



**An Oifig Náisiúnta Géineolaíochta  
agus Géanómaíochta  
HSE National Genetics and Genomics Office**

**April 2025**

## Contents

<b>1. INTRODUCTION .....</b>	<b>3</b>
<b>2. 2024 ACHIEVEMENTS.....</b>	<b>4</b>
<b>1. Priority Deliverable .....</b>	<b>4</b>
Develop a National Genomic Test Directory for Rare and Inherited Disease .....	4
<b>2. Priority Deliverable .....</b>	<b>5</b>
Development of a National Genetic and Genomic Workforce Plan .....	5
<b>3. Priority Deliverable .....</b>	<b>6</b>
Through continued Communications and PPI activity, progression of Communications Research, and development of proposed new information materials for clinicians and patients .....	6
<b>4. Priority Deliverable .....</b>	<b>7</b>
By continuing to develop equitable national genetics and genomics services, including clinical genetics services, genomics laboratory capabilities and associated bioinformatics.....	7
<b>5. Priority Deliverable .....</b>	<b>8</b>
Develop a national genetic and genomic data and technology roadmap with input through engagement with the national offices. ....	8
<b>6. Priority Deliverable .....</b>	<b>8</b>
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.....	8
<b>3. FUTURE DEVELOPMENTS .....</b>	<b>8</b>
<b>4. THE NATIONAL GENETICS AND GENOMICS OFFICE .....</b>	<b>9</b>

## 1. Introduction

The Health Service Executive (HSE) published the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland (the Strategy) in December 2022. The development of the Strategy involved engagement with patient representatives and advocates, over one hundred experts, and other key stakeholders including the Department of Health (DoH) and HSE national programmes.

In alignment with Sláintecare, the Strategy outlines a vision for 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 a strong governance structure. Development of this service will be informed by patient and public involvement, and it will support and engage with pioneering research and innovation.

The National Genetics and Genomics Office (NGGO) was established in 2023 to implement the Strategy. The thirteen strategic areas of focus of the Strategy outline the way forward for genetic and genomic services in Ireland and 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.

The NGGO identified six priority deliverables for 2024 that were aligned to the Strategy's themes and the related areas of focus. This allowed for visibility of the themes and areas of focus within the detail of 2024 implementation plans and projected outcomes. This report will review the key achievements of 2024 and outline the NGGO's 2025 priority deliverables.

## 2. 2024 Achievements

In 2024 the National Genetics and Genomics Office planned to achieve six priority deliverables:

1. Development of a National Genomic Test Directory for Rare and Inherited Disease.
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. Development of a national genetic and genomic data and technology roadmap with input from the HSE's 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.

### 1. Priority Deliverable

Develop a National Genomic Test Directory for Rare and Inherited Disease

#### ENHANCING GENETIC AND GENOMIC CLINICAL SERVICES

Work on this priority deliverable commenced in 2023 and the first version of the National Genomic Test Directory for Rare and Inherited Disease (Test Directory) was published for advisory purposes at the end of 2024. The Test Directory sets out the most appropriate genomic test that can be requested for a clinical indication, the clinical eligibility criteria for a patient, the most appropriate point in the clinical pathway to request the test and the clinical specialties that can request the test.

At the end of 2023, the NGGO held a series of meetings and workshops with clinical specialties to discuss commencing work on the Test Directory. By the end of 2024, most specialties concluded the initial feedback process, and the remaining specialties will be contacted during 2025 as part of further rounds of engagement.

Version 1 of the Test Directory includes clinical indications separated into five Parts identified under the headings: Cardiology, Lipids, Metabolic Medicine, Mitochondrial and Ophthalmology. The production of this version required a more in-depth, advanced 6-month process of input and engagement with clinicians from the

associated specialties. This process ensured Version 1 reflected the needs of the Irish patient population.

The Test Directory and a related 'Frequently Asked Questions' document are both available on the NGGO webpage on the HSE website here: [National Genetics and Genomics - HSE.ie](https://www.hse.ie/national-genetics-and-genomics)

A joint HSE and Department of Health launch of the Test Directory is scheduled for the end of January 2025. The expansion of the Test Directory is now an ongoing body of work for the NGGO with plans to release further versions of the Test Directory incorporating additional specialties during 2025 and 2026.

## 2. Priority Deliverable

### Development of a National Genetic and Genomic Workforce Plan

#### **BUILDING THE GENETICS AND GENOMICS WORKFORCE FOR THE FUTURE**

During the year, the NGGO worked closely with HSE National Doctors Training and Planning (NDTP) to develop a [Medical Workforce Plan for Clinical Genetics](#). This stakeholder-informed review will inform specialist training capacity in Clinical Genetics and will enable the NDTP to plan training requirements for an expanded medical workforce in the coming years as envisaged by the Strategy. This report is an update of the 2019 plan for the specialty.

The plan acknowledges the role of the NGGO in the development of clinical genetics services and the impact this may have on workforce modelling and projections, and on the need to review the plan in the near future. The Medical Workforce Plan for Clinical Genetics will be available on the NDTP website in January 2025.

In 2024, the NGGO began developing a submission for the eligibility criteria (and grade codes) for genetic counsellor posts. Once submitted and approved, these codes will distinguish genetic counsellors within the health service workforce, will ensure recognised job descriptions are available for each grade, and additionally will facilitate the HSE to accurately identify and count the current genetic counsellor workforce. This will assist in future workforce planning.

The NGGO recognises the work outstanding under this deliverable and in 2025 the Office will work to produce the first Strategic Genomics Workforce Plan.

### 3. Priority Deliverable

Through 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**

The NGGO Communications and Public and Patient Working Group continued their activity in 2024. Membership of the Group includes patient representatives and advocates, a clinical geneticist, genetic counsellor, research advocacy expert, and a member of the HSE Communications team. It is co-chaired by Assistant National Director of the NGGO and the CEO of Health Research Charities Ireland. The group met five times during the year with an additional questions and answers session with the Chief Clinical Officer. A specific questions and answers session was held with the Working Group on the development of the NGGO 5-year plan.

During 2024, Amárach Research completed a piece of qualitative research commissioned by the NGGO on Genomic Health Literacy in Ireland. The final report is available [here on the NGGO webpage](#). The aim of the research was to provide insights into the information needs of patients (and their family) using genetic and genomic services at different stages of patient journeys, and to identify how best to address these needs.

The research identified a timeline with possible points of information or education need along the patient journey and the NGGO has used the findings to map the development of education materials to meet the service user and patients' needs. In Quarter 3 2024, the NGGO invited members of the extended patient panel to join a project group to create and develop a series of patient information leaflets. The time commitment for the pilot project to develop the first leaflet involved three meetings held virtually and email interactions between meetings. The HSE communications team assisted the project group throughout the development of the leaflet. The leaflet is titled 'Genetic and Genomic Testing' and was approved for plain English by the National Adult Literacy Association (NALA). It will be available on the HSE website in early 2025.

The NGGO commissioned a webinar series from the RCSI aimed at improving genomic knowledge among clinicians. The series was completed in Quarter 3 2024 and is now available for all interested HSE staff to access on HSELand: Search for 'Fundamentals of Genetic and Genomic Testing@ when you log into HSELand here: <https://www.hseland.ie/dash/Account/Login>

The series consists of 7 modules:

Module 1: From Bases to Genomes - What I Need to Know.

Module 2: How Genetic Variation Arises from Cell Division.

Module 3: When Should I Test?

Module 4: What Test Should I Order and What Is That Test Looking at (and not)?

Module 5: How Do I Interpret the Results of Genetic Testing?

Module 6: Horizon Scanning - What's Coming Down the Line?

Module 7: Ethical Considerations in Genetic Testing and Newborn Screening.

#### 4. Priority Deliverable

By continuing to develop equitable national genetics and genomics services, including clinical genetics services, genomics laboratory capabilities and associated bioinformatics

#### ENHANCING GENETIC AND GENOMIC CLINICAL SERVICES

In 2024, funding was made available to expand Clinical Services. A Consultant Clinical Geneticist post, 3 Genetic Counsellor posts and a Genomic Resource Associate were funded. The NGGO expects recruitment of these posts to be completed by the second quarter of 2025.

In 2024, the NGGO commenced preparation for the development of a centralised National Genomic Processing Service for the pre-and post-analytical aspects of delivering the Test Directory. This is a first step in developing genomic laboratory capabilities. In 2025, the location of this service will be determined by an Expression of Interest and Invitation to Participate process. The National Genomic Processing Service (NGPS), once in place, has been designed to ensure that for samples sent abroad for testing, the right test will be requested for the right patient, at the right time, and requested by the right person; this will improve patients' experience of genomic testing.

## 5. Priority Deliverable

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

In 2024, the NGGO commenced proposals to develop a Bioinformatics and Genetic Data Infrastructure Roadmap. This is a key piece of work for delivery by the NGGO in 2025.

## 6. Priority Deliverable

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

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

## 3. Future developments

In 2024 the NGGO developed a 5-year implementation plan for the delivery of the Strategy. The NGGO has used this to inform the 10 priority deliverables of 2025. The 10 priority deliverables for 2025 are aligned to the Strategy's themes and the related areas of focus. These deliverables also include some projects of work not started or not completed in 2024.

### 2025 10 Priority Deliverables:

1. Complete a Bioinformatics and Genetic Data Infrastructure Roadmap
2. Complete the evaluation and service arrangement phases of the NGPS to facilitate service operationalisation
3. Finalise a proposal for next phase laboratory infrastructure development
4. Identify a process for the national procurement of genetic and genomic tests included in the Test Directory
5. Develop a Clinical Services Model
6. Develop a National Genomic Testing Consent Form



7. Develop the NGGO Communications Plan
8. Address skill gaps within the NGGO
9. Submit eligibility criteria business case for Clinical Scientist roles
10. Produce the first Strategic Genomics Workforce Plan

The NGGO will work to deliver on these 10 priorities throughout 2025.

## **4. The National Genetics and Genomics Office**

The purpose of the NGGO is to lead on the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland. The NGGO leadership team includes experts in clinical genetics and genomics, and genomic laboratory science.

### **National Leads**

Assistant National Director: Helen Shortt

NGGO National Clinical Director: Dr Emma McCann

NGGO National Clinical Laboratory Director: Dr Richard Hagan

The NGGO sits within the Office of the Chief Clinical Officer (CCO) in the HSE and reports through the CCO to the Chief Executive Officer to the Board of the HSE. The NGGO works closely with the Department of Health and other key stakeholders and clinical programmes in relation to the implementation of the Strategy. The NGGO reports on the implementation of the Strategy to an Implementation Steering Group that includes representation from patients.