National Genomic Test Directory

Rare and Inherited Disease
Version 1

Frequently Asked Questions

This FAQs document is valid for Version 1 of the National Genomic Test Directory: December 2024

Table of Contents

1. Introduction

The HSE established the National Genetics and Genomics Office (NGGO) in 2023 to implement the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland (the Strategy). The Strategy outlines the way forward for the genetics and genomics service in Ireland through the progression of work under 5 priority themes;

1. Coordinating a National Approach to Genetics and Genomics
2. Ensuring Patient and Public Involvement and Partnership
3. Building a Genetics and Genomics Workforce for the Future
4. Enhancing Genetic and Genomic Clinical Services
5. Strengthening Infrastructures to Drive Advances in Genetics and Genomics

The development of a National Genomic Test Directory was identified as a way forward in enhancing genetic and genomic clinical services by promoting evidence based, equitable, and timely access to genetic and genomic tests.

This frequently asked questions (FAQs) document is in reference to version 1 of the National Genomic Test Directory for Rare and Inherited Disease (the Test Directory), consisting of clinical indications for Cardiology, Lipids, Metabolic, Mitochondrial, and Ophthalmology. The Test Directory is in a process of phased development and the National Genomic Test Directory FAQs will be updated accordingly along with the Test Directory.

2. About the National Genomic Test Directory for Rare and Inherited Disease

The National Genomic Test Directory for Rare and Inherited Disease will set out the most appropriate genetic or 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. Version 1 of the Test Directory will be published for advisory purposes at the end of 2024. Version 1 will contain details of testing for a subset of specialties (Cardiology, Lipids, Metabolic, Mitochondrial, and Ophthalmology) with subsequent releases incorporating additional specialties. The development of a pre-analytical service in 2025 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 this will reduce clinical risk associated with genomic testing in Ireland.

2.1 The National Genomic Processing Service

The NGGO is planning for the delivery of a national centre which will be core to the delivery of a national genomic medicine service and the implementation of the Strategy. In the interim the NGGO will establish a National Genomic Processing Service (NGPS) for samples sent abroad for genetic and genomic testing. The development of a centralised NGPS for the pre-and post-analytical aspects of the Test Directory will deliver the first phase in the development of genomic laboratory services.

3 Frequently Asked Questions

3.1 What is the National Genomic Test Directory for Rare and Inherited Disease and where is it published?

The Test Directory defines all genomic tests for rare and inherited disease available within the scope of a national genomic medicine service. For each clinical indication, the Test Directory will also set out the eligibility criteria in relation to requesting the appropriate genomic test. The Test Directory is available on the NGGO webpage on the HSE website: National Genetics and Genomics - HSE.ie.

3.2 How is the content of the Test Directory developed?

The NGGO evaluated international examples of genomic test directories and identified the UK NHS England National Genomic Test Directory as the most suitable to use to develop a similar HSE National Genomic Test Directory for Rare and Inherited Disease.

Through a series of collaborative workshops with clinical specialists and their laboratory colleagues, the NGGO worked to identify the appropriate test, requesting specialties and clinical indications relevant to the population of Ireland and the Irish health care system. Version 1 of the Test Directory includes tests for clinical indications from the specialties of Cardiology, Lipids, Metabolic, Mitochondrial, and Ophthalmology.

The NGGO has also coordinated with colleagues in the National Cancer Control Programme (NCCP) who are developing the HSE National Genomic Test Directory for Cancer to ensure alignment of the directories and respective strategies. Information and access to the current NCCP Genomic Test Directory for Cancer is available on the NCCP webpage.

3.3 What does the implementation of a Test Directory mean for patients?

To meet patient need, clinical laboratories currently send many samples to laboratories outside of Ireland for genetic and genomic testing. The development of a National Genomic Processing Service (NGPS) for the pre- and post-analytical aspects of delivering the Test Directory for samples sent outside of Ireland is a fundamental deliverable of the Strategy. The implementation of the Test Directory and the NGPS will mitigate multiple patient, operational, and financial risks. The Test Directory will clarify which tests are appropriate for the patient for each clinical indication and eligibility criteria. The Test Directory and the services delivered through the NGPS will secure:

- Equity of access for patients to the full range of clinically appropriate genomic tests available in the Test Directory;
- Access to up to date and appropriate technology for the best diagnostic and clinical outcomes;
- A standardised testing approach for all patient samples;
- The ability to introduce new genomic tests in the future and keep pace with the latest research and evidence.

3.4 What tests can be ordered from the Test Directory?

The Test Directory identifies for each clinical indication the most appropriate test, testing methodology, and the patients who are eligible to access a test. The requesting specialties is a list of the clinical specialties who require access to the test for their patient cohort.

3.5 How can tests be ordered from the Test Directory?

Clinicians should follow local process to request genomic tests using the most appropriate clinical indication. All referrals for testing should be triaged by the applicable specialty pathway to ensure the most appropriate test is requested.

Clinicians requesting a genomic test can do so by;

- Requesting the appropriate clinical indication as provided in the Test Directory.
- If the clinician is aware that some of the constituent tests which are offered as part of the clinical indication are not needed, they should specify to the laboratory which constituent tests are required and which are not.

As much relevant clinical details as possible should be provided to allow the NGPS to efficiently ensure the test selection is appropriate and to aid interpretation of results by the analysing laboratory.

After selecting a genomic test, the NGPS Test Request Form will require completion by the requesting clinician. As a pre-requisite for submission of the sample to the NGPS, completion of the Minimum Data is required. The NGPS Test Request Form will be available on the NGGO webpage on the HSE website: National Genetics and Genomics - HSE.ie from the NGPS 'go-live' date.

In instances where the clinical indication described to the NGPS indicates that a constituent(s) part of a test is more suitable than the requested test, the constituent(s) part will be requested by the NGPS. Testing should be targeted at patients where a genetic or genomic diagnosis will guide management for the proband or family.

3.6 Who is eligible for testing?

The Test Directory sets out the clinical indications where there is clear scientific evidence for the value of genomic testing for patients. Each clinical indication included in the Test Directory has a set of testing criteria that sets out which patients are eligible for testing.

3.7 Can whole genome sequencing (WGS) be ordered as part of routine care?

Clinicians wishing to refer a patient for WGS should first discuss the case with a Consultant Clinical Geneticist, who will be able to advise on the most appropriate tests available.

3.8 Who can order tests from the Test Directory?

As many of the tests are only relevant for certain specialties e.g. Clinical Genetics or Cardiology, the Test Directory indicates which clinical specialties are able to order a specific genomic test.

3.9 Where is the testing carried out?

Genomic testing for patients is provided by specialised international laboratories, with the pre-and post-analytical phases being provided by the NGPS. The NGPS will route the sample to the laboratory identified as most suitable for the provision of the requested test. In the early phases of Test Directory development and operationalisation, the NGPS will accept requests to route samples to laboratories selected by requesting clinicians (the selected laboratory must meet the test requirements set out for the clinical indication in the Test Directory as assessed by the NGPS).

3.10 Who can I contact if I have a question about the current version of the Test Directory?

In most cases, the NGPS will be able to assist with any queries concerning the testing for the clinical indications in the Test Directory. In some cases, queries should be referred to a clinical geneticist who

may be able to provide advice. Guidance should be sought in accordance with the clinical urgency of the test in question. Any queries relating to the content, structure and format of the National Genomic Test Directory can be directed via email to the NGGO at Genomics.Office@hse.ie.

3.11 How will the results be returned to me?

The report issued in respect of each genomic test approved by the NGPS will be returned to the Requesting Entity according to local arrangements. This may include Requesting Entities receiving an electronic version of the report.

3.12 How is the testing in the Test Directory paid for?

Arrangements for payment will not change: The Requesting Entity (public hospital) will be invoiced directly by the external laboratory provider.

3.13 Can genomic tests not in the Test Directory be accessed?

Only genomic tests included in the Test Directory can be requested through the NGPS.

3.14 How and when will the Test Directory be updated?

The Test Directory will be reviewed and updated on an annual basis. To support the process to update the Test Directory, HSE NGGO will establish a Test Directory Clinical Advisory Group. The review process will ensure that the Test Directory is up to date with the latest advances in science and technology. Once a specialty has been added to the Test Directory, an application to propose updates to the Test Directory can be submitted to the NGGO Genomics.Office@hse.ie. The Test Directory Clinical Advisory Group will review the proposed amendments, make recommendations and agree the amendments to the Test Directory each year. Following approval by the Test Directory Clinical Advisory Group, HSE NGGO will publish an updated Test Directory in each year.

3.15 What type of amendments can be proposed to be made to the Test Directory?

Applications to propose updates to the Test Directory may seek to:

- Add new clinical indications to the Test Directory;
- Amend the eligibility criteria, requesting specialties, constituent tests, test targets or technologies for existing clinical indications;
- Amend the content of gene panels;
- Decommission a test where it is obsolete or no longer supported by clinical, scientific or economic evidence;
- Move a clinical indication or test to an alternative test method e.g. to WES

4 Contact Information

If you have any questions about the National Genomic Test Directory or genomic testing available through the National Genomic Processing Service, please contact the National Genetics & Genomics Office at Genomics.Office@hse.ie