ERDERA

European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for Ireland

National Mirror Group

Ireland has a National Mirror Group (NMG), which was launched on 3rd September 2024. The NMG is coordinated by Ireland's Health Research Board. This NMG has overseen the collection of data from Ireland via a data contributing committee. Contributors are listed at the end of this report.

Definition of a Rare Disease

Ireland adopts the formal European Union (EU) definition of a rare disease (i.e. those with a prevalence of no more than 5 patients per 10,000 persons. This definition is laid down in Regulation EC no. 141/2000 on Orphan Medicinal Products, Directive 2011/24/EU on Cross Border Healthcare as well as in the Council Recommendation on an action in the field of rare diseases of 8 June 2009.). Ireland's National Strategy espouses this definition. There are also references within the strategy to the approximate number of people living with a rare disease in Ireland, which is over 300,000.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Ireland has a live, time bound, National Strategy for rare disease. Ireland's 'National Rare Disease Strategy' can viewed here. The strategy covers a five-year period, between 2025-2030. Previously, there was a National Rare Disease Plan for Ireland between 2014-2018.

Elaboration and Adoption/How was the original Strategy elaborated?

The first 'National Rare Disease Plan for Ireland 2014-2018' contained 46 recommendations focused on improving diagnosis, identifying centres of excellence, and establishing dedicated rare disease pathways. Whilst significant progress was made through the establishment of the National Rare Diseases Office in the HSE (Health Service Executive in Ireland), including publication of a Model of Care for Rare Diseases and the inclusion of Ireland in 18 European Reference Networks, there were other areas where implementation did not progress as may have been envisaged, or where new technologies and advancements have overtaken these recommendations. Ireland renewed focus on

Co-funded by
the European Union

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.
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rare disease and updated their vision and aims to ensure that a new strategy for rare disease meets the needs of people living with rare diseases, over the next five years.

Ireland's new 'National Rare Disease Strategy' was officially launched by the Minister for Health, on 27 August 2025. It is not enshrined in laws or regulations but elements enabling the strategy, such as digital health records, are part of other legislation. In the development of this Strategy, the Department of Health commissioned the Health Information and Quality Authority (HIQA) to conduct a review of National Rare Disease Plans and Strategies in 12 other countries. The review focused on identifying the main themes and aims in national rare disease strategies, the structures used to support strategy implementation, and how these compare to Ireland's previous National Rare Disease Plan. The main themes identified across all 12 countries included screening and diagnosis, access to healthcare and coordination of services, rare disease research, and representation of people with rare diseases.

In April 2023, a dedicated policy unit was established within the Office of the Chief Medical Officer (CMO) to commence work and to engage with stakeholders across the Department of Health, the HSE and advocacy groups, to develop a successor to the 'National Rare Disease Plan 2014-2018'. In December 2023, the Chief Medical Officer established the National Rare Disease Steering Group. The Steering Group's Terms of Reference were as follows:

1. To develop a National Rare Disease Strategy and associated action plan for the consideration of the Chief Medical Officer and Minister for Health. The National Rare Disease Strategy will set out the vision for Rare Disease services in Ireland and outline the actions required to achieve this.
2. Consider how best to improve access to rare disease services and consider how best to ensure appropriate access for people living with a rare disease in Ireland, with a particular focus on integrated care.
3. Have regard for Sláintecare (improvement plan and strategy for reforming Ireland's health and social care system), the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland, the European Health Data Space, the Health Information Bill, and other relevant European and international rare disease policies.
4. Consider how best to integrate European Reference Networks into the national health system.
5. Undertake a robust consultation process to inform the development of the new plan. This process will include, as appropriate, people living with rare diseases, stakeholder engagement and public consultation.
6. Identify the core practical requirements of a Rare Disease Registry Framework, with due regard for existing registries, the integration of ORPHAcodes, ERN reporting requirements and service development needs.
7. Consider how to best promote rare disease awareness, including education among healthcare professionals, policy makers, and the public.
8. Consider how to best promote and support participation in national and international research.
9. Consider the governance and funding implication of any of the recommendations arising out of the new National Rare Disease Strategy.

The National Rare Disease Steering Group was comprised of patient representatives, patient advocates, clinicians, researchers, and representatives from the National Rare Diseases Office, National Genetics and Genomics Office, European Reference Networks, Health Research Board, HSE Child Health Public Heath, the office of the Chief Clinical Officer and the National Clinical Programme for People with Disability. The group was led by an independent Chair with secretariat support from the Department of Health. The National Rare Disease Steering Group met regularly over the course of developing this Strategy, which included several workshop meetings to achieve consensus on specific questions. The Steering Group took a broad consultative approach which included the establishment of a National Rare Disease Patient Forum, a large public consultation, and direct engagements with key stakeholders. The Steering Group also invited several contributors to present across key areas including:

- Models of Care and Transitional Care
- Screening
- Genetics and Genomic Medicine
- European Reference Networks
- Orphan Medicinal Products
- Medical Card Eligibility
- Health Information Systems & Registries
- Research and Clinical Trials

Is there funding for the strategy? How is it Implemented and/or Monitored or Evaluated?

Ireland’s National Strategy for Rare Disease has no funding or resources committed to the strategy itself but there are funds dedicated to delivering actions within the strategy.

A different group has been initiated specifically to oversee the implementation of Ireland’s National Strategy for Rare Disease. The Implementation Oversight Group (IOG) has not formally begun but it will review the roll-out and timebound performance of recommendations being implemented as part of the National Strategy for Rare Disease and will feedback on performance to the Department of Health. The following stakeholder types will be included in this group:

- Patients/people with lived experience of a rare condition
- Ireland’s National Alliance of rare disease patient Organisations
- Health Ministry/Competent National Authority in charge of Health or Care
- Research Ministry/ Competent National Authority in charge of Research
- Research funders
- Researchers/clinicians from rare disease centres
- ERN coordinators or representatives

Formal monitoring/evaluation will take place of Ireland’s National Strategy and this will be carried out by the IOG. Each recommendation within the strategy has a lead agency responsible for it with indicative timelines. The IOG will engage with the agencies responsible for updates and roll-out of changes, collating challenges or delays and feeding this back to the Department of Health for co-ordination and governance. When operational, the IOG will monitor the strategy and meet regularly to review timelines and delivery.

Research in National Plans or national research strategies relevant to rare disease

Rare disease research is specifically addressed within Ireland's National Strategy for Rare Disease. There is a section dedicated to rare disease research, which highlights the importance of research and collaboration, particularly in relation to international collaboration. The National Strategy includes specific mention of international initiatives including ERDERA, European Joint Programme for Rare Disease (EJP RD), JARDIN, 1+ Million Genomes project (1+MG), EP PerMed (European Partnership for Personalised Medicine) and ICPerMed (International Consortium for Personalised Medicine), European Reference Networks (ERN), ERAnet and the European Clinical Research Infrastructure Network (ECRIN).

Topics relating to rare disease research included in the National Strategy are:

- International/transnational RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Biobanks/biosample catalogues for rare diseases
- Ontologies, codification or data standardisation
- Diagnostics research ('solving unknown conditions')
- Basic research (e.g. cell lines, animal models etc)
- Clinical/Translational research
- Sociological (e.g. Quality-of-Life-related) or socio-economic research

Ireland's National Strategy includes the following recommendations relating to rare disease research:

- Establish a National Rare Diseases Research Group, including people living with a rare disease (PLWRD) as partners, to provide a structured, coordinated, multi-actor and strategic approach to ensuring more high-quality rare disease research in Ireland. This group aims to build research capacity and capability from basic research to conducting clinical research (including clinical trials) within the health services (including ERN hospital sites) and universities.
- Increase the opportunities for PLWRD to access, as potential participants, in high-quality clinical trials and healthcare interventions by supporting the Rare Disease Clinical Trial Network and through Ireland's participation in ERDERA.
- The development of a rare disease registry to allow suitable participants to be identified and offered the opportunity to participate in research and trials relevant to them.
- Increase opportunities for rare disease specific funded research calls targeting both indigenous research and greater participation in international consortia.
- Ensure healthcare professionals are supported and encouraged to pursue research in rare diseases and provided with the dedicated time to do so.

There are no financial commitments outlined within the strategy, specifically for research. However, government agencies currently involved with supporting rare disease research, including the Health Research Board, will continue to include elements of the rare disease strategy for research within their financial estimates, submitted as part of business planning to the Department of Health on an annual basis.

Within the terms of Reference for the steering group that developed Ireland's National Strategy, it is stated that the strategy will synergise with other national strategies and policies. The Steering Group drafting the strategy involved key stakeholders to ensure clinical and research alignment with wider developments nationally, in order to compliment other activities and strategies, as well as international developments where possible.

Select Achievements of the National Plan

The first National Rare Disease Plan for Ireland (2014-2018) contained 46 recommendations focused on improving diagnosis, identifying centres of excellence, and establishing dedicated rare disease pathways. The key achievements thus far are:

1. Establishment of the National Rare Diseases Office in the HSE
2. Publication of a Model of Care for Rare Diseases
3. Ireland joining 18 out of 24 European Reference Networks
4. Establishment of Rare Disease Technology Review Committee to support decision making around reimbursement of some high value orphan medicines.

Rare Disease Research Programmes and Funding

There are specific and regular research programme/funding calls dedicated to rare disease research in Ireland. These programmes/funding calls are funded by public funds.

The Health Research Board (HRB) is the national funding agency for health and social care research in Ireland. It has funded the following rare disease initiatives:

- Rare Disease Catalyst Consortium

  This consortium brings all relevant stakeholders together under an investment worth €3 million across three years. The consortium connects Irish-based researchers, clinicians, and patient advocates with international networks to strengthen rare disease research. Activities include enabling the use of European Reference Network disease registries, rare disease researcher training, increasing the influence of rare disease patients in research and developing research pathways for undiagnosed patients under the upcoming ERDERA partnership programme.

- Rare Disease Clinical Trials Network (RDCTN)

  The RDCTN acts as a collaborative hub for rare disease trials, with the HRB investing €1 million over five years to increase opportunities for patients to access high-quality clinical trials. The HRB funded initiative addresses challenges of conducting rare disease clinical trials in Ireland by supporting education and training, regulatory frameworks, public and patient involvement and industry engagement.

- Joint Transnational Calls

  The HRB participates in ERDERA and co-funds participation in Joint Transnational Calls for rare diseases.

- Joint funding with charities

  The HRB partners with Health Research Charities Ireland (HRCI) to fund innovative research in many areas relevant to HRCI members and has proven to be an important funding avenue for rare disease research, accounting for approximately €7m of the HRB spend on rare disease in the last 10 years.

- The HRB also supports a selection of research career awards, EU research projects and the Irish Platform for Patient Organisations, Science and Industry (IPPOSI).

In total, the HRB has invested over €18.8 million over the last decade in rare disease research funding programmes/calls. There is policy in place to ensure that a portion of the national research budget specifically is allocated to rare disease research.

Rare Disease Registration and Biobanking

Ireland does not currently have national or regional registries for rare disease. However, Ireland's National Strategy for rare disease includes a recommendation to establish a national rare disease registry, underpinned by legislation, as necessary. The registry will enable improved planning, coordination of care and monitoring of outcomes for PLWRD by supporting service and workforce planning, epidemiology and pharmacoeconomic data. Additionally, an appropriately robust system of rare disease data reporting is required across health and social care sectors. A project team will be put in place to scope the technical and practical design elements of a rare disease registry, and to develop a detailed specification plan for a national rare disease registry.

There are disease-specific registries/registries for groups of rare conditions in Ireland. These registries are often established by charities, supported by the community that have lived experience of the disease and/or health care professionals that treat the condition.

Ireland does not have a national biobank specifically for rare disease biosamples but individual biobanks exist for rare disease samples.

Organisation of Rare Disease Care

Centres of Expertise

There is a policy in place in Ireland to designate Centre's of Expertise for rare disease. There are centres in place for groups of rare disease and individual rare diseases. In total, there are 40 Centres of Expertise in Ireland. Ireland uses EUCERD (European Union Committee of Experts on Rare Diseases) criteria for designating centres of expertise. The following requirements are included within the criteria:

- Contribute to state-of-the-art research
- Bring together, or coordinate, multidisciplinary specialised medical and paramedical expertise such as physiotherapists, speech therapists, dieticians, genetic counsellors, etc
- Bring together, or coordinate, or liaise with social services or social workers, in order to serve the specific medical, rehabilitation and palliative needs of rare diseases patients
- Collaborate with patient organisations to bring in the patients' perspective
- Provide education and training to healthcare professionals from all disciplines, including paramedical specialists and non-healthcare professionals (such as school teachers, personal/homecare facilitators) whenever possible
- Contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals
- Participate in data collection for clinical research and public health purposes

ERNs

Ireland is involved in multiple European Reference Networks. For the latest details on participating HCPs, click here.

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Newborn Screening

Nine conditions are included within Ireland's Newborn Screening programme. An additional two conditions have been approved for screening but implementation is pending. Further details are available here.

Ireland is involved in international initiatives about newborn screening decisions such as Screen4Care.

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Diagnostics

The National Genetics and Genomics Office (NGGO) was established in 2023 to drive the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland and improve Ireland's genetic and genomic medicine services. More information is available here. The NGGO is responsible for coordinating a national approach to genetics and genomics, supporting the development of a skilled genetic and genomic workforce, enhancing clinical services for patients and families, and strengthening laboratory and data infrastructure to drive advances in genetic and genomic medicine services.

A large cohort of clinical genetic testing is currently sent abroad. To address this, a National Genomic Processing Service for co-ordination nationally has been established. Public genetic testing is reimbursed at present. This is an ongoing area of activity in Ireland where services are being established and formalised at a national level. The NGGO are overseeing the 'National Strategy for Accelerating Genetic and Genomic Medicine in Ireland' which includes streamlining of clinical services. Ireland has recently launched the 'National Genomic Test Directory for Rare and Inherited Disease'.

Ireland is involved in initiatives to address undiagnosed patients/people with currently undiagnosable conditions. The HRB funded the Rare Disease Catalyst Grant (RDCAT-2023-001), which has a dedicated work package to integrate with wider efforts in ERDERA focused on 'unsolved patients' cases'. RDCat specifically enables a research pathway to be developed for those without a clinical diagnosis in Ireland. The pilot initiative involves repatriation and reanalysis of data from patients in Ireland, from international clinical laboratories. It demonstrates what will be needed for a future clinical pathway and enables Ireland to contribute to wider EU efforts and the EU4Health Joint Action Integration grant.

Ireland has a policy in place to ensure national providers provide genetic counselling for patients with a suspected or confirmed rare disease.

National Alliances of Rare Disease Patient Organisations

Rare Diseases Ireland (RDI) is Ireland's National Alliance of rare disease patient organisations. Further information about RDI is available here.

RDI were involved in the steering committee which established Ireland's strategy for rare disease, including the research section of the strategy. RDI will be involved in setting Ireland's strategy for research in the future through the National Rare Diseases Research Group, when up and running. RDI are part of the applicant team of existing research grants funded by the HRB such as RDCat, which funds a post within RDI.

Information Resources for Rare Disease

National Orphanet Engagement

Ireland has an operational, national, Orphanet team, which is hosted by the HSE's National Rare Diseases Office. The Orphanet team is funded through national funding. Orphanet Ireland registers Irish data and information into Orphanet. This includes information on Irish centres of expertise, patient organisations, national registries, national biobanks, national research and national clinical trials are registered, if known, and meet the Orphanet criteria.

Helplines

Ireland has a national helpline for rare disease that is publicly funded and available for anyone to use. The National Rare Disease Information Service is managed by the HSE National Rare Diseases Office (NRDO). The Information Service is led by NRDO Information Scientists who provide evidenced base information on rare disease and signposts to rare disease support and resources. The information line runs through a designated email address and queries are sent to NRDO Information Scientists, who use validated resources e.g. Orphanet, ERN websites, HSE website, and liaise with clinical experts to ensure responses are accurate and current. Written responses are provided within 14 working days.

Training and Education

Ireland has a number of rare disease training activities available. The following topics are included within these training activities:

- Diagnostics
- Awareness of Rare Disease
- Data management
- Data quality
- FAIR data
- Standards and quality of genetics/genomics data in clinical practice and laboratories
- Registries
- Biobanks
- PPIE
- Clinical research

The NRDO has developed an e-learning, Rare Disease Education Programme for all healthcare professionals in Ireland. They also deliver an annual training and education session to undergraduate medical students at University College Dublin (UCD). The NRDO Information Scientist for Orphanet delivers training sessions to key stakeholders including clinicians and researchers on Orphanet and Orphacodes. All training delivered by the NRDO is free of charge.

The NGGO in partnership with Royal College of Surgeons in Ireland (RCSI) and The Mater Misericordiae University Hospital (MMUH) developed a training course for healthcare professionals named the Fundamentals of Genetic Testing. Ireland’s Rare Disease Clinical Trial Network (RDCTN) host training initiatives and the RDCat are developing training as part of their grant, including offerings for higher education.

The majority of training is made available free of charge with costs covered by wider grants and all training is in English.

Orphan Medicinal Products (OMPs)

There is no easy data source to provide information on OMPs available in Ireland. Recently, work was carried out by Rare Diseases Ireland on OMPs (as defined by EMA) availability in Ireland. The findings are summarised in the table below:

Year EMA Authorisation	Number authorised	Number withdrawn from market	Number reimbursed by HSE	% eligible OMPs reimbursed by HSE
2019	5	1	1	25% (1 of 4)
2020	20	3	7	41% (7 of 17)
2021	17	2	6	40% (6 of 15)
2022	24	2	4	18% (4 of 22)
2023	10	0	1	10% (1 of 10)

The above data was extracted from European Medicines Agency & National Centre for Pharmacoeconomics websites on 9/10/2025.

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Disclaimer: The data collection activities which enabled this 2025 national report were supported by the ERDERA. ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595. Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them, nor should this document be viewed as an official national 'position'.

Co-funded by the European Union

The Data Contributing Committee of Ireland, which provided this 2025 data (correct as of the end of November 2025) in the context of the Resource on the State of the Art of Rare Disease Activities, is composed of the following individuals:

- Gavin Lawler, Health Research Board (HRB)
- Oonagh Ward, Health Research Board (HRB)
- Atif Awan, Health Service Executive (HSE) National Rare Diseases Office (NRDO)
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