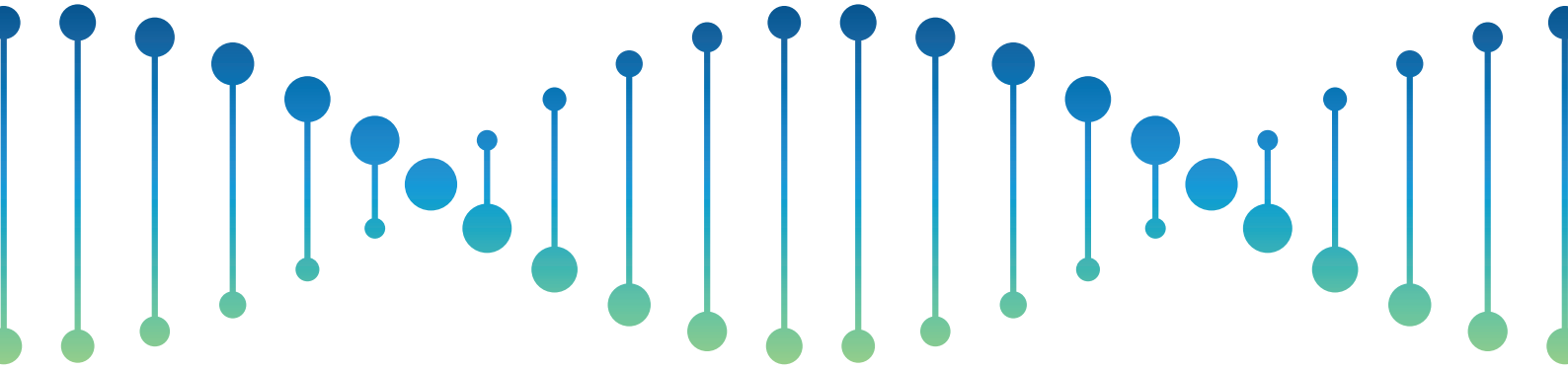
Genomics in Scotland: Building our Future



Summary

A five-year national strategy

April 2024

Scottish Government
Riaghaltas na h-Alba

1. Sources of support

Some of the content in this Strategy and the associated documents may have an emotional impact on you. Support is always available, and some of the national sources of help are listed here.

Genetic Alliance UK

Genetic Alliance UK is an alliance of over 200 organisations supporting people with genetic, rare and undiagnosed conditions in the UK. You can access information on different genetic conditions, genetic testing and search for support groups via their website: https://geneticalliance.org.uk/ or email: contactus@geneticalliance.org.uk

Macmillan Cancer Support

Macmillan support people living with and affected by cancer. They provide a wealth of resources about different types of cancer, testing and treatments as well as information on how to get help coping with the physical, emotional and financial impact of cancer. You can access information and links via their website: https://www.macmillan.org.uk/ and the Macmillan support line 0808 808 00 00

NHS Inform

NHS Inform is Scotland's national health information service to help provide people in Scotland with accurate information and resources to help people make informed decisions about their own health and the health of people they care for. You can access information via their website: https://www.nhsinform.scot/ or their telephone helpline 0800 22 44 88

Mind to Mind

If you're feeling anxious, stressed or low, or having problems sleeping or dealing with grief, find out how you can improve your mental wellbeing by hearing what others have found helpful by visiting: https://www.nhsinform.scot/mind-to-mind

2. Contents

3. What is genomic medicine?

Genomic medicine is the use of genetic information (the instructions within our body's 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.

4. How do we use genomics in medicine?

There are many different types of tests available to look for changes in individual genes, chromosomes, or proteins within the body.

These can be used for:

- **Diagnosis,**
- As part of **screening**, for **monitoring** of an existing condition,
- To make decisions about **treatment**.

Healthcare professionals will consider different factors when choosing the right test for each person including what condition, or conditions, is suspected and the genetic changes usually associated with those conditions. Tests can focus on small segments of a person's DNA (single gene or small numbers of genes) or on large numbers of genes or whole sections of a person's genome (all their genes together).

Some of the genetic changes that healthcare professionals look for are inherited (they are passed from parents to children).

There are also genetic variants that are not inherited but develop during a person's lifetime or in response to different environmental factors.

Most cancers, for example, are linked to non-inherited genetic changes but healthcare professionals may still in these cases look at inherited genetic changes because it can help guide treatment decisions (some people will respond better to some treatments than others because of their genetic make-up).

Genomic medicine has a very wide range of potential uses. This strategy is focused on its application to cancer and rare and inherited conditions, as the areas that need attention most urgently in Scotland.

5. What is a strategy and why do we need one?

A strategy is an overall plan to set out our direction of travel and the key things we want to achieve.

This strategy is our plan to build a better genomic medicine service in Scotland.

It is accompanied by a list of actions setting out what we will do to work towards this plan in the first year. This will be followed up by further plans during the strategy term.

6. What do we want to achieve through this work?

Our vision is to establish a genomic medicine service that improves the lives of people in Scotland in order to:

- Provide the genomic tests needed to support the best possible medical care for people in Scotland.
- Support people and their families before, during and after genomic testing.
- Ensure we have enough people across the NHS with the skills and knowledge to carry out genomic testing and to understand and interpret test information.
- Build safe and secure data systems able to cope with the large amounts of complex information generated by genomic testing.
- Talk with people across Scotland about genomic medicine, what it means for them and what they may be worried or excited about.
- Support research, development, and innovation in genomic medicine to keep learning and make sure that new knowledge can benefit people in Scotland.

7. Where will we focus?

To achieve our goals we need to expand access to genomic testing and, alongside this, build stronger foundations (our 'building blocks') for genomic medicine in Scotland.

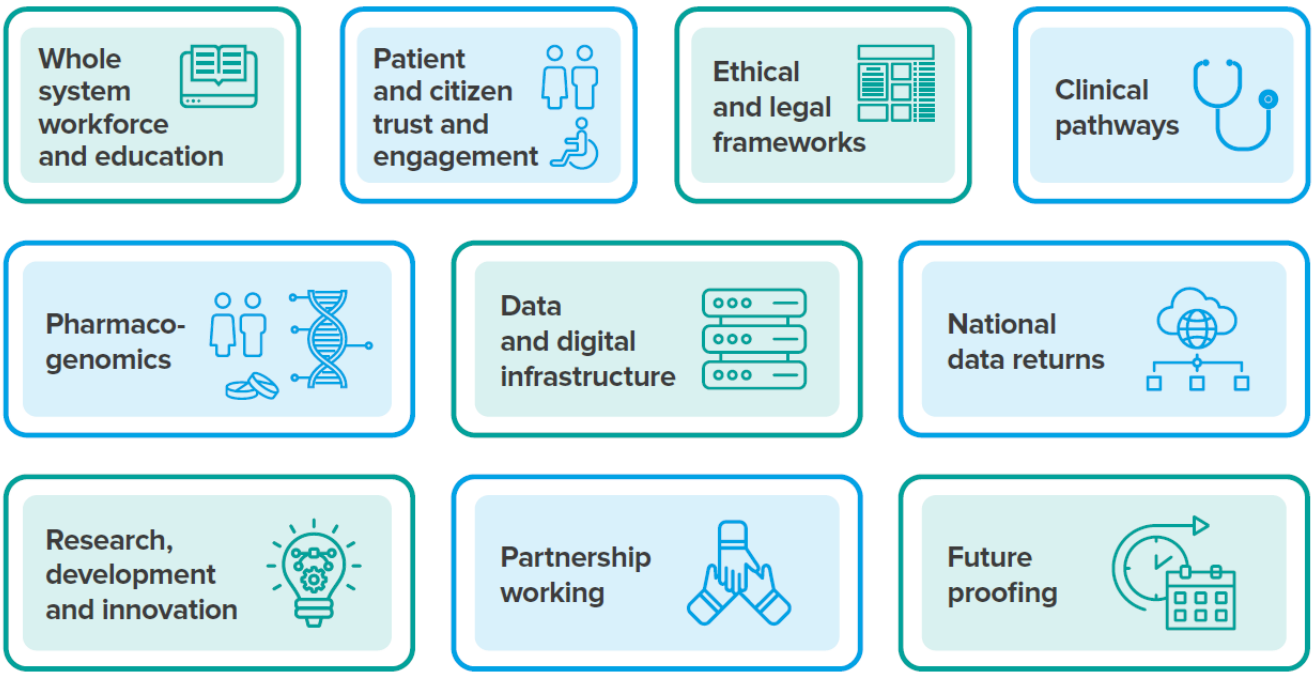


Figure 1. Our building blocks to develop our genomic medicine service in Scotland.

7.1 Prompt and equal access to genomic testing

We want to make sure that genomic medicine in Scotland supports diagnosis and access to the right care, at the right time, for the right person. Timely access to the right test will shorten the time people have to wait for their genomic results. This can lead to a treatment plan starting sooner and being able to improve people's lives from quicker intervention.

7.2 Whole system workforce & education

Our workforce, those who staff NHS Scotland with their care, experience and knowledge, are our greatest asset. We need to support our workforce through training, clear career pathways and opportunities for development within the fast-moving discipline of genomic medicine.

7.3 Patient and citizen trust and engagement

As genomic medicine develops, we need to engage with people using services to take on board their lived experience and what matters most to them. Promoting person-centred care and resources will also help people know what options are available, what questions to ask and how to get the most out of services that are available.

7.4 Ethical and legal considerations

The safe and appropriate management and use of genomic information needs careful consideration so that people in Scotland can be confident that their information is being used securely and in ways that they have agreed with.

7.5 Clinical pathways

A clinical pathway is a checklist that includes all the steps that should be taken in a person's care, by all the various healthcare professionals involved, to make sure care and treatment are given properly. We will work to understand where different clinical specialties are included in existing pathways, how additional supporting roles could improve the patient experience, and how care can be better co-ordinated across the NHS. Co-ordinating care helps everyone. Healthcare professionals from different specialties and from different parts of Scotland should be able to quickly share expertise and consider the whole picture of a person's condition. People attending appointments may need to travel less and get more out of their appointments.

7.6 Pharmacogenomics

Pharmacogenomics is an area of scientific research that studies how a person's genome affects how they respond to certain medicines and allows us to 'target' how we use these medicines to reduce side effects and improve treatment. Increasing people's understanding of pharmacogenomics, and supporting its use across the NHS, will allow more people to benefit from treatment that can improve their lives, and promote the safe and more effective use of medicines.

7.7 Data and Digital Infrastructure

A data and digital infrastructure means having a way of safely storing people's genomic information, and making it easier to share among the people who need to use it to improve people's care within the NHS. It will also enable scientists and healthcare professionals to work together to get the most out of that information to improve people's lives.

7.8 National data returns

Linking genomic information to existing national registries (for example the cancer and rare disease registries) and national health and social care information sets managed by Public Health Scotland will help us to understand the impact of genomic testing, and how effective genomic medicine is in terms of improving people's lives. This can help shape decisions about where we need to invest in our health services. It can also help scientists and healthcare professionals to work together more easily, all of which will help improve our services and the care that people receive in Scotland.

7.9 Research, development and innovation

We know how important research is, so that services, care and treatment can keep improving and benefit from scientific discoveries and new knowledge. We want our genomic medicine service to enable and support research, development and innovation so that more people in Scotland can benefit.

7.10 Partnership working and future-proofing

An important part of this strategy is developing structures and partnerships across Scotland that are flexible to change as genomic medicine develops, and that allow us to offer more and more people access to the genomic testing that they need.

This publication is available at **www.gov.scot**

Any enquiries regarding this publication should be sent to us at

The Scottish Government
St Andrew's House
Edinburgh
EH1 3DG

ISBN: 978-1-83601-152-1 (web only)

Published by The Scottish Government, April 2024

Produced for The Scottish Government by APS Group Scotland, 21 Tennant Street, Edinburgh EH6 5NA
PPDAS1443226 (04/24)