

# **Genomics in Scotland: Building our Future**

**A strategy for delivering an equitable, person-centred, population-based genomics service for Scotland:**

**Implementation Plan 2024-2025**

**Year One**

# Genomics in Scotland Strategy Implementation Plan

The Genomics in Scotland: Building our Future strategy sets out Scotland's commitment to the development of a robust and sustainable national genomic medicine service. 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.

The plan sets out the thirteen overarching aims from the strategy and details the initial actions that efforts will be focused upon over the following 12 months. A Scottish Government policy team is in place to ensure that the plan is implemented effectively, underpinned by the Scottish Strategic Network for Genomic Medicine (SSNGM) and its Patient Involvement Advisory Group. The plan also sets out the key delivery partners who will work towards the delivery of these initial actions. This is by no means an exhaustive list. As emphasised throughout the strategy document, partnership working across academia, the NHS, the third sector, industry and the wider public are essential to the implementation of every aspect of this work and we will work to identify opportunities and engagement across each of the actions outlined below.

A key element of the initial year will be the development of a core set of performance measures to monitor progress against the overall strategy and to more effectively benchmark Scotland's genomic medicine service. We will report on these on an annual basis across the five-year term of the current strategy.

Thereafter we will publish subsequent implementation and delivery plans, building on the initial foundational work set out within this document.

## Year One actions

The table below outlines the overarching strategic aims and the priority actions for implementation within the initial year. These actions are intended to begin the foundational work, collaboration and consultation needed to ensure that investment and efforts across later implementation plans are targeted for maximum impact.

Reporting on progress against these actions, and the proposed performance measures, will be included within the first annual update and progress report which will be published in 2025.

### Priority: Timely and equitable access to testing

**Strategic Aim 1:** We will develop collaborative multi-disciplinary genomic testing services that include 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 genomic disease and the clinical management of patients.

Actions	Timescale
Establish a test approval pipeline from horizon scanning to assessment as a single point of entry for onboarding of new tests.	In progress
Develop a sustainable funding and commissioning model to support the timely development and implementation of new tests and indications, moving away from the current approach of releasing funding on a case-by-case basis.	Initiated 2024-25
Integrate health economics within complex funding and service delivery assessments.	Initiated 2024-25
Establish links with the Scottish Pathology Network (SPAN) to streamline sample processing to lay the foundation for ongoing pathway development and improvement.	In progress
Quantify gaps between Scottish test directories and those offered elsewhere to inform future requirements for Scottish test provision.	In progress

**Priority: Supporting healthcare professionals with test directories and appropriate signposting**

**Strategic Aim 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.

Actions	Timescale
Support healthcare professionals to use existing test directories with clear referral criteria and signposting to request forms and guidance.	In progress
Education at requesting level to support healthcare professionals to order the right test at the right time.	In progress
Working with healthcare professionals across Scotland to embed testing within end-to-end clinical pathways with appropriate turnaround times agreed.	In progress

**Priority: Support early decision making and patient care before and during pregnancy**

**Strategic Aim 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.

Actions	Timescale
Scope options for provision of urgent genomic testing in neonatal and paediatric care.	Initiated 2024-25

**Priority: Explore and develop delivery models for expansion in genomic testing**

**Strategic Aim 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.

Actions	Timescale
Options appraisal of service delivery models and infrastructure needed to support expansion of genomic testing and adoption of new technologies.	Initiated 2024-25
Explore options and supporting mechanisms to incorporate clinical trial targets.	Initiated 2024-25

**Priority: Clinical management pathways**

**Strategic Aim 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.

Actions	Timescale
Develop protocols for diagnostics and reporting to support multidisciplinary team (MDT) and molecular tumour board (MTB) working in cancer and rare and inherited diseases, incorporating lessons learned from the establishment of haematology MDT and a solid tumour MTB pilot.	Initiated 2024-25
Support the development and signposting of national guidelines for hereditary cancer.	Initiated 2024-25
Support the development and signposting of national guidelines for rare and inherited diseases involving genetic testing.	Initiated 2024-25
Support national efforts to link genomic test results with wider healthcare systems to facilitate access to surveillance pathways.	Initiated 2024-25

### Priority: Workforce modelling

**Strategic Aim 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.

Actions	Timescale
Development of a national workforce development and training plan for staff supporting genomic medicine in conjunction with other workforce training developments in Scotland.	Initiated 2024-25
Development of a laboratory workforce plan.	Initiated 2024-25
Map and scope existing bioinformatic support and develop requirements in parallel with service delivery modelling.	Initiated 2024-25

### Priority: Workforce education, training and development

**Strategic Aim 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.

Actions	Timescale
Map genomic training and educational resources available within Scotland and available through collaborations with UK partners.	In progress
Access secured to existing educational resources developed by NHS England and third sector organisations for healthcare professionals across Scotland, with appropriate signposting.	Initiated 2024-25
Collaborate with UK partners to support the ongoing development and maintenance of educational materials.	Initiated 2024-25
Recommendations for job planning and educational needs for clinical and scientific staff to support MDT/MTB working.	Initiated 2024-25
Development, in collaboration with UK partners, of requirements for a bioinformatics training programme.	Initiated 2024-25

**Priority: Alignment with National Digital Strategy and allied healthcare data**

**Strategic Aim 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.

Actions	Timescale
Ensure laboratory data is in a standardised format for sharing nationally.	In progress
Support the implementation of a new genomics module within the national Laboratory Information Management System (LIMS) programme.	In progress

**Priority: Development of digital infrastructure**

**Strategic Aim 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.

Actions	Timescale
Assess the feasibility of an NHS Scotland genomic variant database.	Initiated 2024-25
Options analysis of solutions for a centralised data store for NHS genomic data.	Initiated 2024-25
Options analysis for a common data environment aligned to the service delivery model, data storage solution and information governance policy to allow laboratories to share data and workload.	Initiated 2024-25

**Priority: Development of information governance and consent models**

**Strategic Aim 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.

Actions	Timescale
Development of an appropriate and proportionate consenting framework for genomic testing.	Initiated 2024-25
Development of a data retention policy for genomic data within NHS Scotland.	Initiated 2024-25
Develop a 'Once for Scotland' Information Governance policy that ensures that the appropriate governance is in place to support genomic data storage, sharing and use.	Initiated 2024-25

**Priority: Development of a genomics data and IT ecosystem for Scotland**

**Strategic Aim 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.

Actions	Timescale
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.	Initiated 2024-25



### Priority: Public engagement and awareness

**Strategic Aim 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.

Actions	Timescale
Identify opportunities to engage with the Scottish public about genomics.	Initiated 2024-25
Identify and signpost appropriate support and guidance.	Initiated 2024-25

### Priority: Maximise research, development and innovation

**Strategic Aim 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 genomic-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.

Actions	Timescale
Establish a pan-Scotland genomics research, development and innovation group to support delivery of our genomics ambition.	Initiated 2024-25
Consider short, medium, and long-term requirements for the delivery of pharmacogenomics using the Accelerated National Innovation Adoption pathway process and a pharmacogenomics working group.	Initiated 2024-25
Integrate with the developing NHS infrastructure for collaboration in Scotland to explore opportunities and mechanisms for triple helix partnership working between NHS, academia and industry.	Initiated 2024-25
Identify and evaluate current research, development and innovation opportunities, skills and strengths across Scotland.	Initiated 2024-25
Engage with researchers as service users to ensure that genomic data can be interpreted and informed by other diagnostic and clinical data.	Initiated 2024-25



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