

The National Rare Disease Strategy Ireland 2025-2030

J.F.A. Murphy.

The Minister for Health launched the National Rare Disease Strategy Ireland¹ in August 2025. It is a high-level document. The stated aims are to enhance the diagnosis, treatment, and support for patients suffering from a rare disease. It is hoped that the strategy will lead to an improvement in the quality of life, equitable access to care across the healthcare system, and the development of new therapies through high quality research. The funding being provided this year is 5.6 million Euro this year, rising to 8 million Euro in 2026.

This strategy document builds on the work being undertaken by the National Rare Disease Office (NRDO). The NRDO was established by the HSE in 2015. It is based at the Mater hospital and it acts as a co-ordination hub. It provides specific disease information and the clinical expertise that is available. It provides social care supports and also has a role in research and development. Advocacy is another important function of the office. It manages the Irish section of Orphanet, the European portal on rare diseases information and orphan drugs. A Model of Care for rare diseases² was subsequently published in 2019.

A rare disease is defined as a life-threatening or chronic debilitating disease that affects no more than 5 people per 10,000. The rarity means that medical awareness and scientific information is frequently deficient. There are 6,000 to 8,000 rare diseases. Although individually rare, they are collectively common. They affect 30 million Europeans, and in Ireland, 300,000 people live with a rare disease. The term used to describe affected individuals with a rare disease throughout the document is PLWRD (person living with a rare disease). PLWRDs regularly attend 4 or more specialist clinics for their care. The care is divided between specialist services and community services. The clinical and support services engagement is frequently more than 10 times annually. 70% of rare diseases are exclusively paediatric in onset. In the year 2000, 4.2% of children were diagnosed with a rare disease by age 17 years, the mortality rate being 12%.

There are frequent delays in reaching a rare disease diagnosis. Over 70% of PLWRDs have seen 3 or more consultants from different specialties on their journey to the eventual diagnosis. The average is a 6.1 years interval before a diagnosis is made in Ireland. Initially, 83% received a mis-diagnosis and 49% were administered inappropriate treatment. The strategy recommends a number of measures to address these diagnostic deficiencies. The screening, and diagnostics need to be expanded. Rare disease modules should be incorporated into undergraduate and postgraduate curricula. The teaching programmes should include clinical practice, diagnostics, pathology, genetics, and the challenges facing the patient in terms of daily living, psychological and mental health wellbeing. Workshops on rare

diseases should be made available as part of CPD. The European reference networks (ERNs) play an important role in the promotion of education in the diagnosis and treatment of patients with rare diseases. It has developed online learning platforms, e-learning courses, and webinars. There are ERN Academies which provide in-depth training on various aspects of rare disease management.

Population screening plays an important role in the early diagnosis and prompt treatment for rare diseases. The governance of screening and implementation has been enhanced by the establishment by NSAC (national screening advisory committee) in 2019. One of the major pillars is the national newborn blood spot programme (NNBSP) that is undertaken on all newborn infants. Currently it screens for 9 diseases with 2 further being added for SMA and non-ADA SCIDS. The programme has the capacity to screen for 50 diseases and more will be added over time. The other newborn screening programme is the national universal newborn hearing screening programme (NUNHSP). The National Genetics and Genomics office established in 2023 is delivering improved access to genetics. For certain disorders prenatal diagnosis is possible.

Only 5% of rare diseases have a specific treatment available for affected patients. The pharmaceutical industry has been discouraged by the high costs and low yields. The Orphan Medicinal Product (OMP) initiative has been a positive development. The committee for the OMPs is the EMA committee responsible for recommending orphan designation of medicines for rare diseases. The committee was established in year 2000. A range of incentives are being offered by governments including a 10-year market exclusivity and research grant funding. To date over 200 orphan medicines have been authorised for patients with rare diseases. Two recent breakthrough pharmaceutical products have been Zolgensma for spinal muscle atrophy (2021), and Libmeldy for metachromatic leukodystrophy (2024).

Rare disease research is critical for the understanding and treatment of rare diseases. Access to high quality clinical trials is important for PLWRDs. One of the barriers to rare disease research is limited time available for clinicians to undertake research. The document promotes the provision of protected time for undertaking research. It also supports the provision of funding and infrastructure support. A further proposal is the expansion of research fellowships and placements to enable the development of a workforce skilled in both clinical care and research.

The HRB has invested 18.8 million in rare disease research as of August 2025 supporting collaborative national and international initiatives. In January 2024, the European Commission provided €56m for rare disease research. The All-Ireland rare disease interdisciplinary research network (RAiN) was established in 2023. It has established a research partnership between UCD and Queen's University, Belfast.

The strategy addresses the importance of public engagement. The general public have never heard of most of the rare diseases. This leads to misunderstanding and isolation for PLWRDs. Awareness campaigns will help to inform communities about the nature and challenges for a person living with a rare disease. This is important from a rights perspective and will help to promote equity and inclusion. The recommendation is the development of a rare disease communication and awareness plan to highlight the issues around rare diseases with the general public, healthcare professionals, and policy makers. This process needs to be progressed in collaboration with the PLWRDs to ensure that it best fits their specific needs.

The strategy is further step on the journey in the provision of timely diagnosis and effective treatment and care of persons suffering from a rare disease

JFA Murphy
Editor

References:

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2. Model of Care for Rare Diseases: The national clinical programme for rare diseases. July 2019: <https://www.hse.ie/eng/services/list/5/rarediseases/rare-disease-publications/model-of-care-for-rare-diseases.pdf>.