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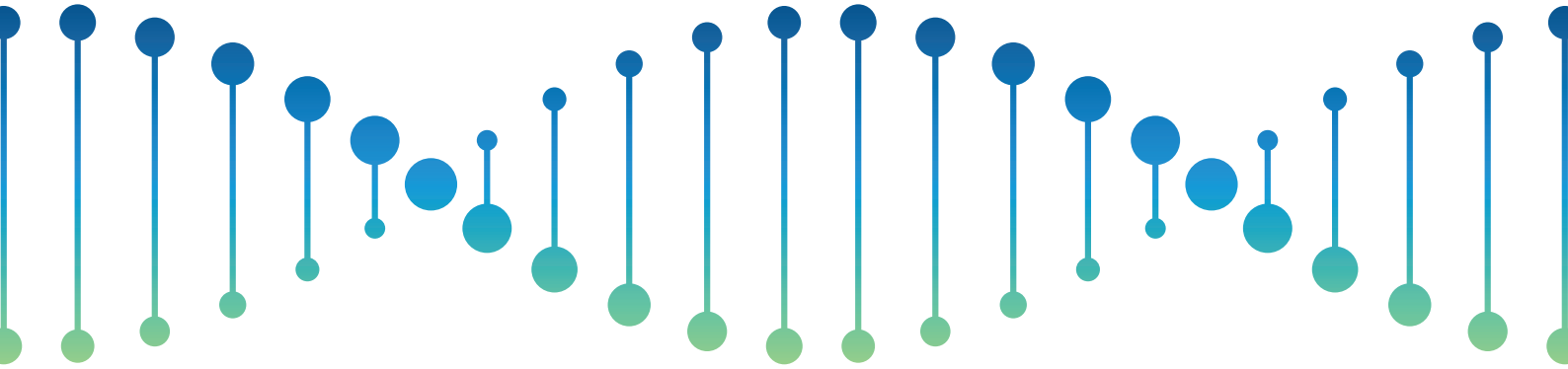
Genomics in Scotland: Building our future



Our strategic intent to deliver an equitable, person-centred, population-based genomics service and infrastructure for Scotland

March 2023

Scottish Government

Genomics in Scotland: Building our Future

Our strategic intent to deliver an equitable, person-centred, population-based genomics service and infrastructure for Scotland.

Ministerial Foreword

This publication is the first in a series, setting out our strategic vision for our genomics future. The Scottish Government is committed to the development of genomic medicine and the use of genetic information to improve the health and wellbeing of people living in Scotland. This was made clear in our commitment to 'Genome UK: The Future of Healthcare' back in September 2020.

Over the next 18 months we will publish a series of documents setting out how we will deliver genetic services which will not only allow us to treat disease but prevent ill health before the prevalence of symptoms. This is crucial to our future but also our immediate recovery priorities.

It is important to recognise that we start this journey with considerable expertise to draw upon. This was evident by our ability to stand up SARS-CoV2 pathogen sequencing services at short notice to support our response to the pandemic.

Outwith this service, Scottish physicians and scientists have been actively participating in the advancement of genomic medicine over the past three decades. We have a great foundation to build upon within our NHS laboratory network but it's important that these laboratories are fit for the future, as we know genomic medicine is on a steep trajectory.

To achieve our ambition of having a genomics ecosystem with the infrastructure to support it will require investment in rapidly evolving technologies, skills and facilities. We will need to be innovative and adaptive to change, working collaboratively through the triple helix approach with academia, industry and the public sector to harness the opportunities at our disposal and deliver an integrated approach to genomics across Scotland.

Humza Yousaf MSP
Cabinet Secretary for Health and Social Care

Chief Scientist for Health Foreword

I am pleased to welcome this strategic intent document that sets out the Scottish Government's path to support the development and adoption of genomic medicine in NHS Scotland.

Greater knowledge of the genome is providing opportunities for the design of new diagnostic approaches and the development and targeting of medicines to improve their effectiveness and/or reduce the potential for side effects. Genomic medicine is an integral part of precision medicine and precision health, where genomics and other health technologies have a potential to transform and improve healthcare. This can enable earlier and more precise diagnoses as well as more targeted prevention and management of diseases.

Research, innovation and collaboration between Scottish Universities and NHS Scotland together with life sciences industry partners will be vital to realising the benefits that medical genomics can offer to patients. We have particular strengths and a skilled and vibrant genomic and precision medicine research community in Scotland. It is crucial that we sustain and build upon our strengths, create and maintain supportive structures for research and innovation as well as strong and effective collaborations both within Scotland and beyond. A clear strategy for Scotland is an important first step.

Professor Dame Anna Dominiczak
Chief Scientist (Health) for the Scottish Government.

Contents

Introduction

Genomic medicine allows us to use genetic information to inform medical care or predict the risk of disease and has the potential to transform healthcare in Scotland.

We have seen a heightened awareness of genomics through the COVID-19 pandemic and, as we recover, we need to harness the potential of genomic medicine (both human and pathogen). By doing so, we can build robust infrastructure and systems that provide access to the best possible care for patients within our NHS, drawing on and nurturing scientific expertise and innovation. Having an open and meaningful dialogue with service users and the wider public is essential if we are to do this.

Our Genomics Vision

- Improve the lives of people in Scotland, the quality of healthcare services and public health surveillance.
- Have the infrastructure, mechanisms and workforce required to capitalise on advances in genomic medicine.
- Make best use of scientific and clinical expertise in both human and pathogen genomics to translate research and innovation into clinical practice.
- Increase awareness and the benefits of genomics across Scotland so people can make informed decisions about their treatment and health.
- To work closely with our partners in Scotland and across the UK to ensure equity of care for patients and share best practice where possible in accordance with the Genome UK shared commitments[i].
- Motivate and accelerate Scotland's thriving Life Sciences and Precision Medicine Sector and stimulate economic growth through the development and application of genomic technologies.

Scottish Strategic Network for Genomic Medicine

NHS Scotland's genetic and molecular pathology laboratories are commissioned through National Services Division (NSD), part of NHS National Services Scotland. Services are delivered through four regional laboratories based in Aberdeen, Dundee, Edinburgh and Glasgow. These genetics laboratories previously collaborated through the Scottish Genetics Laboratory Consortium (SGLC) which has now been replaced by the Scottish Strategic Network for Genomic Medicine (SSNGM).

Established in August 2022, the aim of the SSNGM is to provide overall strategic leadership and oversight for genomics in Scotland. The Network structure allows us to draw on the body of expertise across Scotland in genomics to support the development and implementation of our Strategy (see Appendix 1: SSNGM structure).

A key area of priority for the SSNGM is implementing the recommendations from the NSD *Major Review Report of Genetics & Molecular Pathology Laboratory Service*[ii] completed in March 2022. With funding from the Scottish Government, NSD has put in place a Genomics Transformation Team which is leading on the development and implementation of an action plan to transform our genomic laboratory services so they are fit for our future strategy.

Investing in our future

The recovery from COVID has coincided with a cost of living crisis and the war in Ukraine, all of which has contributed to an incredibly challenging public finance landscape.

We accept this is a challenge for the genomics community. Spending on genomics is often new and innovative with significant spending in the short term, for example, on sophisticated genetic testing. This testing can, however, lead to greater savings in the longer term through the earlier and/or more effective treatment of different conditions.

Measuring the health economics of genomic medicine continues to be an important part of our approach, providing an evidence base for the prevention of ill health as well as treatment for already established conditions in patients. As new testing and innovations become available we will flex and adapt our services to ensure we are providing the most efficient and value for money service for the NHS in Scotland.

Scotland's Genomics Strategy

Genomics is a fast-moving and rapidly evolving field. This document is the first of three publications, and it sets out the intended direction of travel for genomic medicine in Scotland.

The second publication will be a five-year genomics Strategy for Scotland, developed in partnership with the SSNGM. The five-year timescale allows us to focus our work within the wider recovery agenda in Scotland and provides flexibility to adapt our approach as the field develops and new technologies emerge.

Finally, we will also publish an implementation plan for the Strategy recognising the need for rapid and accelerated implementation in several areas particularly for those living with cancer and rare diseases.

Over the next five years, the genomics landscape in Scotland will be transformed as we work to ensure that patients have equitable access to timely, personalised and high-quality genomic medicine for a range of conditions.

Diagnosis and Precision Medicine

Genome UK Vision: we will help people live longer, healthier lives by using new genomic technologies to routinely identify the genetic determinants of rare diseases, infectious diseases and cancer. We will detect cancers earlier, and we will provide personalised treatments to illness.

Using genomic technologies, we can predict ill health and help to provide better diagnosis and treatment through the use of precision medicine to support people to live longer and healthier lives. The genomic medicine landscape is developing rapidly. In the last five years, we have seen the implementation of impactful genetic testing for a variety of conditions which has helped inform patient treatment, allowing patients to receive the right treatment at the right time. Pathogen genomics is also helping to deliver 'precision public health' by guiding investigations of infection outbreaks, antimicrobial stewardship, better-targeted disease control and infection surveillance.

In December 2022, we published Scotland's Rare Disease Action Plan[iii], aligned to both the UK Rare Disease Framework[iv] and Genome UK. Around 80% of rare diseases are currently understood to have a genetic origin, and genomic healthcare provides a significant opportunity to shorten the 'diagnostic odyssey' for individuals living with these conditions. Our Genomic Strategy will align to our Rare Disease Action Plan and we will engage with the Rare Disease Implementation Board as well as key rare disease organisations like Genetic Alliance UK to ensure our Strategy is informed by those who will benefit most.

We expect that the next five years will see considerable growth in cancer genomics, at a time when cancer care in Scotland is continuing to recover from the impact of Covid-19. Our cancer genomics expansion will support the testing required to help patients benefit from targeted treatments and ensure that patients do not receive treatments where there are no clinical benefits and avoid unnecessary side effects. By building genomics into the cancer pathway, we hope to be able to detect relapses earlier and more effectively, and consider the screening implications for patients and their families.

Pharmacogenomics (the study of genetics in relation to medicine, and the influence of genes on an individual's response to medicines) is also an enabler for precision medicine. The introduction of pharmacogenomics into routine practice has the potential to transform the use of medicines with pharmacy services and we are already looking to the future and building on a growing evidence base through pharmacogenomic research programmes at the Universities of Glasgow and Dundee.

To support our approach to diagnosis and precision medicine, we will need to;

- Support the development of, and ensure alignment between Scotland's Genomics Strategy, Rare Disease Action Plan and the new Cancer Strategy.

- Expand our genomic cancer tests to ensure clinicians have timely access to better diagnostic, prognostic and potential therapeutic information for their cancer patients, resulting in better outcomes.
- Seek to develop capabilities to enable greater use of genomic testing results to support opportunities for patients to enter genome-directed clinical trials of treatments for both cancer and rare diseases, recognising the benefits that this research can bring to individual patients and the NHS.
- Work with the Rare Disease Implementation Board for Scotland to improve diagnostic pathways for those living with rare conditions.
- Consider investment in infrastructure and leadership (as part of the wider Precision Medicine portfolio) for pharmacogenomics, pharmacy services and for the wider multidisciplinary team, as laboratory and digital capabilities are crucial in the provision of services.
- Better understand the future opportunities for Advanced Therapeutic Medicinal Products (ATMPs), recognising that there are wide-ranging implications and challenges for their managed introduction.
- Work with the SSNGM to promote a harmonised and multidisciplinary approach to the roll out of pharmacogenomics across Scotland.
- Consider the requirements for future services, and if necessary, the scope for increasing capacity for Whole Exome Sequencing (WES) and/or Whole Genome Sequencing (WGS).

Prevention

Genome UK vision: We will use genomics to accurately predict the risk of chronic diseases and our national screening programmes will use genomics to identify at-risk populations.

Genomic medicine has great potential not only in the treatment of an individual who has been diagnosed with a condition, but also identifying healthy individuals at higher risk of developing a condition and taking early and evasive action. It can help us to understand more complex, multiple gene interactions, as well as the interactions between genes and the environment and can help develop new methods for diagnosis, treatment and sometimes even prevention of conditions.

This is primarily done through screening. While not all screening will take the form of a national screening programme, we will collaborate with the Scottish Screening Committee which is aligned to the UK National Screening Committee.

In January 2023, the First Minister set out the pressures facing the NHS in Scotland and the need to relieve the burden on our hospitals. To do this we must take more of a proactive and preventative approach to health. Genomic medicine has a key role to play in managing the pressures faced by our NHS. It allows health conditions to be predicted before they become prevalent through the use of testing for predisposition to certain conditions to then allow us to prevent ill health before requiring Primary Care input or hospitalisation. Genomics also allows us to apply precision medicine to patients who are already ill in order to inhibit further side effects from medications or, depending on a person's genetic makeup, utilise precision medications for maximum effect.

To support our approach to prevention our Strategy will cover how we:

- Enable NHS Scotland to move from diagnosing and treating conditions once symptoms are prevalent to utilising genomics to predict and prevent ill health.
- Work with the Scottish Screening Committee and National Screening Oversight to understand how we can best link with national screening programmes that use genomics in order to provide interventions to those with increased risks.
- Support the development of pathways to identify those at a higher risk of disease ensuring follow up and treatment pathways are in place.
- Work closely with the Pathogen Genomics Oversight Group (PaGOG) and the pathogen genomics community to expand our understanding of pathogen genomics, especially in relation to Infection Prevention and Control (IPC) and antimicrobial resistance (AMR)
- Maintain engagement with Genomics England Ltd on the roll out of their Newborn Screening Research Project and consider opportunities for interaction and shared learning which can be applied in Scotland.
- Consider the expansion of Scotland's existing Non-Invasive Prenatal Testing (NIPT) and Preimplantation Genetic Testing (PGT) programmes for inherited conditions to take account of new and advancing technologies.

Research and Innovation

Genome UK Vision: We will extend the UK's world-leading position at the forefront of discovery-led and translational genomics research, continually expanding our collective genomics knowledge base. We will develop an ecosystem of world-leading secure genomics datasets, powering international research and supporting a seamless transition of impactful research findings into the healthcare setting backed up by robust implementation research programmes.

The development of genomic medicine in Scotland has benefited greatly from leading academic expertise in, and partnership working between, Scottish Universities and the NHS as exemplified by the Scottish Genomes Partnership. We want to continue to support collaborative research and innovation activities between the Scottish academic, NHS and life science sectors through continued investment by the Chief Scientist's Office (CSO) research funding committees[v] and fellowship schemes[vi]. In addition, we are continuing to work with Our Future Health[vii] to support Scottish participation in this ground-breaking research programme.

It is vital that the innovation and multidisciplinary networks fostered under the Scottish Genome Partnership[viii], and broadened during the Covid-19 pandemic, be built upon and strengthened in order to translate new developments into tangible benefits for Scotland as part of the wider recovery agenda.

Case Study: Response-mode funded research project - Liquid Biopsy

A team at the University of Edinburgh's Institute of Genetics and Cancer have established a range of blood-based tests, known as liquid biopsy, which detect and monitor cancer DNA fragments shed by tumours into the blood stream in Oropharyngeal squamous cell carcinoma (OPSCC) patients.

Having recently established tests for which a positive result gives a 10-fold increased risk of tumour recurrence in Human Papillomavirus (HPV) associated tumours, this Chief Scientist Office (CSO) funded project now looks at the OPSCC group with the poorest outcomes (non-HPV tumours). The project will apply state-of-the-art DNA detection and sequencing technologies to analyse tumour-derived DNA fragments in the bloodstream, to follow treatment response and to develop new methods for detecting relapse and resistance to treatment in non-HPV OPSCC.

The tests for HPV-associated tumours are now being taken into the national HPV reference laboratory and the ultimate aim is that tests for both HPV-associated and non-HPV tumours can be translated into standard of care to improve the clinical assessment, and quality of life and overall outcomes for OPSCC patients in Scotland.

While we can take confidence from the progress to date, we also recognise the need for a transformation of the infrastructure and mechanisms to advance genomic research in Scotland, with a focus on the following areas:

- Storage, use and sharing of genetic data guided by the development of the NHS Scotland National Digital Platform.
- Development of a meaningful dialogue with stakeholders including patient groups, service users and the wider public about the wider implications of human and pathogen genomics.
- Working with industry to understand their research requirements, and how we can collaborate to mutual benefit.
- Improving the national research governance infrastructure to remove unnecessary barriers and allow an improved one nation approach to approving and conducting studies in rare disease and cancer.
- Development and promotion of innovation and a culture of translational research and knowledge exchange as an integral part of the genomic and bioinformatics workforce in Scotland.

Cross Cutting Themes

Patient and Public Engagement

Genome UK Vision: We will build and maintain trust in genomic healthcare with patients, the public and NHS workforce, ensuring that they are involved and engaged in how we design and implement genomic healthcare, including the use of data and ethical considerations.

Our biggest priority in all this work is patients. As part of our person-centred approach, we will partner with patients, families, charities and other representative groups to establish a patient panel as part of our Network to ensure that patient needs inform the development, monitoring and delivery of services. Increasing awareness and engagement in this way will allow patients to make informed choices about the services they use and provide reassurance to the wider public.

Public Health and Pathogen Genomics

The formation of the SSNGM and the work to deliver a genomics strategy for Scotland is a chance to assess the continuing development of both human and pathogen genomics and how these areas can complement each other to provide maximum benefits for patients and wider public health, potentially as part of a wider 'One Health' approach. Public Health Scotland and collaborative partners across Scotland are already demonstrating the value of genomics in managing individual cases of disease and wider surveillance activity (see case study below). We also want to build on the successful multidisciplinary partnerships built around sequencing for Covid-19 to ensure that we sustain and nurture this expertise.

Case Study: Speeding up diagnosis and treatment management decisions for Tuberculosis (TB) using whole genome sequencing

Since February 2022, Scotland's Mycobacteria Reference Laboratory in Edinburgh has used Whole Genome Sequencing (WGS) to sequence M. tuberculosis, and other Mycobacterium species that cause disease.

Using this single test, they have improved the turnaround time to a matter of days whereas this was previously 4 – 6 weeks for both information on diagnosis and antibiotics susceptibility. This helps to ensure that the right antibiotics are prescribed more quickly thus allowing patients to recover faster and, by reducing the number of ineffective courses of antibiotics, limiting the opportunities for antibiotic resistance to develop. The information from WGS also provides high-resolution genotyping that can identify closely related strains that potentially come from the same source.

This level of detail that WGS provides goes beyond what was previously available to trace the transmission of TB. It is being used by public health teams to track the transmission of TB, the spread of drug-resistant strains and to guide the investigation of outbreaks.

Workforce

Genome UK Vision: We will support the NHS workforce, academia and industry workforce to develop and acquire the necessary scientific and clinical skill sets and understanding of genomics, including bioinformatics. We will support the workforce across all sectors to communicate about genomics in an accessible way. We will prioritise workforce and training in spending and policy considerations. We will implement a framework of skills across the sectors, identifying the major skills shortages in each and propose new ways of training to keep up with demand. We will develop clinical pathways and standards of care that fully incorporate the latest genomic testing and results.

The experience of Covid-19 has emphasised the importance of the NHS and the people who work within it, including those working in laboratories and other clinical support roles the length and breadth of the country.

We will work to support the necessary planning, training and development of both new and existing NHS staff to allow them to deliver a service that keeps pace with developments in the field, recognising that improved testing and patient pathways will impact upon the requirements for clinical staff. In particular, we will consider how we embed a bioinformatics workforce within our infrastructures.

We will also look to plan and encourage our future workforce, working with Universities, Colleges, schools and others to encourage uptake of Science Technology Engineering and Maths (STEM) subjects and position a career in healthcare science, including genomics, as an attractive ambition for young talent.

Data

Genome UK Vision: We will standardise the way we record and store genomic, phenotypic and healthcare data to support the use of advanced analytics in both care and research and to ensure that a patient's genomic data can repeatedly inform their care throughout their life.

Through our Research and Innovation pillar we are already committed to developing a Scottish genomic data repository that will join up the four Scottish NHS genetics laboratories, encourage greater connectivity and allow secure storage and sharing of genetic data to support diagnosis and care of patients.

Access to genomic testing, and the ability to safely and securely share and interpret data can also open up opportunities for patient treatment, including through clinical trials. We intend for Scottish genomic data to be used to support patient access to clinical trials across cancer and rare diseases. We will consider how we can develop the necessary infrastructure, processes, and skills to enable the use of genomic data in alignment with Scotland's first Data Strategy for Health and Social Care[ix].

Industrial Growth

Genome UK Vision: We will make the UK the best location globally to start and scale new genomics healthcare companies and innovations, attracting direct investment in genomics by the global life sciences industry and increasing our share of clinical trials in the UK.

Scotland has a thriving life sciences economy, which employs 41,700 people across over 700 organisations. We want Scotland to be internationally recognised as a world leader in genomics, attracting researchers, small and mid-size enterprises, and industry to our country.

Scotland has gained momentum in pharmacogenomics in the last few years led by the Scottish Pharmacogenomic Working Group and drawing upon expertise from across Scotland's industry, academia, and government.

Scottish Enterprise will work closely with industry to align with the ambitions of the Life Sciences Strategy for Scotland and will engage with the Life Sciences Scotland Industry Leadership Group, the Scottish Health Industry Partnership (SHIP) and other industry groups to align the work of our network with our Innovation Pipeline and Accelerated National Innovation Adoption (ANIA) Pathway to encourage growth and drive innovation.

Ethics

Genome UK Vision: All our genomic data systems will continue to apply consistent high standards around data security and the UK model will be recognised as being the gold standard for how to apply strong and consistent ethical and regulatory standards that support rapid healthcare innovation, adhere to legal frameworks, and maintain public and professional trust.

The handling of an individual's personal data, in this case genomic data, raises a number of ethical considerations, such as how to store data securely over time, how and when it would be appropriate to share such data, and with whom. Early engagement on the wider ethical issues associated with genomics will form a core part of our strategy, with efforts to engage with patient groups, the wider Scottish public and centres of expertise in bioethics and data governance.

We are also aware that the majority of current genetic datasets are skewed to reflect people with predominately white western European ancestry. We intend to work with others to increase representation of hitherto under-represented groups with a view to building genomic datasets that are reflective of the population we serve.

Our patient panel and wider public engagement and participation efforts will be critical to making advances in this area whilst ensuring that our regulatory and ethical frameworks support rapid healthcare innovation and are both understood and accepted.

Environmental impact

The Scottish Government recognises environmental impact as a priority across all of our policies. The evidence of a global climate emergency is irrefutable, and to tackle this emergency we need the public and private sector to take action to slow and adapt to the effects of climate change as set out in our updated Climate Change Plan and Scottish Climate Change Adaptation Programme (SCCAP).

Scotland has made great progress on our journey to net zero and in August 2022 we published the NHS Scotland climate emergency and sustainability strategy: 2022-2026[x], but there is more to do within the genomic medicine service to reduce harmful environmental impacts. We will learn from examples (see case study below) to ensure the transformation of our genomics infrastructure supports our climate change agenda.

Case Study: Q^2 Solutions MyGreenLab certification

Q^2 Solutions, a wholly owned subsidiary and laboratory division of IQVIA, has recognised the impact that laboratories have on the environment and have set out several commitments publicly, as part of their Environment, Social and Governance (ESG) programme, to reduce the impact on the communities they operate in globally. This includes a commitment to become a net-zero business via the Science Based Target initiative (SBTi), with the target to be published by end of 2023. This will align Q^2 Solutions business operations with the Paris Agreement from COP 21.

In 2021, Q^2 Solutions enrolled their Livingston laboratory in Scotland on to the My Green Lab Certification programme. This certification programme is recognised by the United Nations Race to Zero campaign as a key measure of progress towards a zero-carbon future and is considered the international gold standard for laboratory sustainability best practices. To successfully gain certification the requirement is to reduce a laboratory's environmental impacts across a variety of categories including energy, water, and waste.

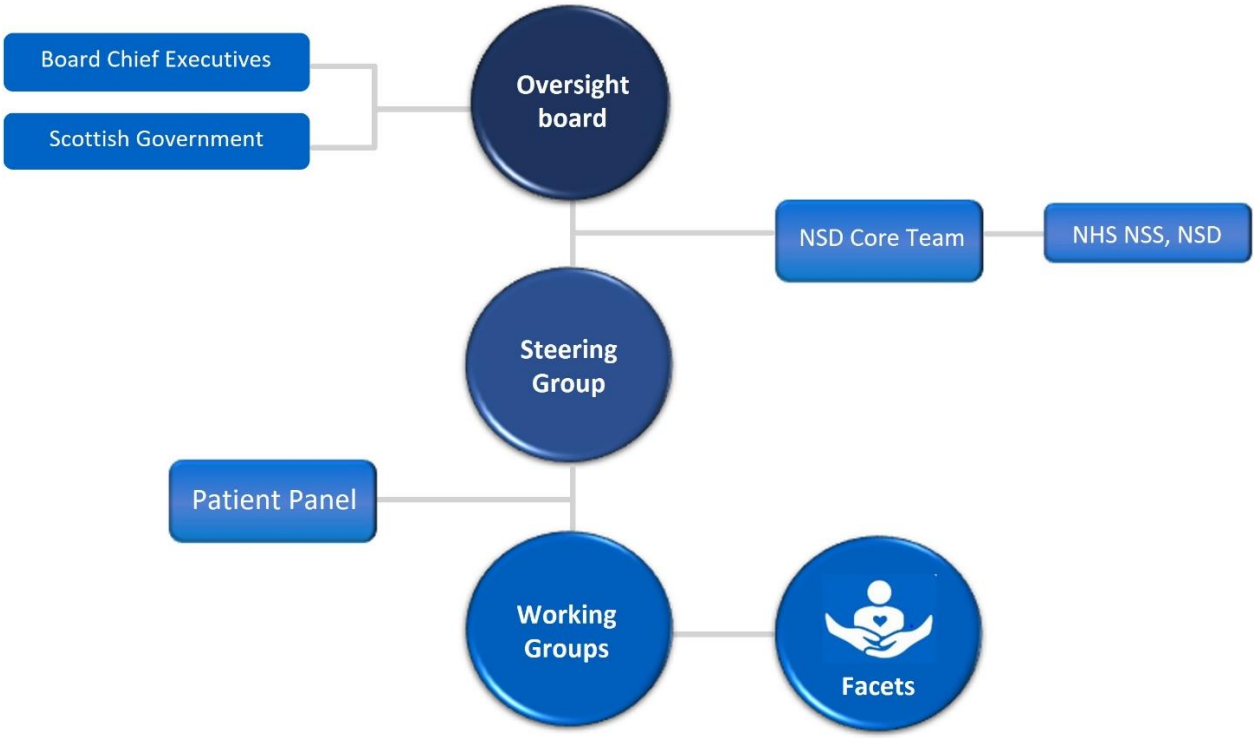
Some of the initiatives Q^2 Solutions have implemented since this programme was launched include energy saving programmes around laboratory equipment and -80 freezers, a new glass recycling programme to divert glass waste away from incineration and work with key suppliers to reduce packaging waste.

As it stands today, Q^2 Solutions has 71 My Green Lab Ambassadors trained on the Livingston site as part of a laboratory operations teams whose role it is to initiate projects to drive sustainability improvements. The Livingston laboratory achieved certification in November 2022 with all other Q^2 Solutions laboratories globally aiming for certification across 2023-2024.

Q^2 Solutions are proud of these achievements to further increase sustainability in their laboratory environments and are committed to embedding sustainable practices across business operations globally to continue safeguarding the planet.

Appendix 1 - Scottish Strategic Network for Genomics Medicine Structure

Below is a diagram that shows the hierarchy and reporting structure for the Scottish Strategic Network for Genomic Medicine, with the Oversight board at the top reporting to NHS Board Chief Executives and the Scottish Government. In addition to the Oversight Board, the Steering Group, patient panel (in development), working groups and facets which exist within the network are all represented, as is the Networks Core Team which is drawn from NHS Scotland National Services Division and the Scottish Government Genomics Policy team.



References

[i] You can access the Genome UK: shared commitments on the UK Government website
[ii] Read the report following the Major Review of Scotland's Genetic and Molecular Pathology Laboratory Services on the NHS National Services Scotland website
[iii] Access Scotland's Rare disease action plan on the Scottish Government website
[iv] View the UK Rare Diseases Framework on the UK Government website
[v] Read more information on the Chief Scientist's Office research funding committees on the website of the Chief Scientist Office
[vi] Read more information on fellowship funding schemes on the website of the Chief Scientist Office
[vii] Read more information our future health on the Our Future Health Website
[viii] Read more information on the Scottish Genome Partnership on the Scottish Genome Partnership website.
[ix] You can view the Health and social care data strategy on the Scottish Government website
[x] Access the NHS Scotland climate emergency and sustainability strategy: 2022-2026 on the Scottish Government website

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