

National Genetics and Genomics Strategy

National Clinical Programmes Conference

7th October 2022

Background



In alignment with the vision of Sláintecare and established clinical programmes such as the National Cancer Control Programme, Strategic Programmes, Office of the Chief Clinical Officer has progressed the development of a National Strategy for Genetics and Genomics with the establishment of a Steering Group. The purpose of this group is to develop a single national strategy for genetics and genomics in Ireland, encompassing care provided across acute, primary and community settings and spanning all ages.

The strategy seeks to determine

- The scope and scale of Ireland's genetics and genomics ambition
- The clinical and research priorities
- Plans for international collaboration and partnerships
- Plans for the reform of the existing clinical and laboratory services in Ireland.

In doing so seeks to address the following key issues

- Analysis and storage arrangements including biobanks
- Governance structures and legislative requirements
- Ethical principles and data privacy and security
- The required infrastructure, equipment and ICT data processing
- Staffing resources and training requirements.

Overview of Steering Group Expertise



Overview of the Focus and Leadership of the Working Groups

Workforce and Collaboration

The necessary skills to develop and attract a diverse workforce of genetic and genomic specialists, and on strategic partnerships and collaborations to raise the profile of Ireland as a leader in genetics and genomics on the world stage.

Prof Michael Gill and
Prof Brendan Loftus

Clinical Practice and Innovation

Ensuring the translation of advancements in genetics and genomics into healthcare services to optimise patient outcomes in a manner which is scientifically grounded, ethical and equitable in its approach.

Prof Owen Smith and
Prof Risteárd Ó Laoide

Data and Infrastructure

Identify priorities regarding the requirements for building upon and improving the country's current genetic and genomic infrastructure.

Prof Walter Kolch and
Prof Mark Lawler

Policy Communication and Engagement

Ensure the public and patients are involved, the patient voice is heard and genomic literacy is improved.

Review the requirements for ensuring informed consent and the policy and legislative requirement.

Deirdre McNamara and
Muiris O'Connor

High Level Overview of Activities Inputting into Strategy Development

- > 100 experts contributing
- > 31 documents submitted for initial recommendations
- 11 Patient Representative positions
- 2 Patient Advocacy webinars
- 2 Healthcare Worker webinars
- > 100 total queries received
- Steering Group Meetings
- Working Group Meetings
- Checkpoint Meetings.

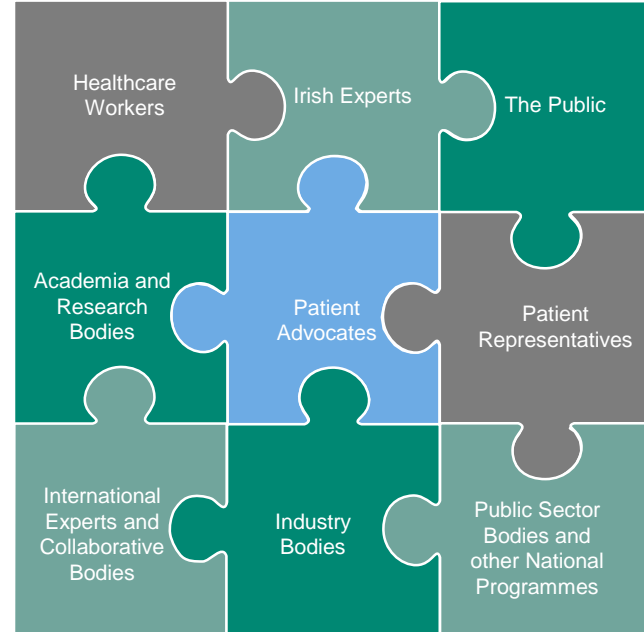


- Meetings with international experts to identify lessons learned
- Industry engagement
- Engagement with IPPOSI citizens jury
- Engagement with other National Working Groups
- Establishment of the current 'as is' in clinical service
- Involvement in international collaborations
- Review of international best practice
- Attendance at conferences.

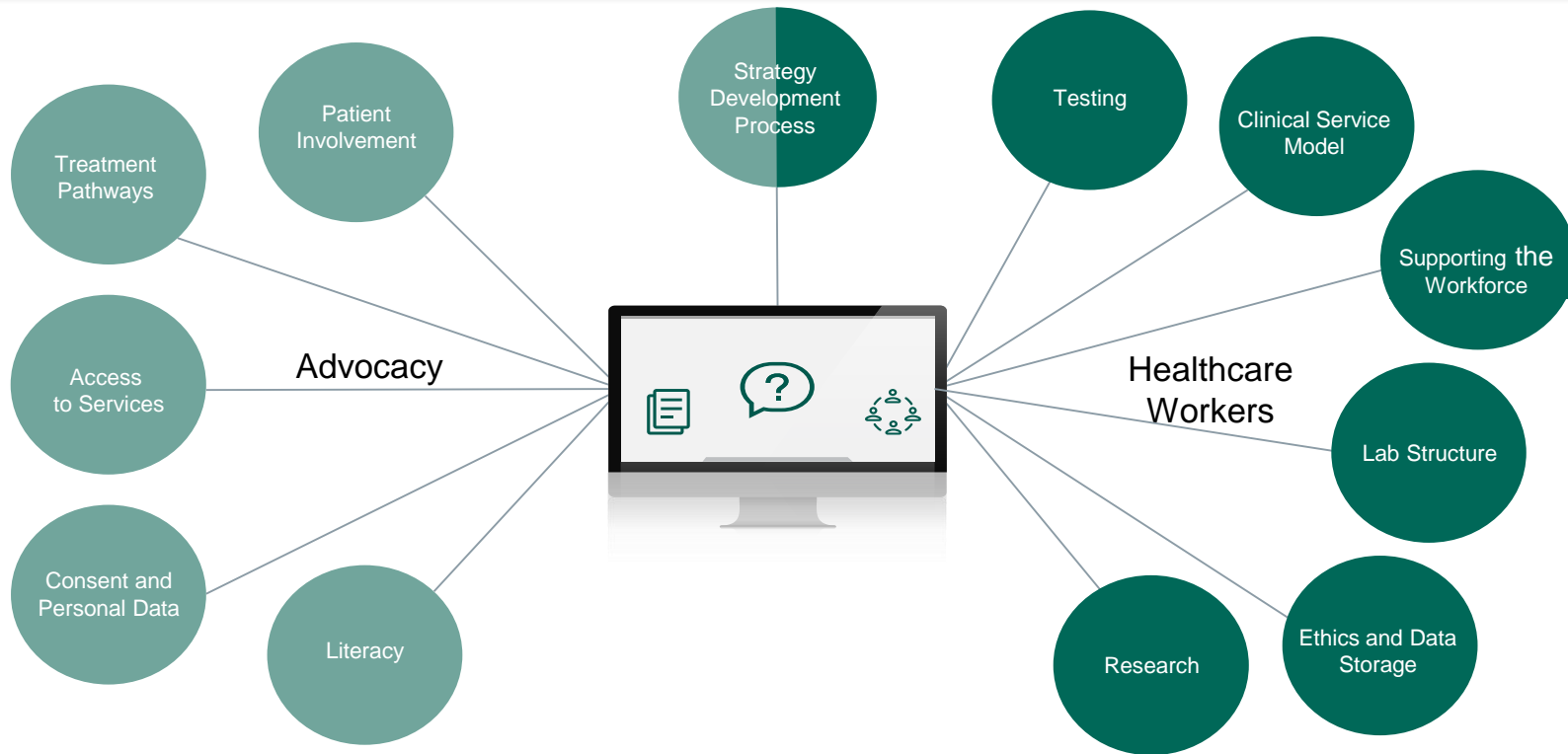
Stakeholder Engagement - Input into the Strategy

The development of this Strategy has benefitted from significant engagement with a diverse range of stakeholder groups.

Stakeholder engagement will continue into Q4 2022 through drafting of the Strategy document, and into 2023 with the implementation of the Strategy.



Emerging Themes from Engagements and Feedback



High Level Overview of Current Challenges in Genetic and Genomic Services in Ireland

Patient Representatives' Ask for Change

“Urgency to improve service - access & efficiency”

“National Genetics Services which are robust, with equivalent access no matter where you live”

“Genetic Counsellors embedded in clinical centres of excellence”

“Access for family members, support and follow-up”

“Literacy of the wider healthcare workforce, including GPs”



Lack of coordination and integration between clinical and research genomics



Insufficient clinical and laboratory infrastructure for genetics and genomics



Ireland lags behind other countries in utilising the power of genomics for patient benefit



Knowledge gaps in the workforce and in the general public



Shortage of trained specialists and long waiting times for services

Vision and Objectives



**Patient
Empowerment**



**Trust and
Engagement**



**Precision
Medicine**



**Workforce and
Training**



**Research and
Innovation**



**Global
Contributor**

Vision and Objectives



Patient Empowerment

Patients will be empowered to shape the development of patient and family -centred services through a transparent, shared decision making process supported by enhanced public and patient education on genetics and genomics

Vision and Objectives



Trust and Engagement

Effective public and patient engagement will build and maintain trust and ensure transparency about the ethical framework and legislation underpinning all aspects of services delivery and research including data ownership and the informed active consent process

Vision and Objectives



Precision Medicine

In order for patients to benefit from advances in precision medicine, Genetics and Genomics will be integrated into normal clinical practice at all stages of the patient pathway, while ensuring equitable, timely access to high quality services

Vision and Objectives



Workforce and Training

A skilled workforce will deliver specialised services and the wider healthcare workforce will confidently use genetic and genomic data to support the delivery of patient care. Our staff will be supported by enhanced and responsive education and training in genetics and genomics

Vision and Objectives



Research and Innovation

Genetic and genomic research and innovation will help to ensure the translation of new knowledge and advances in technology into improved patient outcomes. This will simultaneously drive economic growth through collaboration and partnership with academia and industry

Vision and Objectives



Global Contributor

Ireland will be a key contributor to advancing genomics and genetics globally through international collaboration, trusted partnerships with industry and building the right infrastructure and secure digital platforms and analytics to drive research and innovation

Guiding Principles

Patient and Family Centred

Services will be delivered in an integrated patient and family centred manner

Collaborative

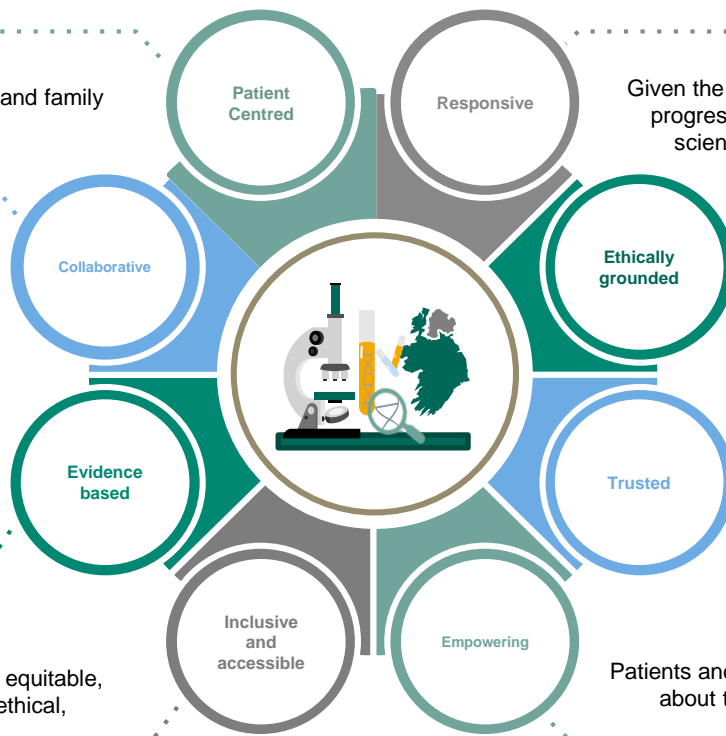
To advance knowledge and fuel innovation in genetics and genomics, we will work collaboratively both nationally and internationally, for the benefit of patients

Evidence based and Cost Effective

The translation of advancements in genetics and genomics into healthcare services will be evidence based and in the best interest of our patients while remaining cost effective and reducing waste

Inclusive and accessible

Genetic and genomic services and research will be equitable, inclusive, accessible, responsive and respectful of ethical, cultural and socio-economic diversity



Responsive

Given the speed at which the fields of genetics and genomics are progressing, we will remain agile and adaptable to advances in scientific knowledge and tools and their clinical and research applications.

Ethically grounded

The use of personal data will meet the highest ethical principles for clinical practice and research and reflect the voice of the citizens of Ireland

Trusted

Effective governance will ensure that Genetic and Genomic clinical services and research are delivered in a manner that fosters trust and builds public and patient confidence in precision medicine

Empowering

Patients and their families are empowered to make informed decisions about the use of genetic and genomic tests and health data in the delivery of their care



Challenges



Lack of coordination and integration between clinical and research genomics



Insufficient clinical and laboratory infrastructure for genetics and genomics



Ireland lags behind other countries in utilising the power of genomics for patient benefit



Knowledge gaps in the workforce and in the general public



Shortage of trained specialists and long waiting times for services

Addressing the Challenges

National Office for Genetics and Genomics

Hub and Spoke for Laboratory and Bioinformatic Service
National Centre of Excellence in Genomic Testing and Bioinformatics

Timely implementation of the priorities outlines in the National Genetics and Genomics Strategy

A national campaign will be developed and implemented to raise awareness of genetics and genomics and increase genetic and genomic literacy amongst the general public

A National Genetics and Genomics Workforce Plan will be developed by and implemented



Examples of Headline Priority Areas of Focus



Establishment of the National Genetics and Genomics Office

- A National Office will be established to provide oversight and governance of genetic and genomic service delivery. The National Office will (1) drive the implementation of the Strategy, (2) deliver service improvements in the short term to address urgent service deficiencies, and (3) lead on longer term continuous service development.



Integrated Patient-Centred Clinical Pathways

- Integrated multidisciplinary pathways will be in place, with care delivered locally by clinical specialities with a special interest in genetics and genomics. Specialties will be supported by local access to expertise in genetic counselling, in line with the Sláintecare vision.
- There will be a national approach to Clinical Genetics service delivery to support local and regional referrals of complex cases.



Developing the Specialised Workforce

- A workforce plan will be developed to support recruitment and retention of the specialist workforce for delivery of clinical and laboratory services.
- Access to accredited training programmes and enhanced educational support will be in place for specialised staff and the wider Healthcare Workforce, including GPs, to improve genetic and genomic literacy and awareness of clinical pathways.



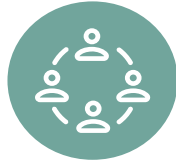
Hub and Spoke Approach to Laboratory Infrastructure

- The future structure for the delivery of genetic and genomic laboratory services will comprise of a centralised hub, delivering National WGS/WES and associated bioinformatics, with existing laboratories in the spokes supporting local service delivery and expanding capabilities to include NGS (panels) in selected local laboratories.
- Services will be supported by the establishment of a comprehensive national test directory to support timely, efficient and appropriate testing at each laboratory level.

Next Steps



**Strategy
Development
and Evidence
Gathering**



**Patient
Advocacy
Engagement**



**Healthcare
Worker
Engagement**



**Industry
Engagement**

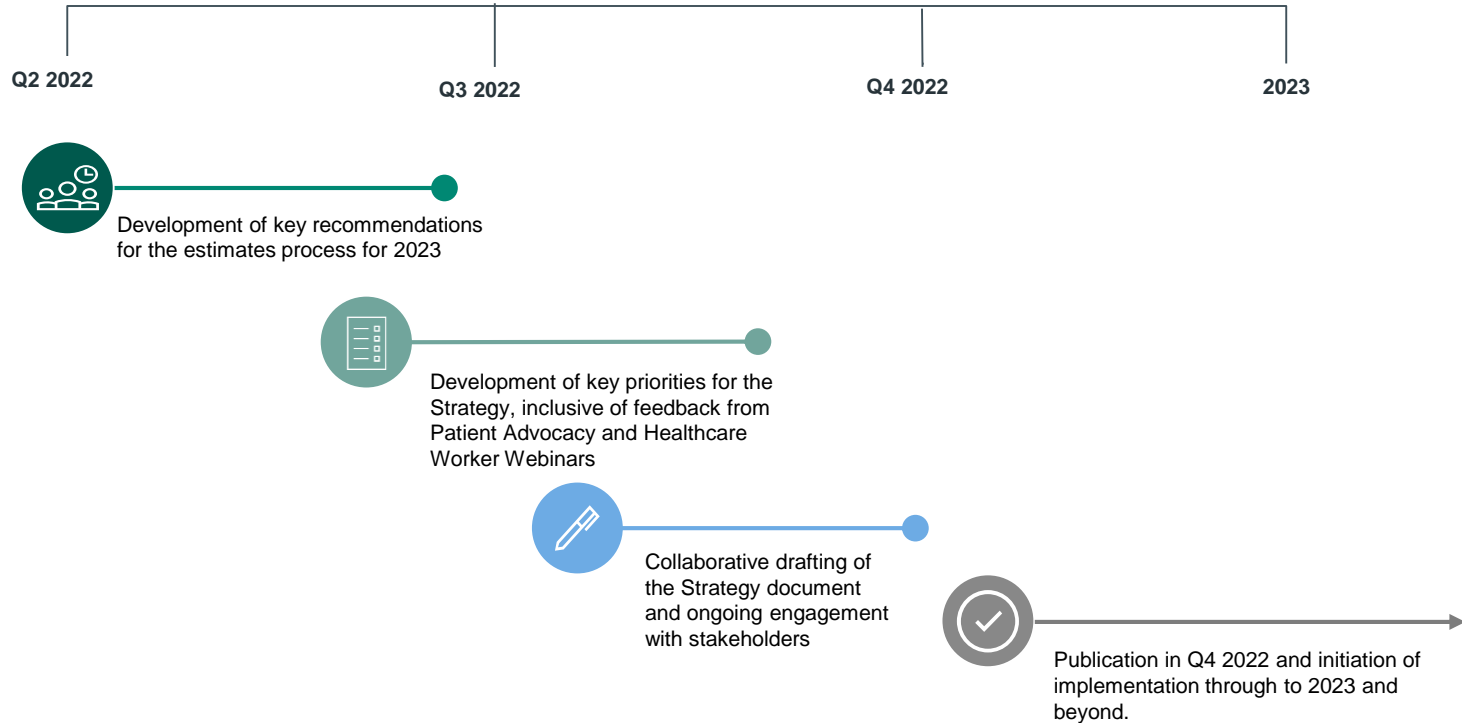


**International
Engagement**



**Focused
Engagement
with other Key
Stakeholders**

Timeline of Key Milestones for Development of the Strategy



Thank You

Contact Information

StrategicProgrammes.CCO@hse.ie