

Informing the Future of Genomic Medicine in Scotland



Summary report

Introduction

- Genome-based technology is transforming healthcare. Medical programmes are underway in several countries, with more than 20 having programmes to sequence the genomes of at least 100,000 subjects. The technology is revealing new diagnoses for patients with rare inherited diseases and a genomic approach is enabling more targeted cancer treatments with better prospect of successful outcomes. Genomic Medicine also provides significant research and commercial opportunities.
- The SSAC received a request from the Scottish Government's Health & Social Care directorate to review the development of Genomic Medicine in Scotland, as an integral part of Precision Medicine, in order to support long-term planning and investment in this fast-evolving area of healthcare.
- This report provides an overview of current Genomic Medicine capabilities in Scotland, compares developments elsewhere in the UK and internationally, and summarises opportunities and benefits for the NHS, research and life sciences sectors in Scotland.
- To produce the report, the SSAC brought together a group of experts from academia, the NHS, and life science sectors, to advise where action and investment are required. The SSAC thanks this group for their expert knowledge, energy and commitment.
- This summary document provides a short overview of the main themes and actions from the full report, which can be found at <http://www.scottishscience.org.uk/>

What is genomics?

Genomics is the study of the complete genetic material of an individual. It can be thought of as an individual's instruction book of life, determining everything from height and eye colour to our risk of developing rare inherited conditions such as cystic fibrosis and haemophilia, and common illnesses such as cancer and heart disease. Genomics is a vital part of Precision Medicine, which is about providing the right treatment at the right time by tailoring treatment to an individual's molecular and genetic characteristics.

Genomic Medicine is the use of genome technology and information in healthcare, and in related research and innovation. It seeks to understand the functions of genes, how genetic variations cause disease and to provide new, precise methods for diagnosis and treatment.

Genomics in Scotland

Scottish physicians and scientists have made world-leading research contributions in rare disease and cancer genomics in the past three decades. There is also a well-established network of clinicians and clinical scientists working in NHS Scotland who have been delivering Genomic Medicine since at least 2013.

In 2015/16, the Scottish Genomes Partnership was established, from a £15 million investment by the Universities of Edinburgh and Glasgow and a £6 million joint investment by the Scottish Government and UK Medical Research Council. This generated sequencing infrastructure for next-generation Genomic Medicine, building on existing expertise and recruiting new talent. The Scottish sequencing centres have sequenced more than 12,000 whole genomes for a range of projects, including research studies. A major success of the partnership has been the strong collaborations built between academia, NHS, industry and other stakeholders, placing Scotland amongst those at the forefront of Genomic Medicine internationally.

A primary aim of the Scottish Genomes Partnership is the improvement of targeted therapies, focusing predominantly, at the present time, on rare diseases and cancer.

Genome technology has transformed in the last 20 years



A reduction in costs, in genome sequencing and data storage, alongside faster and more accurate data production is revolutionising healthcare.



The aim of a single genomic test is to provide a 'one-stop-shop' that avoids the need for other diagnostic tests, saving money through more rapid diagnosis and improved health.

Rare disease diagnostics

A rare disease is defined as a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000. One in seventeen people in Scotland will be affected by a rare disease at some point in their lives.

Scotland has played a leading role in research to diagnose rare diseases, such as the pioneering UK10K, Deciphering Developmental Disorders (DDD) study and the 100,000 Genomes Project. DDD identified more than 30 new genes for developmental disorders and has helped more than 13,000 families with these conditions.

In rare diseases caused by a glitch in a single gene, genome sequencing can pinpoint the exact molecular change underlying the disease.

In common diseases usually caused by an interplay of genetic variation and the environment, genome sequencing can identify instances where the disease is caused by an abnormality in a single gene.

Genomic Sequencing can offer precise, quick diagnosis leading to:

- a better patient experience,
- improved care for patients and their families,
- fewer prolonged, invasive and often expensive clinical investigations,
- accurate reproductive counselling and pre-symptomatic diagnosis to reduce the risk of familial disease recurrence, and
- potential cost efficiencies.

Case Study

A child born with paralysed malformed legs and severe developmental delay was first seen in the genetic clinic at the age of two years.

Over the following six years the child and family underwent a large number of investigations to identify the underlying cause of the problems.

These included MRI scans and electrophysiological testing of muscle and nerves. These required general anaesthetics as well as many targeted and genetic blood tests.

With all standard avenues exhausted and no explanation, the child's parents decided to enrol them in the DDD research study for trio-based whole exome analysis. This identified a mutation in a gene not present in either parent which indicated a very rare genetic condition.

The diagnosis from this research study gave the family an answer to the cause of the child's problems, reassured them that they, and other family members were unlikely to have another child with the same condition, and allowed them and their doctors to learn from children worldwide with similar problems in terms of clinical outlook and appropriate care.

Transforming models of cancer care

Genome sequencing is already having a major impact on cancer diagnosis and management worldwide, and is playing an increasingly central role in prevention and treatment.

Cancer develops through changes to genomes in our body tissues and these changes determine how a cancer grows and behaves, including its response to treatment.

Using genomic technologies we can predict which patients will benefit from a particular treatment, improve outcomes, minimise use of ineffective treatments and reduce unnecessary complications.

Increasing numbers of targeted therapies for cancer are also challenging traditional models of care. A more personalised model of cancer care is now needed in NHS Scotland to help doctors tailor treatments to specific genomic markers.

We are ideally positioned to offer world-class genomic profiling to all Scottish cancer patients through a combination of local sequencing capacity, expertise in clinical trials and Precision Medicine, engaged and committed NHS laboratories, and access to a wealth of treatment and outcome data.

Case Study

KYT is a US program for pancreatic cancer patients. Pancreatic cancer has a median survival of six months and only 10% of patients survive for a year after diagnosis.

In KYT, patients have their tumour profiled using genomic and other molecular technologies.

Half of patients profiled to date have had an 'actionable change' identified in their cancer, meaning that a specific targeted treatment is available.

Patients who had a treatment that was matched to their actionable change survived more than twice as long as patients who had unmatched treatment.



40%

40% of Scotland's population will be diagnosed with cancer at some point in their lives



25-30%



Cancer is the registered cause of death for 25-30% of people in Scotland



There are over **30,000 new cases** of cancer diagnoses each year in Scotland.

+30,000

Opportunities provided by digital health

Scotland has recognised the enormous opportunities for better use of data to improve health care but its enviable electronic health record system could be improved significantly through the addition of information about a patient's genome.

Scotland's 2018 Digital Health and Care Strategy commits to using technology to reshape and improve health and care services. Genomic data will be an important aspect of the proposed national digital platform to record and deliver real-time health data at point-of-care.

A key goal for the NHS should be to develop a 'Self-learning Health System': robust data acquired through routine care and use of innovative medicines that inform future treatment decisions as well as resource provision.

Scotland's state-of-the-art genome sequencing centres are creating additional opportunities to engage with commercial companies, including the clinical trials market. An area of specific interest for Precision Medicine is the integration of data from diverse sources to improve clinical decision-making and attract industry projects. New commercial opportunities for big data projects and clinical interpretation have already arisen from Genomic and Precision Medicine research in Scotland.



Due to the modest size of Scotland, it should be possible to combine routine healthcare and genomic information to obtain relatively complete data for the entire Scottish population. This could underpin a data-driven approach to public health and disease prevention that is unlikely to have been attempted at a national scale elsewhere. Safe and effective governance of data will be critical to ensure public confidence.

Key recommendations

Leadership

The Scottish Government **should convene** a Scotland-wide Leadership Group to advise how best to support the development of Genomic Medicine in Scotland; challenge barriers to progress; maximise impact for the benefit of healthcare, research and life sciences in Scotland; engage and involve the public; and increase Scotland's influence at UK and international levels, positioning Scotland as a world leader in genomics in the wider context of Precision Medicine.

Clinical Implementation

With Scottish Government support, NHS Scotland **should expedite** the evaluation and adoption of genomic testing strategies into clinical pathways where there is good evidence that these lead to improved patient outcomes.

Workforce

NHS Scotland working with NHS Education for Scotland, ScotGen, Skills Development and academic institutions **should lead** in co-ordinating the development and delivery of the training courses and educational resources required to develop essential expertise required to support and drive world-class Genomic Medicine capabilities in NHS Scotland.

Digital Health

The Scottish Government **should take full account** of the digital infrastructure needed to enable genomics within clinical pathways and to support the use of genomic data for research and innovation. This infrastructure includes: digital skills, supercomputing hardware, high capacity connected networks, and systems for data management and security.

Research and Innovation

The Scottish Government, working with research funders, industry, enterprise agencies and higher education providers, **should consider** how best to support genomic research and innovation in Scotland, including patient and public involvement and engagement, and maximise the opportunities for inward investment, building on Scotland's current position of excellence.

Industry-Facing Activity

The Scottish Government, working with enterprise agencies, **should build** on the strengths of Scotland's 'triple helix partnership' between academia, the NHS and industry and consider how best to position as assets across Scotland to maximise future engagement and partnership opportunities which will accelerate genomics development as an integral part of Precision Medicine.



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