



BY EMAIL ONLY

Deputy Colm Burke
Dáil Éireann
Leinster House
Kildare Street
Dublin 2

7th April 2025

PQ 13649/25-To ask the Minister for Health her plans to address the shortage of accredited genetic testing services for patients in Ireland; her views on public hospital laboratories not having the resources, equipment, or accreditation to meet the clinical genetic testing needs of patients and clinicians; how she proposes to address this; and if she will make a statement on the matter.

PQ13651/25-To ask the Minister for Health the guidance provided by the HSE to public consultant clinicians on where patient blood samples should be sent for genetic testing; what countries Irish patient blood samples are sent to for testing; the process for validating and approving these testing laboratories; if they send the samples to ISO 15189 accredited laboratories in Ireland; and if she will make a statement on the matter.

Dear Deputy Burke,

Many thanks for your representation.

The National Strategy for Accelerating Genetic and Genomic Medicine in Ireland was published in 2022. Many stakeholders including patients and staff contributed to its development. The strategy identifies several key strategic areas of focus that, when achieved, will transform existing services including genetic and genomic testing. The HSE's National Genetics and Genomics Office (NGGO) is working to achieve the following:

- Coordinating a national approach to genetics and genomics.
- Ensuring patient and public involvement (PPI) and partnerships
- Building the genetics and genomics workforce for the future
- Enhancing genetic and genomic clinical services
- Strengthening infrastructures to drive advances in genetics and genomics.

At the end of 2024, the NGGO published the first version of the National Genomic Test Directory for Rare and Inherited Disease (the Test Directory) as an advisory document. The Test Directory, developed by the NGGO through a series of collaborative workshops with clinical specialists and their laboratory colleagues, identifies the appropriate test, requesting specialties and clinical indications and criteria setting out which patients should be considered



for testing under that indication, and the requesting specialties is a list of the clinical specialties who would be expected to request the test.

Version 1 of the Test Directory includes tests for clinical indications from the specialties of Cardiology, Lipids, Metabolic, Mitochondrial, and Ophthalmology. Throughout 2025 and early 2026, the NGGO will work with clinicians and laboratory scientists to develop further versions of the Test Directory that will provide advice on genomic testing for a further number of clinical specialties.

At present individual hospitals make their own arrangements for samples they send for genomic testing. The NGGO estimates that upwards of 20,000 samples are sent abroad each year to in the region of 60-80 laboratories. A small amount of genomic testing is being carried out in the public health system at present.

The development of a centralised National Genomic Processing Service (NGPS) will enable the operationalisation of the Test Directory by ensuring that the right test is requested for the right patient at the right time in the right laboratory and will provide the NGGO with information on numbers of samples sent abroad for genomic testing.

In 2025, the NGGO, with support from the Department of Health, is advancing the development of the NGPS. The NGPS will support the use of the Test Directory and will be a hub for receipt of specimens from hospitals across Ireland to refer for genomic testing by laboratories abroad. The service will also coordinate the reporting of results back to the requesting hospitals and consultants.

In addition, during 2025, the NGGO will plan for a public procurement process for genomic tests associated with the clinical indications specified in the National Genomic Test Directory. In addition, the procurement process will require tendering laboratories to have a quality assurance system in place as a prerequisite.

The NGGO continues to work closely with the Department of Health on the implementation of the National Strategy including options for enhancing genomic testing capacity.

I hope this provides you with some assistance.

Yours sincerely

Sharon Hayden
General Manager
Office of the Chief Clinical Officer