Rare Diseases Ireland

MANIFESTO FOR RARE DISEASES

300,000 people living with rare diseases in Ireland, as well as their families and caregivers, call for an Ireland where they can have longer and better lives and achieve their full potential, in a society that values their wellbeing and leaves no one behind.

1 in 2,000
people, or less, affected by any rare disease

300,000
people live with rare diseases in Ireland

6,000+
number of distinct rare diseases

72%
rare diseases are genetic in origin

In Ireland 1 in 17 people are living with rare diseases that are typically chronic, progressive, degenerative, disabling and frequently life-threatening. Rare diseases have a profound impact on the daily lives of affected individuals and their families.

The next government must take steps to build on the progress that is being made with development of a new National Rare Disease Strategy for Ireland. Delivering on the ambitions of the strategy will improve the lives of tens-of-thousands of people living with rare diseases by delivering faster diagnosis (with improved counting, coding and healthcare planning), better coordination of care, improved access to health and social care, including mental health care, and improved access to specialist care and treatment, including care and treatment available internationally.

This manifesto sets out our priorities for the next five years. We are calling for people living with rare diseases to be a priority group for the next Government, and all other relevant policymakers. Strong political leadership is required to set clear expectations, ensure that all parts of the system work together, and provide the resources needed to deliver change.

The following recommendations are key to building an equitable inclusive Ireland.

LEADERSHIP, GOVERNANCE AND ACCOUNTABILITY ACROSS HSE AND DEPARTMENT OF HEALTH

EARLIER, FASTER AND MORE ACCURATE DIAGNOSIS

INTEGRATED COORDINATED CARE PATHWAYS WITHIN AND BETWEEN HEALTH REGIONS AND EXPERT CENTRES

TIMELY EQUITABLE ACCESS TO INNOVATIVE TREATMENTS AND TOOLS

LEADERSHIP, GOVERNANCE AND ACCOUNTABILITY ACROSS HSE AND DEPARTMENT OF HEALTH

Rare and complex diseases can be difficult to diagnose, challenging to treat and complicated to care for. The next government must put in place and invest in the structures at Department of Health (DoH) and Health Service Executive (HSE) to build on the progress that is being made with development of a new National Rare Disease Strategy for Ireland.

Genuine partnership between Government, DoH, HSE, healthcare professionals, people living with rare diseases, families, caregivers, patient organisations, researchers and industry is required to unblock barriers that are holding back progress for those living with rare diseases.

- **Establish a dedicated cross-sectoral National Rare Disease Strategy Oversight Group with appropriate leadership, authority, and accountability to provide oversight and governance ensuring the effective funding, implementation and audit review of the National Strategy.**
- **Develop and resource a rare disease service at national level in the HSE and DOH to drive implementation of the National Strategy.**
- **Support and enable effective patient partnership and representation of people living with rare diseases across all levels of healthcare decision-making.**

EARLIER, FASTER AND MORE ACCURATE DIAGNOSIS

The search for an accurate diagnosis is very often a prolonged “odyssey” for people living with rare diseases. Lack of awareness of rare diseases among healthcare professionals, limited capacity in screening and genetic and genomic testing services and inadequate referral pathways give rise to a six-year average delay in diagnosis. Delayed diagnosis leads to deterioration in both physical and mental health, as well as financial and social distress, for both the person living with a rare disease and their family. Diagnosis is the first step towards improved health and wellbeing.

- **Sustainably invest in continuous expansion of the newborn screening ‘heel prick’ test program at HSE and the National Screening Advisory Committee. At minimum double (from 9 to the European average of 18) the number of rare diseases screened at birth.**
- **Establish and integrate pathways that ensure timely and equitable access to genetic and genomic services for all across the country, including at GP level.**
- **Invest in laboratory and data infrastructure to deliver on National Genetics & Genomics Strategy.**

INTEGRATED COORDINATED CARE PATHWAYS WITHIN AND BETWEEN HEALTH REGIONS AND EXPERT CENTRES

Rare diseases are not only rare due to ‘small’ numbers affected by them, but also due to the scarcity of healthcare experts who can treat them. This has created a 'lottery' in which some people living with rare diseases can access experts and benefit from their knowledge, experience and access to research and trials, but others are unable to access this same level of expert care and treatment. New health regions must be designed and resourced to improve health outcomes for people living with rare diseases and eliminate these inherent inequalities.

- **Commission and implement HSE approved national rare disease care pathways across the health regions. Demonstrate ‘the right care in the right place at the right time’ principles for coordinated complex care.**
- **Establish and integrate care pathways that deliver timely equitable access to the right expert centre for all people living with rare diseases, with/without a diagnosis.**
- **Invest in sustainable European Reference Network (ERN) integration into the healthcare system to enable access to the knowledge, experience, research and trials, and highly specialised care of international experts.**

TIMELY EQUITABLE ACCESS TO INNOVATIVE TREATMENTS AND TOOLS

In recent years, scientific developments have deepened understanding of rare diseases and led to advances that offer hope of improved outcomes. Innovations such as genomic testing, advanced therapy medicinal products (ATMPs), also known as cell and gene therapies, digital care support tools and technologies such as artificial intelligence (AI) coupled with digital health records could help address the myriad inequalities that people living with rare diseases face.

In spite of Ireland’s leadership position in industries such as medtech, pharma, biotech and digital, our healthcare system is ill prepared to adopt these innovations.

- **Appoint a cross-sectoral group to develop and implement a strategy that ensures innovative medicines are available to people living with rare diseases within one year of receiving European Medicines Agency (EMA) marketing authorisation, with appropriate safe delivery and outcome evaluation, including outcome registries.**
- **Establish multi-stakeholder ‘National ATMP Clinical Network Programme’ to accelerate development of infrastructure and personnel to provide access to cell and gene therapies in Irish expert centres.**
- **Develop ethical framework to inform adoption and/or expansion of innovations such as AI-driven diagnostics and genomic newborn screening.**

5 years
to add one disease to 'heel-prick' test

75%
Europe screen for more diseases in 'heel-prick' test

58% PLWRD
never referred to an expert centre

54 of 63
EMA authorised (2019-22) meds not available 01/24

39 ATMP
submissions expected at EMA over 2 yrs to 03/25

31% PLWRD
no medical expert in Ireland for their rare disease

€0
invested over 7 yrs in ERN integration and infrastructure

6.1 years
average time to confirmed rare disease diagnosis

0 of 11
national rare disease care pathways commissioned

60% PLWRD
misdiagnosed with another physical disease

Rare diseases in children living in Ireland

4.2% children diagnosed with a rare disease **by age 17**

~2,000 (3.7%) teens with a life-long rare disease **transition each year** to adult care

11.8% mortality by age 18 in those living with rare diseases

58.6% deaths in those **<15 years old** have an underlying rare disease

Children with rare diseases use **25 times** more **hospital bed-days** as compared to their peers

51.9% children's **hospital bed-days** are used by the **4.2%** living with a rare disease

Most rare diseases are life-long, chronic and disabling, and sometimes progressive and life-threatening.

Gunne E, Lambert DM, Ward AJ, Murphy DN, Treacy EP, Lynch SA. An estimate of the cumulative paediatric prevalence of rare diseases in Ireland and comment on the literature. *Eur J Hum Genet.* **2022 Nov;30(11):1211-1215**

Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, Murphy D, Le Cam Y, Rath A. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* **2020 Feb;28(2):165-173**.

EFPIA Patients W.A.I.T. Indicator 2023 Survey **2025 Jun**

Rare Reality – Living with a rare disease in Ireland – Healthcare experiences **2022 Jan**