Within the development of data solutions and digital infrastructure, there is a need for CHI-linked genomic data, in conjunction with common data models and standards, that can support the gathering of intelligence and translation into datasets for research and integration with other sources of data to allow real world evidence studies of the value of different genomic technologies and tests across Scotland.

Working to address racialised inequalities within data systems
The Expert Reference Group on COVID-19 and Ethnicity established by the Scottish Government in 2020 to consider the impact of COVID-19 on minority ethnic communities considered evidence of systemic data inadequacies, risks and harms within Scotland.[22] There is also a known lack of diversity within genomic data and variant repositories and structural biases within many new tools and technologies. We will work with the Anti-Racism Observatory to ensure that infrastructure and processes developed to support genomic medicine in Scotland do not perpetuate racialised systemic inequity and that there are clear guidelines on how race and ethnicity data is collected and used responsibly as risk markers rather than risk factors.

Data for research, innovation and service improvement
We will also find ways to capture data on genomic medicine service processes and clinical outcomes to assess their reach, effectiveness, adoption, implementation and maintenance, in conjunction with work around a national genomic data return as detailed below.

What will this mean for people of Scotland?

Having a fit-for-purpose data infrastructure will ensure that people's genomic data is stored safely and securely. It will also enable scientists and clinicians to get the most out of working with data to improve people's lives, with interoperability across systems to enable greater collaboration.

18. Genomic data returns and building a robust evidence base

We want to ensure that genomic data can be incorporated into national datasets and registries to support screening, population-based health surveillance and research and, in doing so, develop an evidence base to gauge the impact of genomic testing and the use of genomic information on patient outcomes.

Background
Genomic data has potential diagnostic, prognostic and therapeutic value across a wide range of clinical disciplines. To support its wider adoption and mainstreaming across health and social care systems we need evidence around patient outcomes, and the health economics of specific testing technologies within clinical care pathways.

Where we are now
There is currently no national data return for genomic medicine. This means that patient-level data generated by NHS genetic laboratories cannot, at present, be integrated into national registries or made available for population-based studies. During the COVID-19 pandemic, colleagues within the Pathogen Genomics Service demonstrated the enormous value of integrating data within a secure environment to other key national datasets. In doing so, national organisations such as PHS have been able to use genomic intelligence to inform policy and public health at the local, regional and national level.

Where we want to be

We want to develop a genomic medicine service for the people of Scotland that is data-driven, and will allow us to assess the impact of genomic testing on outcomes for patients, families and the wider population. Such a service will ensure that new testing and technologies are effective and enable the use of genomic data in research and for population-level screening.

A national minimum data set for genomics and integration into PHS as a national data return
We will develop, in conjunction with PHS, a national minimum dataset for genomics that can be linked with the Scottish Cancer Registry and Intelligence Service, the Congenital Conditions and Rare Diseases Registration and Information Service for Scotland (CARDRISS) and other national datasets, including the national Picture Archiving and Communication System (PACS).

The development of this dataset will form part of the development activity supporting the creation of a genomics module within the national LIMS, to ensure that specifications focus not only on individual patient management, but include capacity to support summary data for national returns. This will require a complimentary investment in the specialist data management and analytical knowledge and skills needed to handle this new data. In doing so, we will develop a vital building block that will enable genomic data to be used at a population-level to aid efforts around screening and service audit, to support research and innovation and to ensure the development of a genomic medicine service grounded in a data-driven approach. It is also important that we work closely in collaboration with PHS on the Pathogen Genomic Strategy to ensure that both human and pathogen genomic data can be brought together more effectively and to ensure transparency around the collection and use of race and ethnicity data.

Development of robust outcome measures
We will consider, in conjunction with the SSNGM Patient Involvement Advisory Group and other partners, the use of different outcome measures, focusing on individuals, families and health systems to include qualitative person-centred measures alongside quantitative data returns.

Health economics and evaluation cycles
Evaluating the health economics of new tests, technologies and delivery models within genomic medicine will be an important part of our approach to achieve value for money and allow us to build a robust evidence base for the benefits of genomic medicine and its wider applications. We will develop models for assessment and build evaluation frameworks in collaboration with the Scottish Health Technologies Group (SHTG) and academic partners.

What will this mean for people of Scotland?

Linking genomic data to other registries will be mutually beneficial, and will help provide a comprehensive source of information for genetic conditions in Scotland. This can help inform decisions about priorities for investment and research, allow scientists and clinicians to collaborate more easily, and all of this will help improve the lives of people in Scotland.

19. Research, Development and Innovation

We aim to develop the genomic medicine service and infrastructure in such a way that research, development and innovation is built in and facilitated by design, and strengthens connections across the NHS, academia, industry, the third sector and citizens about research opportunities, needs and infrastructure.

Background
Scotland has academic, NHS and industry centres of research excellence with a wealth of knowledge and expertise in innovation in both genomic medicine and pharmacogenomics. The development of genomic medicine in Scotland has benefited enormously from collaboration across the sectors and, in particular, partnership working between Scottish universities and the NHS. This has been exemplified by the Scottish Genomes Partnership, the WES service and the CSO Precision Medicine Alliance Scotland programme. Scotland has established a number of resources supporting genomic research including SHARE and also participates in a number of important UK-wide research initiatives including UK Biobank, and is supporting plans for Our Future Health.

Where we are now
While there are examples of excellent research, development and innovation in genomic medicine and collaborative working across Scotland as exemplified by the Scottish Genomes Partnership, there are opportunities to improve health outcomes through greater innovation and translation of research into clinical practice. The Chief Scientist Office (CSO) has funded a series of research projects under the Precision Medicine Alliance Scotland (see Case Study 19.2) including the i-Diabetes Platform led by NHS Tayside for enhanced phenotyping of patients with diabetes for Precision Diagnosis, Prognosis and Treatment. Genomic research is often an integral part of standard care, and not easily separated from clinical services. Within cancer care pathways, for example, patients who have exhausted all treatment options may have access to new experimental therapies via industry or academic clinical trials, while those with rare conditions can benefit from advances in genomic knowledge that may result in new information many years after they were originally tested. Patient-level and population-level genomic data needs to be accessible, securely and safely, to enable research and innovation that results in improved patient outcomes and better standards of care.

Where we want to be

We want a genomic medicine service that includes research and innovation at its core with clinical services directly informed by research, and clinical services also supporting research activity. Research roles should be included and recognised as an essential element. To do this, we want to ensure that genomic data can be integrated with registry and national data returns with a consistent consent model that supports research use of data. We also need to join up more efficiently with existing innovation adoption pathways to promote collaboration and research activity across the NHS, academia and industry, with effective use of translational spaces to better test and integrate innovation.

Integration of genomic data into national returns accessible for research

As described above, a key objective is to ensure that both patient-level and population-level standardised genomic data forms a national data return under PHS. We will work closely with Research Data Scotland (RDS), PHS and supervisory authorities to ensure the incorporation of genomic information within national and UK metadata catalogues and explore how best to streamline its use for research, development and innovation, with RDS and the Scottish Safe Haven Network acting as the gateway for research access.

Consent models and information governance

There are already established processes in place for the use of national health datasets held by PHS for population-based analyses. We will also implement a model whereby consent for use of patient-level genomic information to support research, development and innovation can be incorporated within genomic testing pathways. We will work closely with the SSNGM Patient Involvement Advisory Group, service users, RDS and supervisory bodies to understand what this would mean in practice and to identify needs and concerns. We need to ensure transparency around the security, privacy, ethical use and process for using genomic data in research by different partners. In doing so, we also recognise the need to work with communities across Scotland that have been under-represented in genomic research to build and sustain trust. We will also ensure research, development and innovation is incorporated into wider IG discussions around the uses of genomic medicine data solutions and digital infrastructure.

Translational spaces

Recognising the key role of innovation, and translation of research into clinical service, we will work closely with existing structures within Scotland to identify potential opportunities as part of horizon scanning and to develop projects identified by our genomic medicine services. The core infrastructure that we will support and engage with include Scotland's end-to-end innovation pathway, consisting of three Regional Test Beds for mid-translational work and the Innovation Design Authority (IDA)'s Accelerated National Innovation Adoption (ANIA) pathway for innovation adoption. We also recognise the importance of working with academic centres, industry partners and organisations such as the CRUK Scotland Centre and the Living Laboratory for Precision Medicine particularly around knowledge exchange and the urgent need to support cancer testing for clinical trial targets and novel approaches to trial design and set-up.

Supporting clinical trial equity of access

Recognising the findings of the Equity of Access to Cancer Clinical Trials Short Life Working Group, we will work with colleagues across government, NHS Research Scotland (NRS) and other key partners such as experimental cancer medicines centres and the CRUK Scotland Centre on the recommendations for 'genomically enabled' clinical trials.[31] Central to this is reducing geographical disadvantage for these clinical trials, by using a 'Once for Scotland' approach. This will include developing mechanisms for national approval and delivery of trials, that can then be available to all eligible patients in Scotland, irrespective of the coordinating centre. Another key focus of the group will be the identification, commissioning and adoption of new targets on a nation-wide basis to support and drive recruitment to research and clinical trials across Scotland.

Partnership working and joint job roles
Research and innovation, including translational work, is recognised as important to staff recruitment and retention within the NHS. Translational work is also of great interest within academia and industry. As part of our wider workforce modelling, we will explore ways to implement innovation and collaborate with academia and industry across the NHS to encourage partnership working, skills development, and knowledge exchange (as demonstrated within Case study 19.1.).

Collaboration around strategic aims
The SSNGM has a crucial role in supporting research, development and innovation in genomics and will work with stakeholders across Scotland to identify key priorities. We will work with the CSO to support, where possible, the identification of national and international funding calls where a national or collaborative approach could be used to support the strategic aims outlined within this strategy. We recognise that industry in particular has a key role in driving innovation within genomic medicine and we will look to work with industry to ensure that genomic medicine can thrive in Scotland in accordance with international standards.

19.1. Case study: The Whole Exome Sequencing (WES) service for early diagnosis of children with severe undiagnosed developmental disorders

The WES service was established for the accurate early diagnosis of infants and young children presenting with severe undiagnosed developmental disorders. The traditional diagnostic pathway (which can include a routine biochemical screen, a basic chromosome evaluation and other investigations such as MRI and muscle biopsy) is often delivered over a period of years and typically only leads to a diagnosis in <10% of patients.

The application of trio-based WES:

- Expedites patient diagnosis, often within an acute hospital setting which may improve prognosis (the likely course of a disease or condition)
- Offers an improved diagnostic yield (the likelihood of a test providing the information needed to make a diagnosis)
- Avoids a protracted care pathway involving multiple tests and treatments and the distress and inconvenience of iterative testing experienced by patients/families
- Provides accurate information for families on the risk of having another child affected with the same disorder. This can help families to decide if they would like pre-implantation genetic diagnosis (PGD) or prenatal diagnosis (PND)

The test amplifies all of the genes in the human body and detects between 45,000 and 50,000 variants per patient. This is then filtered against genes known to cause developmental disorders, by frequency, by consequence and then compared to the variants present in the parents in order to distil any causative variant/s that may be present.

The service started in July 2019 and since then 1032 patients have been tested. A diagnosis has been made in 30% of the patients with causative variants being detected in 279 different genes. The service also regularly re-analyses data if any improvements are made to the analysis pipeline or new causative genes are added to the gene list, which has resulted in another 8 families receiving an answer and a diagnosis for their child.

19.2. Case study: The Precision Medicine Alliance Scotland (PMAS) programme

The PMAS programme aimed to develop and deliver Precision Medicine approaches in the NHS to the diagnosis and treatment of conditions that disproportionately impact those living under difficult socio-economic circumstances across Scotland.

Following an open funding call and an independent expert review, four projects were funded in 2021 in critical care, diabetes, liver disease and multiple sclerosis (MS) with a total investment of £10 million over 4 years:

- TRAITS: Time-critical precision medicine for hospitalised adults with acute illness to efficiently generate evidence on patient-centred outcomes and support a programme of discovery (Professor Manu Shankar-Hari, University of Edinburgh/NHS Lothian)
- iDiabetes platform: Enhanced phenotyping of patients with diabetes for precision diagnosis, prognosis and treatment (Professor Ewan Pearson, University of Dundee/NHS Tayside)
- Centre for Precision Cell Therapy for the Liver: Using precise advanced therapies to target the liver to treat severe liver disease, reduce liver mortality and morbidity and the need for liver transplantation (Professor Stuart Forbes, University of Edinburgh/NHS Lothian)
- Precision-MS: Integrating precision metrics of brain health into early treatment of MS to help people make more informed treatment decisions about their disease (Professor David Hunt, University of Edinburgh/NHS Lothian)

What will this mean for people of Scotland?

Integrated, collaborative research that enables greater participation helps to realise the potential of genomic medicine for patients. The more that we participate in ground-breaking research, the more we can remain agile and adapt to the constant developments in genomics, and the more people stand to benefit from improved outcomes.

20. Future-proofing our service and building capacity

Throughout this strategy we have referred to the importance of putting in place robust building blocks on which Scotland's genomic medicine service can develop and innovate. We need to ensure, as far as possible, that these foundations are future-proofed and scalable by design to allow us to expand and enhance Scotland's genomic capability at pace and work towards an integrated genomics ecosystem that can best improve patient and population health outcomes.

Background
In each of the sections we set out our ambition for critical infrastructure and identifed the underlying components and harmonisation work that we need to establish as key enablers for genomic medicine in Scotland. Running in parallel with these efforts we need to optimise and build capacity across our laboratory infrastructure and change the funding models underpinning this activity.

Where we are now
Our genomic laboratories, consisting of four laboratories based across Scotland, were inspected as part of an NSD Major Service Review published in 2022. The review concluded that, while each laboratory delivered a high-quality service for NHS Scotland, the organisation of the laboratory network as a whole was no longer fit for optimal service delivery and that change was needed to support the laboratories to expand.

Within current commissioning arrangements, the Scottish genomic laboratories are categorised as specialist services. Through these arrangements we have seen considerable investment both from Health Boards themselves as well as the Scottish Government directly. Current arrangements are based on a point in-time assessment of the demands of genomics services which can, however, hinder their ability to account for future growth and expansion. We also know that our current model is not able to take full advantage of national partnership opportunities across academic, industry and the third sector because of the way in which they are both commissioned and structured.

Where we want to be
We want to build sustainable and scalable infrastructure able to adapt to the rapidly evolving discipline of genomic medicine. As part of these efforts, we want to build capacity across our laboratory network to optimise service delivery, develop our capability around changing genomic technologies and develop robust financial models able to support and sustain this change.

Capacity building
We will embed genomic medicine into clinical pathways so that capacity is built holistically across a number of clinical specialties. We will seek to develop an infrastructure that will deliver now and allow growth with the advances and innovations anticipated over the next 10 years and beyond. This means considering novel and innovative solutions to build capacity within the term of this strategy.

Working with the SSNGM, NHS Board Chief Executives and the genomic laboratory network, we will identify ways to optimise our laboratory delivery model, ensuring we are utilising the expertise and infrastructure we already have in place to deliver a scalable, cost-efficient genomic service. This will include considering how we take advantage of Artificial Intelligence (AI), digital solutions and opportunities available to us outside of our current NHS delivery models and in collaboration with partners across academia, industry and the third sector.

Next-generation sequencing (NGS) panels
A key priority for cancer service delivery is the expansion of NGS panels and the use of more comprehensive, larger gene panels. Working with the SSNGM and the laboratory network, we will support the development and validation of NGS panels to enhance cancer testing services. In doing so, we also need to develop and appropriately resource processes to allow for the regular review and revision of panel targets, to align with best practice and remain agile in the context of a fast-paced research environment.

Development of whole genome sequencing capability
We recognise the importance of using and developing the most up-to-date genomic technologies and we will develop our testing strategies and clinical pathways to accommodate these, including whole genome sequencing (WGS). It is important that we develop both our capability and capacity to ensure that we can be agile in adopting new and more effective technologies at the earliest opportunity, and translating these into clinical care where there is clinical benefit.

Fostering collaborative partnerships
We have already acknowledged the importance of proactive horizon scanning to allow us to plan services that are agile and responsive to change. To do this, we must foster collaborative partnerships across the NHS and bodies such as NES and Health Improvement Scotland, with academia, the third sector and industry. We have seen many examples of the benefits of academic collaboration on genomic medicine throughout the UK in developing services and technologies that benefit patients and families. While in Scotland we have benefitted from academic partnerships, we recognise that we need to strengthen our relations with industry as key stakeholders within genomic medicine and develop a systematic approach to matching service needs with industry offerings. We will work to build partnerships with different stakeholders for the benefit of the people of Scotland while ensuring transparency and the principles of providing value for our public services are maintained.

Funding and Commissioning
Genomic medicine is no longer a specialist service and is core to the delivery of our wider health services. The growth of genomic medicine requires a funding model that can react quickly, ensuring that new tests approved through our SG-TAG process can be funded and implemented to benefit patients in a timely manner.

The current commissioning model however does allow us to take a 'Once for Scotland' approach to the delivery of the genomic medicine service. We will implement alternative funding models which sustain the national approach our services have already benefitted from and which are tailored to genomic technologies. We will develop a structure which allows the continued investment from Health Boards as well as the Scottish Government, but look to also include partners that can support and enhance the capacity of our services. We want to develop services which are not only equitable across the UK but are internationally renowned.

Environmental sustainability
The climate footprint of healthcare globally in 2021 was estimated to be 4.4% of the global net greenhouse gas emissions (around 2 gigatons of carbon dioxide equivalent).[41] If it was a country, this would make the healthcare sector the fifth largest emitter on the planet. Genomic medicine's contribution to this is significant: through sample shipment and processing, laboratory management, biobanking, data use and storage. There is significant scope for change. Pharmacogenomics, for example, has the potential to reduce wastage of medicines and reduce hospital admissions relating to adverse drug reactions and represents a significant opportunity to reduce the environmental impact of medicine use. As we develop the building blocks of a national service and infrastructure, there will be opportunities to build in sustainability by design and learn by linking in with existing efforts.

NHS Scotland recognises the need to reduce the environmental impact of its operations with a Climate Emergency and Sustainability 2022-26 Strategy aiming for NHS Scotland, as an organisation, to reach net zero by 2040.[42] Overall progress across NHS Scotland is assessed annually using the National Sustainability Assessment Tool (NSAT). As part of this five-year genomic medicine strategy we are committed to assessing the climate impact of our services and infrastructure and seeking ways to mitigate this impact, looking at examples of best practice across the NHS, academia and industry, and engaging with current initiatives such as the Laboratory Efficiency Assessment Framework and My Green Lab certification.

What will this mean for people of Scotland?

Having structures in place that are agile, able to respond to developments and build capacity will ensure that more people in Scotland benefit from the advances in genomics in the years to come. This ensures we are taking a true 'Once for Scotland' approach where patients can access appropriate testing and treatment regardless of their Health Board area.

21. Next steps – implementation & governance

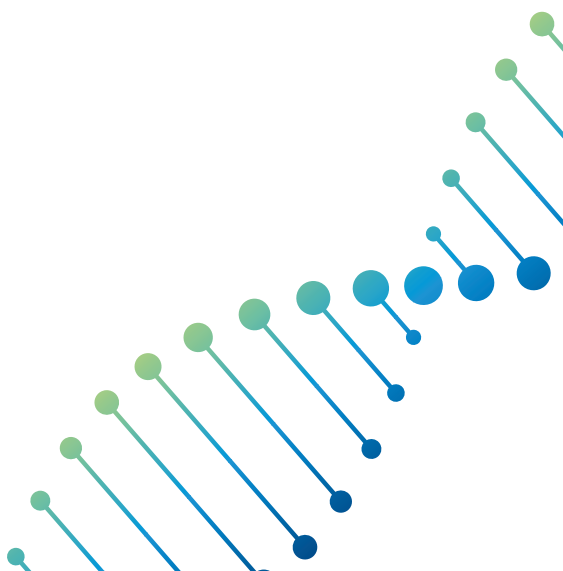
Genomics in Scotland: Building our future is a five-year national strategy which sets out to address short-term urgent needs and lay the necessary foundations for our genomic medicine service in the future. In doing so, we will use the three horizons model to provide an adaptive framework for our future growth and innovation, with further detail of each phase to be published within implementation plans throughout the term of this strategy. The first of these will focus on the foundational and preparatory work needed, in collaboration with stakeholders across Scotland, to ensure that investment and partnership working is targeted for maximum impact and value as part of the ongoing recovery and renewal of our health services as a whole.

The longer-term innovation to embed genomic medicine within the wider health and care system and create greater opportunity to benefit from genomics will be beyond the scope of this strategy but is dependent on the development of the foundations set out within this document.

This strategy supports and aligns with a number of national strategies and frameworks, both within Scotland and across the UK. Within a field as multi-disciplinary as genomics, there are a number of dependencies and limitations. Integrating with national structures and organisations and collaborating with a wide range of stakeholders is critical to the success of this strategy.

As part of delivery we will develop a set of performance measures to monitor and report on both the implementation of this strategy and the delivery of genomic medicine services in Scotland.

In developing this strategy for an equitable, person-centred, rights-based and population-based genomic medicine service for Scotland, our underlying principles and our overall ambition is to encourage different sectors and partners across Scotland to collaborate, innovate, invest and engage as one towards common goals.



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Annex 1: Overview of our current genomic medicine workforce

Healthcare Science workforce

Clinical Scientists

Clinical Scientists, registered with the Health and Care Professions Council (HCPC), are core professionals within genomic medicine and are responsible for interpreting genomic results and authorising all diagnostic reports issued by NHS Scotland genomic laboratories. They are vital in helping clinicians to understand complex genomic information and how this can be communicated to patients and guide decision making. Clinical Scientist training in Genetics and Molecular Pathology in Scotland is a three-year modular training programme developed and overseen by senior clinical scientists within the genomic laboratories. This training was developed and is delivered on behalf of NHS Education for Scotland (NES) and follows many of the requirements of the National School for Healthcare Science's (NSHSC) Scientist Training Programme (STP) although is not formally accredited. Upon completion of the three year programme, trainees can choose to progress through either the Academy for Healthcare Science (AHCS) Certificate of Equivalence route or the Association of Clinical Scientists (ACS) Certificate of Attainment to achieve registration with the HCPC as a Clinical Scientist. A workforce planning exercise undertaken in 2018 by the genetic laboratories predicted that a minimum 10 trainee clinical science posts in Scotland per annum would be required to keep pace with staff retirement and future service growth. With increasing demand for service provision and the move towards more flexible working patterns, this number is understood to be significantly higher now.

Assistants and Practitioners

This group of laboratory staff are essential to the operation of the genomic laboratories as they deliver all the diagnostic testing and generate the information that is then interpreted and reported by Clinical Scientists. HCS Assistants and Practitioners have no formal training programme and train on the job and there is no formal registration in place or clear career progression routes for technical staff beyond NHS Band 7.

Biomedical Scientists

Biomedical Scientists (BMS) undergo formal training and are registered with the HCPC. Although there is an option for BMS to become HCPC registered via the Institute of Biomedical Science (IBMS) competency portfolio, there is currently no specialist portfolio to allow BMS staff to train within molecular pathology and genomic medicine. There is considerable work to be done around the role that BMS can play in the genomic medicine service with limited BMS staff in experienced roles able to deliver service-specific training.

Data Science and Analysts

The establishment of national genomic data returns and the integration of data with existing registries and other national datasets is dependent on data managers and data analysts with specialist knowledge and skills. Training routes vary substantially but data scientists typically train in computer science, data science or a related field followed by post-graduate education, professional certification and in-house training.

Bioinformaticians and Data Scientists

The volume and complexity of genomic data means that bioinformatic and data science support is crucial and this need will only increase as testing expands to include large panels, WES and WGS. Across the four genomic laboratories, bioinformatic support is in place within the NHS in only two Health Boards and the remainder access bioinformatic support via collaborations with academic institutions. The training routes for bioinformatics and data science are diverse and are transferable across the NHS, academia and industry, to an extent that retention of bioinformatic staff within NHS genomic medicine services has been acutely problematic.

Genetic Counsellors

Genetic counsellors work with patients and families to offer genetic and genomic information and support that enable them to make decisions about their health and care. A diverse range of HCPs enter the field and the training route is via a master's level degree. The majority of genetic counsellors in Scotland were trained via Scotland's sole MSc programme in genetic counselling. This course, although fully accredited by the UK and European professional genetic counselling boards, was forced to close to new entrants in 2022 because of staff shortages.

Medical workforce

Clinical Geneticists

Clinical genetics is a medical specialty that focuses on the investigation, diagnosis and management of rare and inherited disease, and assessing the risk of some conditions based on a person's genetic make-up. Training is accessed via the UK-wide Physician Higher Specialty Training Recruitment programme, with annual trainee numbers set for Scotland by the Scottish Deanery. Medical registrars begin training in Clinical Genetics at Specialty Training level three and train on average for six years within three training centres in Scotland (Aberdeen, Edinburgh and Glasgow). Workforce planning carried out in 2022 by the Clinical Genetics Forum predicted a shortfall in newly qualified registrars by 2030, a problem compounded by a UK-wide shortage of clinical genetics consultants, with the number of trainees in Scotland fixed at seven per annum since 2013.

Genetic Nurse Specialists

A small number of nurses are employed as genetic nurse specialists in Scottish centres. They provide specialist nurse expertise to patients with a wide range of genetic conditions rather than being single condition-specific. These nurses have a diverse range of roles that can include care coordination, patient follow-up, patient support, research recruitment, multidisciplinary team (MDT) and joint clinic working. The range of roles is dependent on the needs of patients and the availability of health and social care services to meet those needs within the Health Board area. This model of service can be part of a very effective mechanism for delivering coordinated patient care.

Pathology
Depending on the route of referral, genomic tests may also be interpreted by medically qualified pathologists and discussions of the implications of genomic data for diagnosis, prognosis or therapy will often need to involve molecular pathologists, haematologists or pathologists from different specialities to ensure that findings are fully considered and implemented. Within particular testing pathways, there will also be changes to the preparation of samples and new techniques required to ensure that genomic analysis is viable and valid. There have been a number of initiatives within the pathology community and the Royal College of Pathologists to prepare pathologists for genomic medicine but challenges remain in terms of the delivery of that training, the resourcing of more time-intensive techniques and coordination across more complex clinical pathways.

Pharmacy and pharmacogenomics
As genomic medicine technology develops and our knowledge about the role of genomics in the metabolism of different medicines increases, this will be an area of significant growth at a 'whole system' level. It is essential that pharmacy teams are integrated into the multidisciplinary approach for implementation of pharmacogenomics and provide clinical and professional leadership across Scotland. The Royal Pharmaceutical Society have issued a position statement with recommendations to support pharmacogenomics and implementation across healthcare systems which details the role of pharmacy professionals within this area.

Oncologists and Haematology
As genetic and genomic information becomes an increasingly important part of cancer care, patient-facing oncology staff are already playing a significant role in the interpretation of genomic data, patient communication and counselling, clinical decision-making and the delivery of targeted therapy. Oncologists, haematologists, and nurses across both oncology and haematology urgently need tailored support to help improve their skills and confidence in integrating genomic medicine into oncology practice and supporting genomic MDTs. At the moment there are no established training pathways or accredited programmes in place, and no formally trained specialist staff within the workforce able to support the mainstreaming of genomic medicine into oncology and haematology services. As other 'mainstream' medical specialties increase their use of genomic testing and, as already seen within oncology and haematology, genomic information shapes service delivery, the same challenges around training and education will be encountered.

Annex 2: Glossary

Biomarker	A molecule, gene or characteristic that can help identify disease or process.
Chromosomes	Chromosomes carry genetic information from cell to cell. Each chromosome consists of a single molecule of DNA, wrapped around many proteins. The number and/or structure of chromosomes is known to be altered in certain genetic conditions.
Circulating tumour DNA (ctDNA)	Fragments of DNA present in the bloodstream, originating from cancerous cell and tumours.
Cytogenetic	The study of chromosomes and their inheritance from parent to child.
DNA	Deoxyribonucleic acid, or DNA, is a long molecule that contains genetic information. This information is encoded in the sequence of individual subunits, or 'nucleotide bases', which make up the DNA.
Exome	Those parts of the gene which contain protein information are known as exons. The exome consists of all exons in the genome.
Genes	A gene is the basic unit of inheritance, passed from parents to children as DNA. Each gene is found in a specific section of the genome and most contain information to produce a specific protein. Differences in the sequence of nucleotide bases mean that there are different variants of each gene.
Genetics	The study of genes, genetic variation, and inheritance in living organisms.
Genetic testing	The use of a laboratory test to examine an person's DNA for variations. In a medical setting, the results of genetic testing can be used to confirm or rule out a suspected genetic condition, to assess the likelihood of parents passing on a genetic variation to their children or to study the genomes of cancer tumours.
Genome	The entire set of DNA instructions in a cell, unique to each person.
Genomic medicine	The use of genetic information to inform and shape medical care or predict the risk of disease.
Genotype	A description of the specific variants of a gene that an individual has. DNA sequencing and other methods can be used to determine a person's genotype.
Germline	Characteristics or changes (germline variants) that are inherited through families.

Haematological malignancy	Cancers of the blood, bone marrow and lymph nodes (leukaemias, lymphomas, myelomas) that occur when abnormal cells grow uncontrollably and interfere with the normal functioning of blood cells.
Inherited	Since DNA is passed from parents to children, genetic variants and any associated characteristics as passed from parents to children.
Molecular genetics	The study of heredity and variation at molecular level.
Molecular pathology	Molecular and genetic approach to the study and diagnosis of inherited disease, cancer and infectious disease.
Next-Generation Sequencing (NGS)	DNA sequencing establishes the sequence of nucleotide bases in a specific sample of DNA. NGS refers to the use of technologies for sequencing DNA that became available after the completion of the Human Genome Project, which are cheaper and faster than their predecessors.
Non-invasive prenatal testing/diagnosis (NIPT/NIPD)	NIPT can be offered to those whose pregnancy may be at risk of a specific monogenic condition or to determine the sex of a foetus in the context of an x-linked condition. In both NIPT and NIPD a maternal blood sample is taken to determine genetic conditions and the main difference is that NIPD is considered a diagnostic test but NIPT can be a screening test meaning that follow-up invasive diagnostic tests can be required.
Pre-implantation genetic diagnosis (PGD)	Pre-implantation genetic diagnosis (PGD) is a technique used to remove a small number of cells from fertilised embryo following in-vitro fertilisation (IVF) to test for a known genetic condition in the family. This methodology is a form of pre-conception genetic testing used before pregnancy to prevent families passing on genetic conditions to their children.
Pathogen	A bacteria, virus or other microorganism that can cause disease in humans, animals or plants.
Pharmacogenomics	Using knowledge of an individual's genotype to give them suitable medicine in order to maximise safety and effectiveness.
Phenotype	An individual's observable characteristics (such as blood type). A person's phenotype is determined by their genotype and environmental factors.

Precision Medicine	Precision Medicine (PM) aims to deliver prevention and treatment tailored to individuals' molecular characteristics. Effective implementation of PM requires seamless integration of laboratory, healthcare data, and decision support systems. Developing and maintaining such a platform relies on global collaborations between clinicians, scientists, patients, healthcare providers, and industry (pharmaceutical, digital companies and healthcare device manufacturers). Governance frameworks must protect and unite patients and communities (equity, public trust, data protection, and privacy), and that research, development, and innovation (rapid open publication, adoption into healthcare systems) are aligned to industry and policy developments (intellectual property protection, regulation, and cost-effectives) so that clinical adoption is rapid.
Protein	Proteins are structures produced from genes, which have many different functions within the body and determine many of our physical and biological characteristics.
RNA	Ribonucleic acid, a molecule present in all living cells. Its acts as a messenger, allowing protein to be produced from DNA genes.
Sequencing	As in genomic sequencing: a process by which the sequence of nucleotide bases in DNA is determined for individual genes or entire genomes.
Solid Tumour	Also known as solid cancers. Abnormal growth of cells that do not contain liquid or cysts.
Somatic	Characteristics or changes (somatic variants) that are acquired during a person's lifetime.
Trio sequencing	Family trio sequencing is a powerful technique that can explain genetic conditions by looking at differences between the exome or genome of an affected child and that of their biological parents.
Variant	DNA sequence differences among individual people or populations. Some variants influence specific biological functions.
Whole Exome Sequencing	Genomic technique for sequencing all of the protein-coding regions of genes in a genome (known as the exome).
Whole Genome Sequencing	Genomic technique for sequencing the complete DNA sequence of an organisms genome – the entire genetic code.

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