Rare Diseases Ireland

Ireland's EU Presidency

Spotlight on Rare Disease Research & Innovation

Oireachtas All Party Group on Rare Diseases

July 1st, 2025

What are rare diseases?

What is a RARE DISEASE?
A disease that affects
fewer than 1 in 2,000 people

1 in 17 people
live with a
rare disease

300,000
in Ireland

6,000+ distinct rare diseases

72% genetic origin

70% with first symptoms appearing in childhood

5,100+ ultra-rare diseases each one affecting **< 5** people in Ireland

Rare Disease burden on children in Ireland

IRISH DATA

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

Today's challenges for people living with rare diseases

- **Diagnostic delays** – on average 6 years in Ireland
- **Limited treatment options** – only ~6% of RDs have approved treatment
- Navigate **fragmented and inadequate care** systems
- **Lack of awareness** among public, policy makers and healthcare professionals
- **Lack of expertise** among healthcare professionals – 31% patients have no access to medical expertise in Ireland
- **Inadequate investment** in R&D and innovation
- Discrimination and isolation

The future is brighter for people living with rare diseases

- **Patient centric innovation** – Patient centred outcome measures as endpoints
- **AI-powered tools** - diagnosis, research, drug discovery
- **Advanced (cell and gene) therapies** - personalised medicine
- **Cross border healthcare** - European Reference Networks
- **Decentralized clinical trials** – patient access to trials (regardless of geography)
- **European health data space** - Pan-European rare disease registries
- **Revision EU regulations & legislation** - Orphan drug incentives, joint procurement

Rare Diseases in past EU presidencies

- **French Presidency - 2022 - Conference:** Care and innovation pathways for a European rare diseases policy
- **Czech Presidency - 2022 - Conference:** Expert Conference on Rare Diseases
- **Hungarian Presidency - 2024 - Conference:** For an EU commitment to tackling rare diseases
- **Polish Presidency - 2025 - Conference:** Towards an EU Action Plan on Rare Diseases

Our ask of you

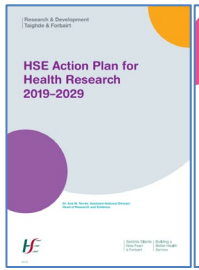
Press government to **host a Rare Disease Conference:**

Embedding research and innovation as standard for all rare disease care

Consortium of patient organisations working together

Policy landscape

- **UN** (2021) **& World Health Assembly** (2025) **Resolutions**
- **Rare 2030 Foresight Study** (2021)
- **EU4Health JARDIN Joint Action** (2024-2026)
- **Ireland's National Rare Disease Strategy** (to be published)
- **National and international Research & Innovation policies**



ERDERA
European Rare Diseases Research Alliance

JARDIN

UNITED NATIONS

World Health Organization

Rare 2030
Foresight in Rare Disease Policy

Rare Diseases Ireland

Rare Disease Conference in Ireland's EU presidency

- **Possible themes:**
  - The **Target 5000** story - from patient-led genotyping services to registries, gene therapies and first-in-man trials and bench research
  - **Advanced therapies** - from UCD's NIBRT to clinical trails at St James' Clinical Research Facility to gene therapy as routine care, screening at birth and potential disease elimination
  - **European Refence Networks** - from sharing skills and expertise across borders to diagnosis, treatment and care to training future experts and sharing data across borders
  - **AI digital tools** - from diagnosis to treatment and care, to health service planning, to research & development and digital twins

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