



# **National Strategy for Accelerating Genetic and Genomic Medicine in Ireland**

Annual Implementation Report 2024

February 2024

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## 1. Introduction

### 1.1 Background

The Health Service Executive (HSE) published the [National Strategy for Accelerating Genetic and Genomic Medicine in Ireland](#) in December 2022. This document was the result of the collaborative and inclusive development phase of the strategy. It involved engagement with patient representatives and advocates, over 100 experts and other key stakeholders including the Department of Health (DoH) and HSE national programmes.

In alignment with Sláintecare, the National Strategy outlines a vision of a sustainable, patient and family centred genetics and genomics service that is equitable and available to patients throughout their lifespan, across the country. A skilled multidisciplinary workforce will provide this service, supported by the National Genetics & Genomics Office and a strong governance structure. The office and service will be guided by patient and public involvement and it will support and engage with pioneering research and innovation.

Last year (2023) saw the start of the implementation phase of the National Strategy, it will continue over the next 4 years. This document will review the key achievements of 2023 and will look forward to 2024. The thirteen strategic areas of focus of the National Strategy outline the way forward for genetic and genomic services in Ireland over this five-year timeframe and beyond. These areas are the foundation of this implementation plan.

They map to five interconnected priority themes:

1. Coordinating a national approach to genetics and genomics,
2. Ensuring Patient and Public Involvement (PPI) and Partnerships,
3. Building the genetics and genomics workforce for the future,
4. Enhancing genetic and genomic clinical services, and
5. Strengthening infrastructures to drive advances in genetics and genomics.

### 1.2 Vision for Genetics and Genomics

The strategic vision for genetics and genomics Ireland is to develop a patient and family centred service that aligns with the values of Sláintecare, through its focus on equity of access and enhanced patient outcomes. This service will cover the patient lifespan and will be accessible across the Regional Health Areas (RHAs). The development of this strategy and its implementation over future years is a key step in actualising this vision.

Eight key principals underpin the National Strategy; they are the foundation of work carried out in the implementation phase:

- **Patient and family centred;** Services to be delivered in an integrated patient and family centred manner.

- **Responsiveness:** 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 application.
- **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 genetic and genomic medicine.
- **Empowering:** Patients and their families will be empowered to make informed decisions about the use of genetic and genomic tests and health data in the delivery of their care.
- **Inclusive and accessible:** Clinical genetics and genomics and associated translational research will be equitable, inclusive, accessible, responsive, and respectful of diversity in society.
- **Evidence-based and cost-effective:** The translation of advances 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.
- **Collaborative:** To advance knowledge and fuel innovation in genetics and genomics, we will work collaboratively, both nationally and internationally for the benefit of patients.

### 1.3 Key Strategic Areas of Focus

Table 1 lists the thirteen strategic areas of focus defined for the development of genetic and genomic services in Ireland. These areas of focus are mapped to the strategy’s priority themes to ensure service delivery is reflective of the strategy. The six 2024 priority deliverables for the year are mapped to five of the strategic areas of focus. This allows for visibility of the Strategy’s priority themes and strategic areas of focus within the detail of the annual implementation plan.

Table 1. Thirteen Strategic areas of focus for the development of genetic and genomic services

<b>Coordinating a national approach to genetics and genomics</b>	<b>1</b>	A national office for genetics and genomics will be established in 2023 under the governance of the HSE and will provide oversight and a standardised approach to the delivery of the genetics and genomics service as outlined in the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.
	<b>2</b>	The Department of Health will engage with stakeholders across the clinical, academic, research and non-profit sector to identify gaps in Irish policy and legislation. This engagement, in conjunction with wider public consultation, will be used to inform future legislative and policy action.

Ensuring Patient and Public Involvement (PPI) and partnerships	3	A national education and communication programme will be developed and implemented in 2023 to raise awareness of genetics and genomics and increase genetic and genomic literacy amongst patients and the public.
	4	Building and maintaining public trust and engagement will ensure sustainability and impact positively on service, research, and policy developments. Meaningful partnerships with the public will be established to ensure that the public and patient voice is at the heart of implementation of the strategy and in the design and development of any new services or initiatives.
	5	There will be a national approach to ensure that standardised guidance on consent for genetic and genomic clinical and research purposes is harmonised and developed in line with relevant guidelines and legislation.
Building the genomics workforce of the future	6	A National Genetics and Genomics Workforce Plan will be developed in 2023 to support the recruitment, retention, education, and career development of the current and future genetics and genomics workforce
Enhancing genetic and genomic clinical services	7	A suite of measures that ensure the delivery of safe, high-quality care will commence in 2023, and processes will be implemented to monitor performance against agreed targets to drive quality improvement.
	8	Locally integrated, multidisciplinary, patient and family centred care pathways will support the continued transition of genetics and genomics into mainstream healthcare by building on existing services, collaborative networks, and expertise to enhance service delivery in a manner that is efficient, equitable, and in accordance with the Sláintecare vision.
	9	Equitable, timely, and evidence-based availability of genetic and genomic tests and technologies in clinical practice will be improved through a coordinated and standardised national approach. The development of a National Test Directory will commence in 2023.
Strengthening infrastructure to drive advances in genetics and genomics	10	The national office will work with services to enhance existing laboratory infrastructure and informatics services to promote the development and use of innovative technologies for testing, sample tracking, and reporting

	11	A National Centre of Excellence in Genomic Testing and Bioinformatics will be established as a single entity, which will sit under the governance of the HSE.
	12	The national office will review existing genetic and genomic data capacity and capability and work toward the establishment of a secure, scalable, and accessible data and analytical infrastructure to support clinical service delivery, bioinformatics, data access, and research.
	13	Engagement, collaboration, and partnership with international organisations, industry, government, and academic partners will be key to enhancing Ireland’s research ecosystems. Procedures, processes, and guidelines will be developed to support the translation of advances in genetics and genomics into current and future clinical practice

### 1.4 National Genetic and Genomics Office Structure

In 2023, there was significant progress in the implementation of the National Strategy. This included the establishment of both the Implementation Steering Group (ISG) for Genetics and Genomics and the National Genetic and Genomics Office (NGGO). The NGGO sits within the Office of Chief Clinical Officer (CCO) of the HSE and reports to the HSE Executive Management Team (EMT) as requested. Figure 1. Illustrates the Strategy Implementation Governance Structure.

The current NGGO leadership team;

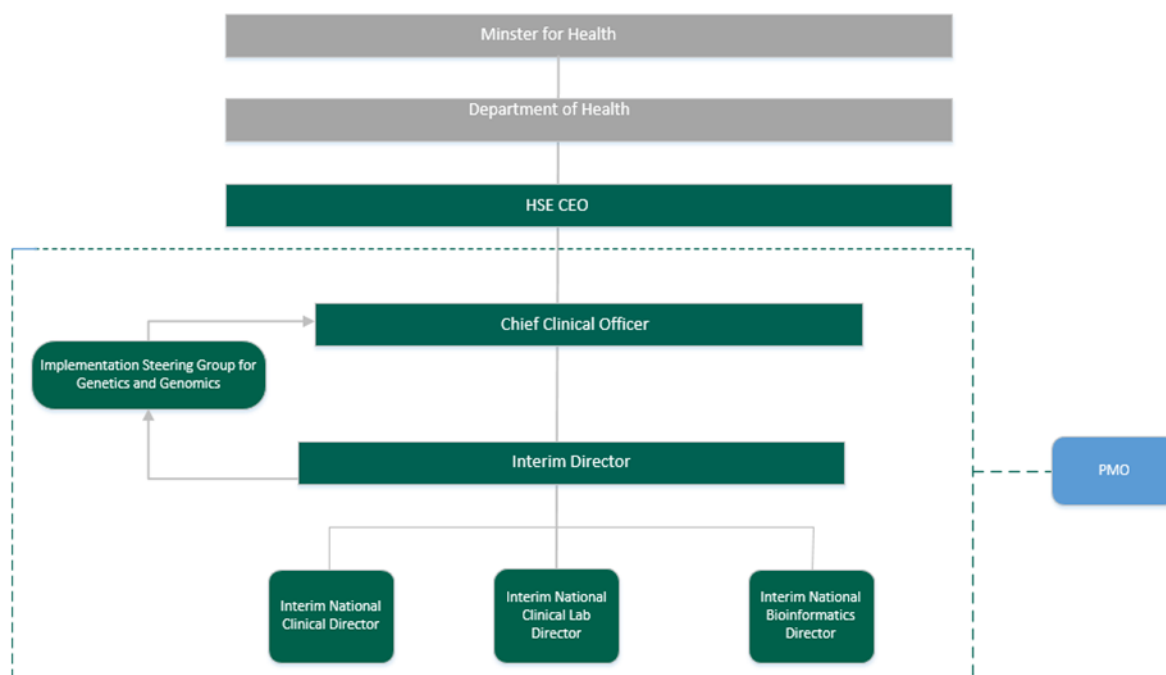
*Interim National Director Dr Mark Bale* was involved in the development phase of the national strategy. He has provided continuity between the development phase and the current implementation phase of the strategy.

*Interim National Clinical Director Dr Emma McCann* joined the NGGO in May 2023. Dr McCann is a Consultant Clinical Geneticist in Children’s Health Ireland at Crumlin.

*Interim National Clinical Laboratory Director Dr Richard Hagan* joined the NGGO in July 2023. Dr Hagan is a Chief Clinical Scientist in the Molecular Biology & Genetics Department of the Irish Blood Transfusions Service.

*Interim National Bioinformatics Director Dr Eppie Jones* joined the NGGO in July 2023. Dr Jones is a Senior Bioinformatician in The Cancer Molecular Diagnostics Laboratory, St James’s Hospital.

Figure 1. Strategy Implementation Governance



## 2. 2023 Implementation Review

### 2.1 2023 Priority deliverables

1. Development and agreement of the implementation plan and governance structures;
2. Establishment of an Implementation Steering Group for Genetics and Genomics to lead and drive the implementation of the National Strategy,
3. Establishment of a National Office for Genetics and Genomics and manage the recruitment of associated roles,
4. Manage the recruitment for frontline workforce roles, funded through the 2023 estimates process,
5. Development of a targeted communications programme to improve public and patient literacy,
6. Continue to collaborate internationally through participation in the 1+Million Genome (1+MG) European project,
7. Participate in the Genomic Data Infrastructure (GDI) project,
8. Development of a National Genetic and Genomic Workforce Plan,
9. Development of a National Test Directory for Genetics and Genomics,
10. Review of existing genetic and genomic data capacity and capabilities.



## 2.2 2023 Achievements

1. **Development and agreement of the implementation plan and governance structures;** A task force was established (January 2023), and had an objective of developing a roadmap to achieve the priority deliverables for 2023. The first annual implementation plan was approved by the HSE EMT and published in April 2023.

2. **Establishment of ISG for Genetics and Genomics to drive and lead the implementation of the strategy:**

The Implementation Steering Group (ISG) was established and held its first meeting in April 2023. The objectives of this group are to provide leadership and oversight to implementation activities and to monitor the progress of the implementation plan. Appendix no. 1 outlines the Terms of Reference for and current membership of this group. The ISG met 5 times in 2023.

3. **Establishment of the NGGO and manage the recruitment of associated roles:**

In order to proceed promptly with the 2023 implementation priority deliverables, the NGGO leads were recruited on an interim basis. The Interim National Clinical Director, Interim National Clinical Laboratory Director and Interim National Bioinformatics Director were in post by mid-2023. Other office team members hired in 2023; programme manager, business coordinator, communications officer and research analyst.

4. **Recruitment for frontline workforce roles, funded through the 2023:**

Progress against this priority was impacted by the HSE recruitment pause. Support and development of the frontline specialist clinical and laboratory genetics workforce will remain an NGGO priority throughout strategy implementation and beyond.

5. **Improve public and patient literacy:**

A research project was started in the second half of 2023, Amárach Research were engaged to assist the NGGO in carrying out this work. The aim of this research is to assess the learning needs of patients, at different stages of their patient journey and to understand how they would like information to be delivered. This will provide insights to inform the development of communications outputs, including style of outputs and required content, to increase genetic and genomic health literacy for patients accessing the national genomic medicine service. This body of work will continue into 2024.

The research question:

*What are the specific learning needs of patients, and parents/carers of patients who have been referred for a genetic assessment, those who have been offered a genetic or genomic test and what is the best way to communicate information to achieve the learning outcomes? What do they need to know about the next steps of the patient care pathway and what information can help them to better understand the processes involved in using Ireland's Genomic Medicine Service?*

IPPOSI (Irish Platform for Patients' Organisation, Science & Industry) published their Citizens' Jury on Genomics in 2022. In 2023, they commenced a programme of work to better understand the genomic literacy needs of the Irish public. The NGGO have supported

this project. In 2023, the NGGO joined the IPPOSI Genomic Education Working Group and will continue to participate in this work in 2024.

#### 6. Participate in the 1+MG European project (see appendix three)

In November 2022 Ireland was officially welcomed into Europe's 1+Million Genomes Initiative. In 2023 the Department of Health developed the 1+MG National Mirror Group. The 1+MG is organised across 13 different work streams, NGGO are represented on the following work streams by the four interim National Directors:

- Workstream 3 Data and Meta standards
- Workstream 4 Quality Standards
- Workstream 5 Data Access and Technical Inter-operability
- Workstream 7 Industry Involvement
- Workstream 10 Common and Complex Diseases
- Workstream 12 Genome of Europe

NGGO is a member of the DoH led 1+MG Mirror group.

The EU 1+M Genomes Initiative formally adopted the [2024-2027 Roadmap](#) outlining the activities at national and EU level to deliver precision medicine. The 1+MG work streams are currently under review and NGGO will engage with the DoH and 1+MG project to facilitate changes and participation.

#### 7. Participate in the EU Genomic Data Infrastructure (GDI) project

The Genomic Data Infrastructure (GDI) project a consortium involving 20 EU Member States with the goal of enabling access to genomics and corresponding clinical data across Europe by creating secure data infrastructure. GDI-IR is the Irish group in this European project. GDI-IR will establish best practices to manage genetic data in the Irish and European setting, protecting the security of the personal data contributed by individuals. Work will be informed by experience and technological developments, with European partners. GDI Ireland established a steering group in 2023 to support the GDI-IR project. Four meetings were held in 2023. NGGO also are members of the related Genome of Ireland Working Group.

#### 8. Development of a National Workforce Plan

The future workforce plan was deprioritised for 2023. It is a key priority for 2024. Development of a genetic and genomic education programme for the non- genetic specialised healthcare workforce in Ireland started in 2023. The RCPI (Royal College of Physicians Ireland) and the RCSI (Royal College of Surgeons Ireland) were engaged to provide webinars and education workshops to healthcare providers. Workshop 1 (from bases to genomes – what I need to know) and workshop 2 (how genetic variation arises from cell division) were held in 2023, with three further workshops planned for 2024.

## 9. Development of a National Genomic Test Directory

Work on the National Genomic Test Directory commenced in quarter 2, 2023; this was behind the 2023 schedule. The first two steps of the process were to identify a methodology suitable for the National Genomic Test Directory, and to prioritise clinical specialities for development of the National Genomic Test Directory.

It was decided that the initial development phases of the Irish National Genomic Test Directory would focus on rare and inherited diseases. Nephrology clinicians were selected as the first specialist group to review the relevant part of the directory to assess suitability for their clinical need. In quarter 4 2023 a workshop was held with nephrologists, and the engagement with this speciality will continue in 2024.

To enable the clinician's use of the genomic test directory, within this work stream a 'Testing Guidance Handbook' working group was established in 2023. The genomic testing guidance handbook has progressed, with many sections completed. Work on the handbook and development of the National Genomic Test Directory for Rare and Inherited Disease will progress in 2024 (2024 priority deliverable 1).

## 10. Review of existing data capacity and capabilities

This priority was deferred to 2024 and will form part of a key deliverable for 2024.

### 2.3 2023 focus on the guiding principles of the strategy

#### 2.3.1 Patient and Public Involvement

The National Strategy was developed with guiding principles of inclusivity, accessibility, trust, empowerment and being patient and family centric. With this in mind, the NGGO placed a strong focus on public and patient involvement from the start of implementation.

In June 2023, the NGGO established a Communications and Patient and Public Involvement (PPI) Working Group to provide advice, expertise and support to inform the development of communications activities to improve public and patient literacy and ensure patient and public voices are sufficiently incorporated into the design and delivery of genetic and genomic services. The working group has acted in an advisory and consultative capacity during the development stages of a genetics and genomics operating model, a genomic test directory and testing guidance for clinicians, and communications to the public to improve genomic health literacy. Further to this, the NGGO began a process to expand its outreach, inviting patients and the public to indicate their interest in participating in future PPI and engagement (PPIE) activities.

To date, those who have expressed interest in PPIE have been invited to provide suggestions and feedback on design options for on a national genomics service should be delivered. PPIE opportunities in Q4 2023 included online consultation workshops, which included patients, their advocates and healthcare professionals, and participation in quantitative and qualitative surveys.

### 2.3.2 NGGO Engagement

To ensure the strategy's guiding principles of collaboration and responsiveness in an evidence-based cost effective service are embedded into genomics processes from implementation, the NGGO team have engaged with partners and stakeholders extensively in 2023. Engagement will continue and develop throughout implementation. This will include ongoing participation in the 1+MG European and GDI projects.

The NGGO recognises its objective of accelerating a sustainable, equitable, patient and family centred genetics and genomics service is an objective shared throughout the HSE. The NGGO have engaged the National Women and Infants Health Programme (NWIHP), the National Cancer Control Programme (NCCP) and the National Rare Disease Office (NRDO) to ensure alignment on projects of shared interest, and have established a calendar of routine meetings.

See appendix 2 for an over view of the working group, professional stakeholders and committees with whom the NGGO have engaged.

### 2.3.3 An inclusive, accessible cost effective service

In 2023 the NGGO commissioned Deloitte Ireland to perform a strategic assessment to inform the future NGGO within the strategic theme of 'enhancing genetic and genomic clinical services'.

The project objectives were:

- To determine and understand the current genetic and genomic laboratory medicine operating model in Ireland including testing capacity, capabilities, knowledge, and resources.
- To undertake a comprehensive review of international best practice to establish key lessons learned and their applicability to genetic and genomic medicine in Ireland moving forwards.
- To develop and appraise various potential and feasible future operating models as part of a comprehensive process, informing the development of a proposed laboratory configuration and associated operating model.
- To develop a series of evidence-based recommendations with respect to the end-to-end operating model for consideration by the HSE.

Deloitte presented their findings to the NGGO in Q3 2023. The Deloitte review detailed a service that had shown growth over the years with local expertise, diverse funding sources, and a strong local drive to enhance these services, although these services are not currently organised into a cohesive national network. This has resulted in variation in technologies available at different sites, and short falls in domestic capacity giving rise to international outsourcing to a range of providers depending on patient, test and site requirements.

Deloitte described eight options to inform the future laboratory operating model, with a final recommendation of a Hub and Node model. Strategically placed nodes within the HSE regional health areas would integrate into acute hospital facilities to make use of existing services where applicable. These nodes would receive support including high volume and specialised testing, network leadership, quality assurance and professional development support from a genomic testing hub.

The NGGO held two well-attended online consultations in October 2023 involving patients and clinical experts to further explore the hub and spoke model for clinical services. NGGO will use this engagement process and related review to inform the ultimate operating model adopted by the HSE and to then progress the development of equitable national genetics and genomics services, including clinical genetics services, genomics laboratory capabilities and associated bioinformatics, a 2024 priority deliverable.

### 3. 2024 Implementation Plan

The NGGO has evaluated the progress made to date, on the strategy overall and the 2023 implementation plan. Based on this evaluation process the NGGO have identified six priority deliverables for 2024, in part these deliverables reflect those that were deferred from 2023.

#### 3.1 2024 Priority Deliverables

1. Development of a National Genomic Test Directory for Rare and Inherited Diseases.
2. Development of a National Genetic and Genomic Workforce Plan.
3. Continued Communications and PPI activity, progression of Communications Research, and development of proposed new information materials for clinicians and patients.
4. Continued development of equitable national genetics and genomics services, including clinical genetics services, genomics laboratory capabilities and associated bioinformatics.
5. Develop a national genetic and genomic data and technology roadmap with input through engagement with the national offices.
6. Continue to collaborate with research initiatives including Genomic Data Infrastructure and Genome of Ireland, and internationally through participation in the 1+Million Genome (1+MG) European project.

#### 3.2 2024 Implementation Plan

The 2024 key deliverables are aligned to the relevant areas of focus (see table 1) to help identify the longer term 5 year implementation plan, based on the strategic vision.

## **2024 Priority Deliverable 1:**

### *Development of a National Genomic Test Directory for Rare and Inherited Disease.*

**ENHANCING GENETIC AND GENOMIC CLINICAL SERVICES**  
**STRATEGIC AREA OF FOCUS (9): NATIONAL GENOMIC TEST DIRECTORY AND GENOMIC TESTING GUIDANCE**  
**HANDBOOK DEVELOPMENT AND IMPLEMENTATION**

This work stream follows on from the work completed in 2023. Primary activity for quarter 1 is assessment of clinical demand and suitability of existing international test directory models through NGGO led speciality workshops with (rare disease) European Reference Network (ERN) leads, national programmes and other appropriate speciality leads. These directory champions will then initiate wider workshops within their speciality, engaging with clinicians nationally, in adult and paediatric services, and patient representatives. At this stage of progress, the PPI working group will be engaged on test directory progress, and feedback sought. These workshops will inform draft version 1 of the National Genomic Test Directory for Rare and Inherited Disease.

Following on from development of draft version 1 and a clinical need gap analysis; quarter 4 of 2024 will see a second stage of assessment of clinical demand and suitability. This will assist in the expansion of the testing included in the directory, and of the refinement of the offering to meet the needs of the Irish patient and their clinician. This stream of work will carry through into 2025. Draft version 1 of the test directory will be one of the initiation documents used to begin the work stream for 2024 Priority Deliverable 4: Continued development of equitable national genetics and genomics services, specifically genomics laboratory capabilities and associated bioinformatics.

#### *Key Outputs & Enablers:*

- Quarter 1: Phase I specialty workshops  
Feedback from workshops  
Feedback from PPI engagement session
- Quarter 2: Draft version 1 of National Genomic Test Directory for Rare and Inherited Disease  
Version 1 Genomic Testing Guidance Handbook
- Quarter 3: National Genomic Test Directory for Rare and Inherited Disease gap analysis
- Quarter 4: Phase II specialty workshops for Draft version 2 of National Genomic Test Directory for Rare and Inherited Disease.

## 2024 Priority Deliverable 2:

*Development of a National Genetic and Genomic Workforce Plan.*

**THE GENETICS AND GENOMICS WORKFORCE FOR THE FUTURE**  
**STRATEGIC AREA OF FOCUS (6): NATIONAL GENETICS AND GENOMICS WORKFORCE PLAN DEVELOPMENT AND IMPLEMENTATION**

This activity was deferred in 2023 and is now a priority deliverable in 2024 with work commencing in quarter 1. The NGGO will use the DoH National Strategic Framework for Health and Social Care Workforce Planning (Working Together for Health) and the HSE HR 2019-2024 'Health Services People Strategy' to guide this work stream with the assistance of the HSE Strategic Workforce Planning and Intelligence Unit. An approach to inform the development of a workforce plan will guide the progress in this strategic area of focus.

Quarter 1 will see work done to establish the 'as-is' of the current clinical and laboratory genomics speciality workforce, and the establishment of a workforce planning (WFP) working group. To create this working group the NGGO will engage with patient representatives, national offices (NCCP, NWIHP, NRDO) and other relevant professional stakeholders (HSCP leads in DoH and HSE, the National Doctors Training & Planning, Genetic Counsellors). Quarter 1 will also see the start of a project of work on the creation of appropriate grade codes for specialist genetic and genomic workforce roles. This work will be done in collaboration with the Strategic Workforce Planning & Intelligence, National HR Directorate.

Following completion of the 'as-is' review and an examination of the influences on the current workforce in quarter 2, the process of strategic planning for this dynamic and agile future workforce will begin in quarter 3. Through the working group a service specification for a nationally equitable clinical genetic service will be developed. This will provide a genomics service for all patients, including those in perinatal, cancer and clinical genetics streams. The workforce plan will be presented to HSE EMT by the end of Q4 2024. The laboratory workforce plan will be done in collaboration with the Laboratory Services Reform Programme.

Within this workforce planning process, education and training needs of the specialist and mainstream genetics and genomics workforce will be examined, the genetic and genomic education programme for the mainstream workforce through the RCPI and the RCSI will continue in the first half of the year. New webinars and education workshops will be produced on topics such as 'what test should I order & what is that test looking at (and not)?'; 'How do I interpret the results of genetic testing?' and 'What's Coming down the line? - Horizon Scanning'

### *Key Outputs & Enablers:*

Quarter 1: HSE HR Workforce planning steering group notification  
'As-is' assessment of current clinical and laboratory workforce

- Current speciality workforce engagement
- Workforce working group established
- Education programme webinars
- Quarter 2: Literature review
- 'As-is' of current education streams
- Grade code review
- Education programme webinars
- Quarter 4: Workforce and education plan published

### **2024 Priority Deliverable 3:**

*Continued Communications and PPI activity, progression of Communications Research, and development of proposed new information materials for clinicians and patients.*

**ENSURING PATIENT AND PUBLIC INVOLVEMENT (PPI) AND PARTNERSHIP**  
**STRATEGIC AREAS OF FOCUS (3,4): NATIONAL EDUCATION AND COMMUNICATION PROGRAMME, PPI IN SHAPING SERVICES, RESEARCH, AND POLICY DEVELOPMENTS**

The communications research project, which commenced in Q4 2023 to identify the information needs of patients referred to genomic services, will continue through to completion by Q1 2024. A key output of this work will be a report outlining the research findings and recommendations for how the HSE can address information gaps among patients and their carers and families to improve the patient experience of using genetic and genomic services. In Quarter 2, a plan will be developed for the development of communications materials, arising from the knowledge gained from the research project.

Throughout 2024, increasing awareness and visibility of the NGGO will be a key priority. In line with the progression of the 2024 deliverables, the NGGO will undertake communications activities to engage with key stakeholders on progress and achievements. These activities will take form in information webinars for healthcare professionals, interested advocacy organisations and patient groups, as appropriate.

#### *Key Outputs & Enablers:*

- Quarter 1: Patient information needs research project reported
- Quarter 2: Patient communication materials plan developed
- Quarter 4: Phase one of patient communication materials available



### **2024 Priority Deliverable 4:**

*Continued development of equitable national genetics and genomics services, including clinical genetics services, genomics laboratory capabilities and associated bioinformatics*

#### **ENHANCING GENETIC AND GENOMIC CLINICAL SERVICES**

**STRATEGIC AREA OF FOCUS (8): DEVELOPMENT OF LOCALLY INTEGRATED, MULTIDISCIPLINARY, PATIENT AND FAMILY CENTRED CARE PATHWAYS AND TRANSITION OF GENETICS AND GENOMICS INTO MAINSTREAM HEALTHCARE BY BUILDING ON EXISTING SERVICES, COLLABORATIVE NETWORKS, AND EXPERTISE TO ENHANCE SERVICE DELIVERY.**

2024 will see the recruitment of frontline genetics and genomics posts, approved through the National Service Development plan. Work on the steps to be taken to ensure that all genetic and genomic professionals have appropriate role recognition will start in quarter 1.

Development of Genomics laboratory capabilities and associated bioinformatics will be informed by the progress of deliverables one, two and five. 2024 will see the NGGO determine, locate and operationalise laboratory processes.

EMT has approved the approach to iteratively produce a first draft of a National Genomic Test Directory but noting that operationalising this – i.e. providing the necessary laboratory services for clinicians to order agreed genomic tests – will be dependent on relevant staff resources, funding, laboratory and bioinformatics infrastructure, and governance.

In quarter 1 priority work for this deliverable will be on the identification of current test providers (in Ireland and/or overseas) and assessment of their accreditation, participation in EQA, Quality Assurance, IVDR legislation, turnaround time criteria.

In quarter 2 priority will be given to identification of available and appropriate laboratory space for efficient DNA extraction and analysis based on draft version one of the National Genomic Test Directory. This will reflect the Genomic Testing Guidance Handbook for arrangements on requesting tests, sample reception for samples currently sent away (inside and outside Ireland) by hospitals. Systems for electronic sample referral, tracking and report return will be identified.

Once ‘send-away’ samples are routed through this laboratory, this information will enable future implementation in this strategic area of focus, along with the Deloitte report, clinical operating models and clinical needs work.

#### *Key Outputs & Enablers:*

- Quarter 2: Assessment of current on-island laboratory capacity  
DNA extraction / wet laboratory capacity identified.  
Process of role recognition for genetic and genomic roles commenced.
- Quarter 3: Agreement completed to develop site for DNA extraction / wet laboratory  
System for electronic tracking and reporting identified
- Quarter 4: Begin to manage and track ‘send-away’ samples through DNA extraction / wet laboratory

### **2024 Priority Deliverable 5:**

*Develop a national genetic and genomic data and technology roadmap with input through engagement with the national offices.*

**STRENGTHENING INFRASTRUCTURE TO DRIVE ADVANCES IN GENETICS AND GENOMICS**  
**STRATEGIC AREA OF FOCUS (12): ESTABLISH DATA AND ANALYTICAL INFRASTRUCTURE**

The 2023 key priority deliverable (10) to review existing genetics and genomics data capacity and capabilities was deferred to 2024. The NGGO will complete a baseline analysis of the existing infrastructure in Ireland and collate international examples of genetic and genomic data and technology infrastructures. This links with work on best practice guidelines and legislation as part of the Genomic Data Infrastructure project, Digital Europe Programme and the Horizon Europe Partnerships on Rare Disease for Personalised Medicine.

The NGGO will collaborate with the PPI Working Group to understand how best to incorporate PPI into this work stream. The public, patient representatives and advocacy groups will also be engaged to understand how to best meet the needs of service users and their families. The NGGO will work with HSE internal experts such as the department of the Chief Information Officer (CIO) and eHealth Ireland to identify opportunities for the integration of genomic data into current infrastructure and future plans,

The NGGO will work with clinical programme such as NSS, NCCP, NWIHP and NRDO to inform the roadmap and to ensure it needs the needs of their patient cohorts, Engage with GDI to inform the scope of the roadmap (infrastructure, data sharing, and research).

#### *Key Outputs & Enablers:*

Quarter 1: Engagement with HSE CIO, DoH, HIQA and HSE identify relevant initiatives to implement the Genomic Data Infrastructure and European Health Data Space regulation.

Quarter 2: Scope future needs for laboratory networks, including genomic data storage and access needs based on Priority Deliverable 4 (test directory and guidance) and engagement with NCCP, NSS, NWIHP and NRDO.

Quarter 3: Roadmap development and PPI + stakeholder engagement

Quarter 4: Finalise genetic and genomic data and technology roadmap

**2024 Priority Deliverable 6:**

*Continue to collaborate with research initiatives including Genomic Data Infrastructure and Genome of Ireland, and internationally through participation in the 1+Million Genome (1+MG) European project.*

**COORDINATING A NATIONAL APPROACH TO GENETICS AND GENOMICS, STRATEGIC AREA OF FOCUS (2):  
POLICY AND LEGISLATIVE REQUIREMENTS**

*Key Outputs & Enablers:*

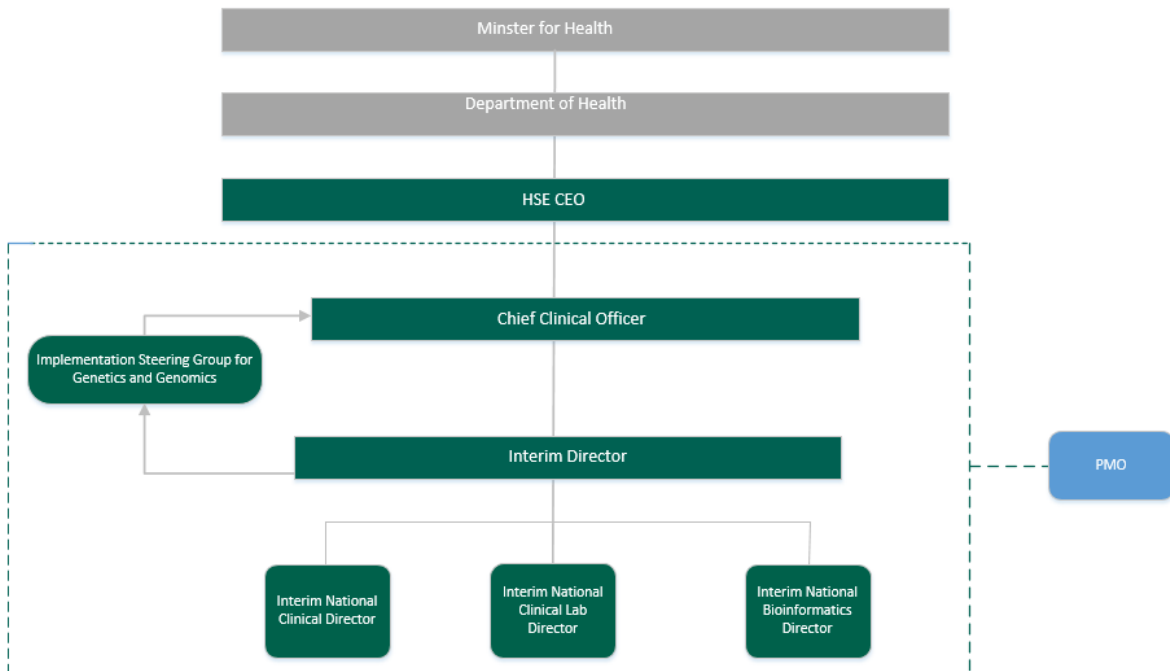
This priority deliverable is continued through from 2023 NGGO work. Appendices 2 and 3 show the NGGO engagement in associated working groups and committees, and the office's stakeholder interaction. The NGGO will continue its work in the 1+Million Genome European Project, 1+MG National Mirror Group and Genomes of Ireland working groups and in the GDI Ireland (GDI-IR) project.

Ireland has submitted an application, via RCSI, to participate in the Genome of Europe initiative. This will initially involve 100,000 Whole Genome Sequences representing the European population. NGGO will work with RCSI who are coordinating further work to identify the use cases that will benefit from an improved Irish (and European) reference genome database. This also aligns with the plans for GDI in Ireland.

The NGGO is continuously engaging with other initiatives, and through 2024 the NGGO will remain open to new and developing research opportunities.

## Appendix One:

### National Genetics and Genomics Office Implementation Steering Group TOR



#### Terms of Reference and membership

##### Purpose of the Implementation Steering Group for Genetics and Genomics

The purpose of this group is to lead and drive forward the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland as per the agreed implementation plan. The group will provide leadership and oversight to the implementation activities as well as monitor and report on the progress of the implementation plan.

##### Responsibilities of the Implementation Steering Group for Genetics and Genomics members

- To contribute, provide insight and advice regarding the operation and implementation of the strategy as it progresses;
- To share relevant information with members of the Implementation Steering Group for Genetics and Genomics;
- To provide advice within their area of expertise and to act as a conduit of information to their parent organisation, committee or clinical groupings;
- To contribute to an individual Workstream if requested to do so by the Implementation Steering Group for Genetics and Genomics in collaboration with the HSE Chief Clinical Officer and Interim Director for the National Genetics and Genomics Office.

##### Structure & Membership

The Implementation Steering Group for Genetics and Genomics will:

- Be chaired by the HSE Chief Clinical Officer and be supported by the National Genetics and Genomics Office;
- The meetings will be attended by the senior staff of the National Genetics and Genomics Office, including:

- Interim National Clinical Director;
- Interim National Clinical Laboratory Director;
- Interim National Bioinformatics Director.
- As appropriate, recommended Working Groups will be formed to carry out activities on its behalf, for example, specific reviews or analysis, implementation guidelines etc. to be completed.

### Reporting and Governance

The Interim Director for the National Genetics and Genomics Office will report on activities relating to this work to the Implementation Steering Group for Genetics and Genomics and the HSE Chief Clinical Officer. Individual members will maintain existing reporting lines in place through their own organisations.

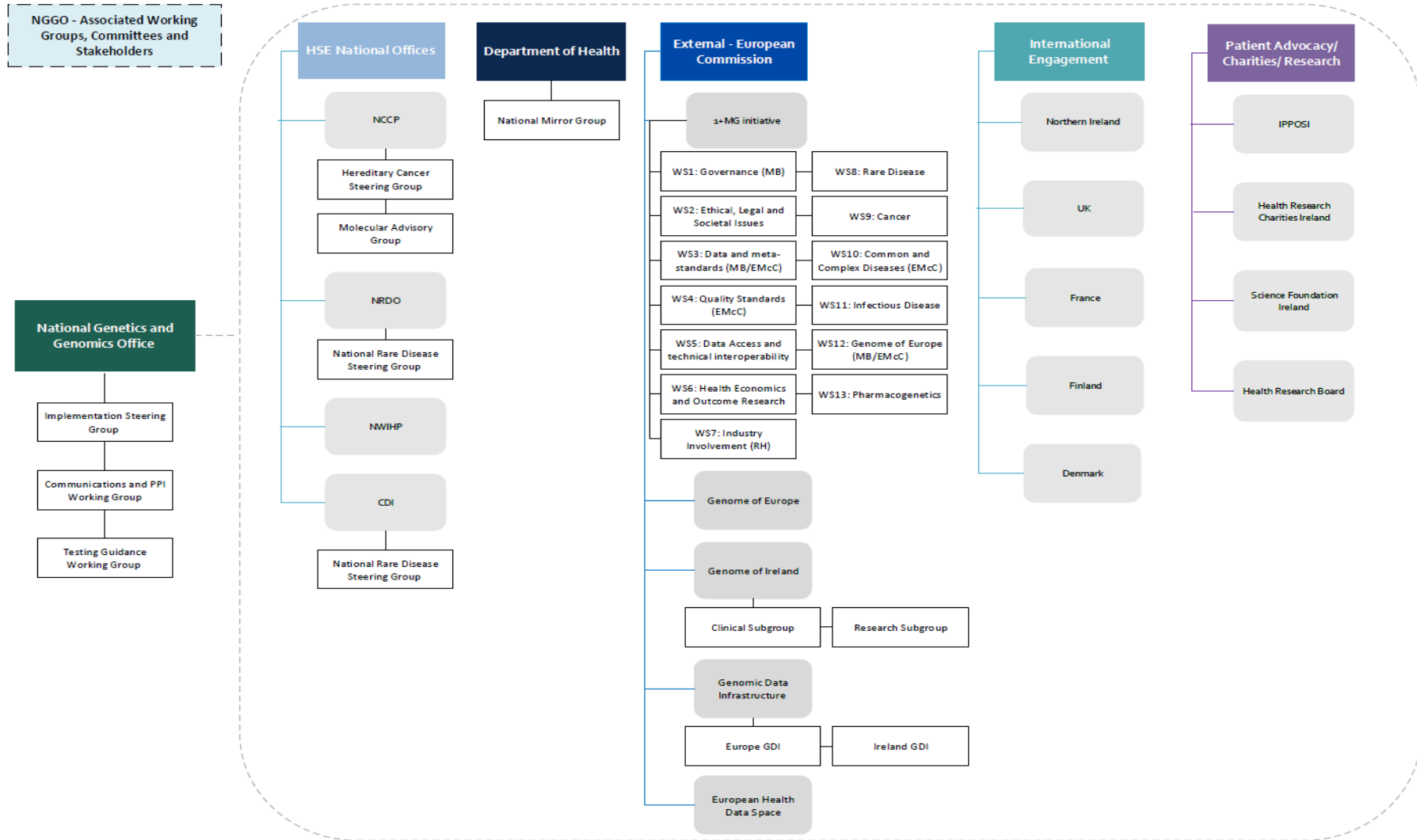
To ensure the coordination of national genetics and genomics activity and a common approach to European engagement, the appointed NGGO representative to the DOH 1+MG National Mirror Group, will be responsible for reporting on this group's activity to the National Mirror Group and the DOH representative on this group for reporting updates from the National Mirror Group back to this Steering Group. Similarly, updates regarding Ireland's participation in the European Reference Networks for Rare Diseases and associated Horizon Europe Partnerships<sup>2</sup>, will be provided to this Steering Group as necessary.

### Terms of Reference and membership

Membership of the Implementation Steering Group for Genetics and Genomics	
<b>Dr Colm Henry</b>	Chief Clinical Officer, HSE
<b>Dr Mark Bale</b>	Interim Director for the National Genetics and Genomics, HSE
<b>Deirdre McNamara</b>	Director of Strategic Programmes, Office of the CCO, HSE
<b>Martina Burns</b>	Assistant National Director, Office of the Chief Information Officer, HSE
<b>Margaret Cuddigan</b>	Patient Representative
<b>Marie Culliton</b>	Scientific Lead, HSE National Clinical Programme for Pathology
<b>Professor Mary Day</b>	National Director of Acute Operations, HSE
<b>Valerie Walsh</b>	Office of the Chief Financial Officer, HSE
<b>Eilish Hardiman</b>	CEO, Children's Health Ireland
<b>Eleanor Masterson</b>	Chief Architectural Advisor, HSE Estates
<b>Avril Daly</b>	Patient Representative
<b>Dr Cliona Murphy</b>	HSE National Women and Infants Health Programme
<b>Professor Risteárd Ó Laoide</b>	Director of HSE National Cancer Control Programme
<b>Christopher Ryan</b>	Head of Research Services & Policy, Department of Health
<b>Professor Eileen Treacy</b>	Clinical Lead for the HSE National Rare Disease Office
<b>Oonagh Ward</b>	Head of Research and Innovation Infrastructures, Health Research Board
<b>Dr Philippa Ryan Withero</b>	Assistant National Director, National HR, Integrated Health Workforce Planning, HSE
<b>Leah Dowdall / Ailish Kelly</b>	Assistant Principal Officer, Department of Health (Alternate Member)
<b>Prof Martin Cormican</b>	National Clinical Lead for Laboratory Services Reform

<b>Dr Emma McCann</b>	Interim Clinical Director for the National Genetics and Genomics, HSE
<b>Dr Richard Hagan</b>	Interim Clinical Laboratory Director for the National Genetics and Genomics, HSE
<b>Dr Eppie Jones</b>	Interim Bioinformatics Director for the National Genetics and Genomics, HSE

Appendix Two: NGGO working groups, committees and stakeholders



### Appendix Three: 1+Million Genome European Project

The Roadmap for 2023-2027 consists of the following delivery tracks and use cases relevant to NGGO and other programmes in Ireland

