# National Rare Disease Strategy 2025-2030

An Roinn Sláinte
Department of Health

# Contents

# Ministerial Foreword

**Minister for Health**
Jennifer Carroll MacNeill, TD

I am honoured to present Ireland's new National Rare Disease Strategy and reiterate the Government's commitment to improving the lives of people living with rare diseases and their circle of support. Building on the foundation of the previous *National Rare Diseases Plan for Ireland 2014-2018*, this Strategy reflects the progress we have made while recognising the work still ahead.

Since the publication of the previous Plan, steps have been taken to address the challenges faced by people living with rare diseases. The establishment of the National Rare Diseases Office marked a pivotal step, providing essential support, information, and advocacy for those navigating the complexities of rare diseases. Yet, we know that for many individuals there remain significant challenges across many areas. Unprecedented funding has also been provided for rare diseases with €6.5m allocated for this year (rising to €8m in 2026). This funding is being used to lay the strong foundations on which we can quickly and proactively drive the implementation of this Strategy's recommendations and deliver real change for the estimated 300,000 people living with a rare disease, their families and circle of support. While this investment is a significant initial step, it is recognised that additional funding through the annual service planning process will be required to fully achieve the goals and aims of this Strategy.

Rare diseases often present complex and multifaceted challenges. Individuals and their families face a diagnostic odyssey, often taking several years to get an accurate and definitive diagnosis, they face difficulties accessing timely and coordinated care, compounded by the rarity and complexity of their conditions. This Strategy reaffirms our commitment to transforming services in alignment with the core principles of Sláintecare – of delivering the right care at the right time as close to home as possible and ensuring that care is equitable, inclusive, and person-centred.

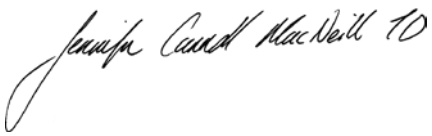
At the heart of this Strategy is a vision to ensure that all people living with a rare disease have access to equitable, inclusive, safe and cross-sectoral care throughout their life journey that will enable them to reach their full potential and to live their best lives.

Through collaboration across health, social care, research, and community sectors, we aim to create a system that responds to the needs of people with rare diseases advancing how we diagnose, treat and care for them. This includes leveraging advancements in genomics and precision medicine to enhance pathways to diagnosis, ensuring, where possible, care is delivered in the community through integrated care pathways that include links to multidisciplinary and interdisciplinary teams, health and social care professionals, and specialists in line with each persons need. We must also continue to support research to improve access to innovative treatments and specialist care to improve outcomes.

The implementation of this Strategy should represent a call to action for all stakeholders—healthcare providers, policymakers, researchers, industry, community organisations and most importantly people living with a rare disease and their families —to work together to make Ireland a leader in rare disease care. I extend my deepest gratitude to all who contributed to its development, including our rare disease partners, whose voices are central to this work.

Together, we can ensure that every person living with a rare disease in Ireland can live a life of dignity, inclusion, and hope.

**Minister for Health**
Jennifer Carroll MacNeill, TD



# Foreword
## by Prof. Cecily Kelleher, Chair

**Chair**
Prof. Cecily Kelleher

It was my privilege to chair the Steering Group that worked on this National Rare Disease Strategy for 2025-2030. Over an 18-month period since December 2023 we have worked to produce this Strategy that one hopes will make a lasting difference for all people in Ireland living with a rare disease, their families and circle of support, now and into the future.

This is an evidence-based report which drew on the multi-disciplinary expertise of advocates, patient representatives, clinicians, scientists, researchers and policy makers to conduct its work. We had a particular strength in having a specially established consultative forum co-chaired by two of our Steering Group members, to express the voice of experience throughout, which is published in an accompanying report, a HIQA conducted review of international best practice, a public consultation and invited presentations from a variety of policy and practice experts to the Steering Group.

The result is eleven recommendations that span the journey across the life course for those diagnosed with a challenging and difficult condition. We recommend an enhanced National Rare Diseases Office, firmly embedded in the new Sláintecare integrated care system that supports individuals from screening and diagnosis and through a lifetime of care. A system that provides equitable and timely access to expert specialist care regardless of where a person may live. The establishment of a state-of-the-art registry, reinforced ERN engagement and commitment to research innovation and impact for the betterment of care and development of evidence-based treatments are recommended.

We recommend also that the wider cost to PLWRD and their families be recognised and resourced and cross-sectoral recognition is afforded to that experience. We also emphasise the need for continuing education and awareness of the general public and all those professionals whose work brings them into contact with those experiencing a rare disease journey.

This is a Strategy, and it requires implementation. This is why the cornerstone of the next phase is the establishment of an Implementation Oversight Group which we are recommending to the Minister.

I would like to thank most sincerely all those who contributed to this work and I hope by 2030 we will see the evidence of what can be achieved.

**Chair**
Prof. Cecily Kelleher

# Executive Summary

## This National Rare Disease Strategy aims to meet the needs of the estimated 300,000 people living with a rare disease in Ireland.

It seeks to establish a comprehensive framework designed to improve diagnosis, treatment, and support for people living with a rare disease by enhancing quality of life, promoting equitable access to healthcare, and fostering innovation in rare disease research and treatment to create a more inclusive healthcare system that addresses the unique challenges posed by rare diseases.

While significant progress was achieved through the National Rare Disease Plan for Ireland 2014-2018, there were many areas where implementation did not progress as may have been envisaged, or where new technologies and advancements have overtaken those recommendations. As such, it is now timely that we renew our focus on rare diseases and update our vision and aims to ensure we have a National Rare Disease Strategy that meets the needs of people living with rare diseases (PLWRD), and their circle of support including families and healthcare workers, over the next five years.

In December 2023, the Chief Medical Officer (CMO) in the Department of Health established a National Rare Disease Steering Group to drive the collaborative and inclusive development of a new National Rare Disease Strategy. The Steering Group membership included patient representatives, patient advocates, clinicians, researchers, and representatives from the National Rare Diseases Office (NRDO), National Genetics and Genomics Office (NGGO), European Reference Networks (ERNs), Health Research Board (HRB), HSE Child Health Public Heath, the office of the Chief Clinical Officer (CCO) and the National Clinical Programme for People with Disability. The Steering Group were tasked with developing **'a National Rare Disease Strategy for the consideration of the Chief Medical Officer and Minister for Health, that sets out the vision for Rare Disease services in Ireland and outline the actions required to achieve this'**[1].

The Steering Group was led by an independent Chair with secretariat support from the Department of Health and was informed by presentations from a range of experts over the course of thirteen meetings and 4 workshops, a report from the National Rare Disease Patient Forum and the results of a wide-ranging public consultation.

This Strategy is first and foremost about people and building trust between all stakeholders through engagement, communication and partnership. By harnessing the expertise, knowledge and enthusiasm that exists across the rare disease community and healthcare providers to deliver on our ambition – preventing rare diseases, enabling earlier diagnosis of rare diseases and providing optimal care and support across the life course to people living with a rare disease to enable people to reach their full potential and to live their best lives.

1. Full Terms of Reference available in Appendix III

## Key Areas Addressed

**Leadership, Accountability and Governance:** The Strategy focuses on ensuring that the Department of Health and Health Service Executive (HSE) collaborate to support the continued enhancement of optimal care and support for all people living with rare diseases and their families. This includes the *expansion of the National Rare Diseases Office* to support and drive the recommendations of this Strategy, and to provide the governance and organisational arrangements to enable planning, management, and delivery of health and social care for people and for communities at a local level across the regions.

**Patient Partnership, Representation and Empowerment:** A central tenet of this Strategy is to ensure that rare disease partners are embedded into the fabric of all health and social care services so that people's needs and priorities are at the heart of policy and service development and delivery, driving more equitable and effective healthcare outcomes. This will be achieved through *the involvement of rare disease partners in the Implementation Oversight Group*, the establishment of a new PLWRD Partnership Advisory Group, and mechanisms to allow rare disease partners to share feedback, ask questions, connect with each other, access educational supports and monitor implementation of this Strategy's recommendations.

**Screening and Diagnosis:** Effective screening and timely diagnosis are critical to enable PLWRD and their families to access effective treatments and therapies to manage their condition appropriately before symptoms deteriorate and permanent damage occurs. This Strategy recommends that *screening, diagnostic and genetic services be expanded through an expansion of the newborn screening programme, support for the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland, and the development of diagnostic pathways* to reduce the diagnostic odyssey experienced by many people living with a rare disease.

**Eligibility, Access, and Integration of Healthcare Services Across the Life Course:** Integrated care is essential in ensuring that all people living with a rare disease and their families have access to equitable, inclusive and integrated health and social care that meets their needs throughout their life. This Strategy seeks to *ensure that this care and support is person-centred, integrated and coordinated among different healthcare professionals, services and settings for those living with a rare disease, as well as those with an undiagnosed rare disease or syndrome without a name (SWAN).* This will require collaboration across the healthcare sector including primary and community care and the acute services, ensuring access to necessary wrap-around services, transition of care from children and adult services and between settings, rehabilitation and end of life care as well as taking into account the financial cost of a rare disease diagnosis.

**Data, Health Information Systems & Registries:** Availability and ease of access to information are crucial for improving the lived experience of people living with a rare disease (PLWRD). Better information-sharing practices enhance patient outcomes, facilitate collaboration, inform policy, support research, and empower people with rare disease. This Strategy recommends the *establishment of a National Rare Disease Registry and to work to ensure the full implementation of ORPHAcodes as the optimal clinical coding system for rare diseases*. This will ensure that accurate rare disease data can be used as an essential tool for service planning, delivery, and evaluation, enabling the development of guidelines and standards, and assessing how Irish health services compare with international benchmarks.

**International Cooperation and European Reference Networks:** The Strategy emphasises the importance of collaboration at national, all-island, and European levels, in building partnerships with people living with a rare disease, rare disease organisations, healthcare providers, researchers, and policymakers to improve outcomes. It also seeks to *ensure that European Reference Networks are fully supported and resourced to enable their integration into the national health service so that the ERNs can play a leading role at a European level in the further development of services for patients and families with a rare disease diagnosis in Ireland*. International collaboration is also important in attracting attention and investment from industry to help stimulate funding and support for research that might otherwise be neglected.

**Rare Disease Research and Innovation:** To advance the understanding of rare diseases, this Strategy emphasises the importance of research. It *aims to increase investment in rare disease research, foster collaborations through international research networks, engage and involve people living with a rare disease, and inform evidence-based policy across all settings*. Improved engagement, collaboration, and partnership both nationally and internationally with industry, government, people living with a rare disease, patient organisations, healthcare professionals and academic partners will help to drive strong research and innovation for all rare diseases. The establishment of a Rare Disease Research Group, with patient partners included, also seeks to ensure a structured, coordinated, multi-actor and broad-based approach to rare diseases research in Ireland

**Orphan Medicines:** This Strategy recognises that orphan medicines have the potential to offer hope where, previously, little to none existed for many people living with a rare disease and, as such, aligns with the Programme for Government commitment to *review options for earlier reimbursement of orphan medicinal products, as well as early access schemes for rare diseases*. It also recognises the importance of enhancing the State's engagement with international initiatives to improve access to medicines.

**Public Information and Awareness Campaigns:** The general public may have never heard the name of most rare diseases, which can lead to misunderstanding and isolation for those affected by these diseases. Awareness campaigns inform the public about the nature of rare diseases, promoting empathy, compassion and empowerment. *By increasing general awareness across society, the recommendations in this Strategy will support communities and society as a whole to become a more understanding and inclusive place for people living with a rare disease*. To achieve this, a partnership approach must be taken to the design of public information and awareness campaigns, with people living with a rare disease, and their representative groups, co-designing information and awareness campaigns informed by lived experiences.

**Rare Disease Education & Training:** Education and training of healthcare professionals in rare diseases is crucial in improving diagnosis, treatment, care and support for people living with a rare disease. Since rare diseases are often complex and poorly understood, properly trained professionals can recognise symptoms earlier, offer a more timely and accurate diagnosis, and provide effective treatment and management of rare conditions. By *ensuring that appropriate modules and training on rare diseases are integrated into the curricula and training programmes at all stages including undergraduate, graduate and continuing professional development of health and social care professionals and carer disciplines*, we can ensure that our health and social care workforce are equipped with the knowledge, skills and awareness to provide better care and support for people living with a rare disease, their families, and carers.

**Implementation and Monitoring:** The successful implementation of Ireland's new National Rare Disease Strategy requires a comprehensive, coordinated, and sustainable approach to implementation and monitoring effectiveness. This strategy and its implementation represent a critical opportunity to transform health and social care for individuals and families affected by rare diseases. Implementation will aim to ensure that the vision of equitable, inclusive, and cross-sectoral care for people living with rare diseases is fully realised by ensuring strong governance and accountability, stakeholder engagement, transparency and inclusivity, as well as ongoing monitoring and evaluation of the Strategy's implementation and impact.

## Recommendation 1

### In relation to Leadership, Accountability and Governance, the Steering Group recommends:

**A.** strengthening and, where required, establishing, clear governance processes that appropriately define and describe the roles, responsibilities and function of all rare disease stakeholders, including patient partners, to ensure appropriate leadership and accountability at both national and regional levels.

**B.** that the National Rare Diseases Office, supported by the Office of the CCO and National Clinical Director for Integrated Care, is expanded and empowered with the necessary infrastructure and resources to drive implementation of this Strategy from a national perspective, and enabled to work in close collaboration with the Health Regions and other National Clinical Programmes, to provide guidance and support for the management, organisation and delivery of rare disease services.

**C.** that the office of the CCO, in collaboration with the Regional Executive Officers, develop clearly defined processes and measurable outcomes to ensure the Health Regions are provided with appropriate guidance and support to operationalise this Strategy.

## Recommendation 2

### In relation to Patient Partnership, Representation and Empowerment, the Steering Group recommends:

**A.** that PLWRD (People Living with a Rare Disease) and their representative groups are equal partners in improving health and social care services. This means PLWRD acting as equal partners across all areas, including the prioritisation, design, development, monitoring and evaluation of policy, services, and research on rare diseases. This partnership approach will be implemented across the country and driven centrally from the HSE and Department of Health, with representation from all areas in line with best practice and relevant national policy, thus ensuring rare disease voices are supported, valued and impactful throughout

**B.** establishing a PLWRD Partnership Advisory Group to support the implementation of the Rare Disease Strategy and to ensure a partnership approach across the various workstreams in the prioritisation, design, development, delivery, monitoring and evaluation of the Strategy's actions.

**C.** Financially supporting PLWRD acting as partners, in line with the HSE National Policy on Reimbursement of Expenses for Patient and Service User Partners, such that they can actively participate as partners without incurring significant cost.

## Recommendation 3

**In relation to Screening and Diagnosis the Steering Group recommends the expansion of screening, diagnostics and genetic counselling services in line with best practice to reduce the time to diagnosis, allow timely access to treatment and research/trial opportunities, and ensure PLWRD and their families are fully informed. This includes:**

**A.** Expediting the prioritisation, review and evaluation process for potential additions to the Newborn Bloodspot Screening Programme, supported by the dedicated review team within HIQA, while retaining the appropriate rigour;

**B.** Supporting and enhancing the current Newborn Bloodspot Screening Programme (NNBSP) to enable further expansion of the programme as per National Screening Advisory Group (NSAC) recommendations and Ministerial approval.

**C.** Supporting the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland as it relates to rare diseases.

**D.** Developing and implementing diagnostic pathways to reduce the time to diagnosis for all people living with a rare disease including access to research pathways for SWAN and undiagnosed people.

## Recommendation 4

**In relation to Eligibility, Access, and Integration of Healthcare Services, the Steering Group recommends that future services should be developed in an equitable, integrated, person and family centric manner aligned to Sláintecare and the Model of Care for Rare Diseases, specifically recommending:**

**A.** That, as part of the application process for medical card assessment, the financial cost of a rare disease diagnosis is recognised and medical expenses arising are taken into consideration.

**B.** The ongoing development and implementation of rare disease integrated care pathways including those for people with an undiagnosed rare diseases or Syndrome without a Name (SWAN).

**C.** The provision of integrated, coordinated, and multidisciplinary care and support within the community across all regions in line with individual needs, with timely referral and access to specialist care and support. This includes the development and greater utilisation of rare disease networks nationally and in other EU Member States and should be supported by understanding population-based needs.

**D.** Full implementation of the Model of Care for Transition from Childhood to Adult Healthcare Providers in Rare Diseases, the National Framework for Transition of Care and the development of associated care pathways to ensure appropriate and flexible transition arrangements for children, adolescents and young people, adults and older persons.

**E.** Access to necessary wraparound support services for PLWRD and their families throughout the life course, inclusive of rehabilitation, respite, palliative care, psychological and mental health supports, and all associated ancillary services in line with individual need.

## Recommendation 5

### In relation to Data, Health Information Systems and Registries, the Steering Group recommends:

**A.** The establishment of a national rare disease registry, underpinned by legislation, as necessary, 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 surveillance and data reporting is required across health and social care sectors.

**B.** A core project team, with the necessary supports, should be established 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.

**C.** The Steering Group recommends that the longer-term vision for the development of EHRs and associated IT systems should include the full integration of ORPHAcodes, as the gold standard nomenclature for rare diseases. A stepwise and an incremental approach to the integration of ORPHAcodes may be required to achieve this. In the interim, the capabilities of existing IT systems (i.e. SNOMED CT) should be leveraged to allow relevant data and information to be extracted for utilisation within a registry, surveillance, and service planning.

## Recommendation 6

In relation to International Cooperation and European Reference Networks, the Steering Group recognises that international and cross-border cooperation, including the integration of the European Reference Networks (ERNs) into the Irish healthcare system are essential to ensure access to expertise and the delivery of high-quality care and support to people living with a rare disease and recommends that:

**A.** International and cross border collaboration is not limited to ERN activity but rather, is actively promoted across the board, inclusive of policy development, clinical practice, training and education, and research

**B.** The HSE establish an ERN Integration Working Group of key stakeholders to develop and implement a roadmap for the integration and operationalisation of ERNs into the national healthcare system.

**C.** The NRDO in conjunction with the ERN Clinical Leads determine the necessary operating/staffing model for a functional ERN, such that they can deliver across all elements of the strategy, including data and research.

**D.** ERNs are fully supported and resourced to ensure effective and sustainable integration into the national healthcare system.

**E.** To ensure the long-term sustainability of ERNs, the DOH and the HSE take into account the recommendations and outcomes and outputs from the EU4Health Joint Action on the Integration of ERNs into National Healthcare Systems (JARDIN) when available.

**F.** The Department of Health and HSE actively encourage and support applications to join the remaining ERNs at the earliest available opportunity.

## Recommendation 7

In relation to Rare Disease Research and Innovation, the Steering Group recommends that a coordinated, and strategic approach to rare disease research across the island of Ireland be put in place. This Includes:

**A.** Establishing a National Rare Diseases Research Group, including PLWRD acting as partners, to provide a structured, coordinated, multi-actor and strategic approach to ensuring more high-quality Rare Diseases research in Ireland. This should aim to build research capacity and capability from basic research to the conducting of clinical research (including clinical trials) within the health services (including ERN hospital sites) and universities

**B.** Increasing the opportunities for PLWRD to access, as potential participants, high-quality clinical trials and healthcare interventions by supporting the RD Clinical Trial Network (HRB) and through Ireland's participation in the European Rare Diseases Research Alliance (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 will support this recommendation.

**C.** Increase opportunities for rare disease specific funded research calls targeting both indigenous research and greater participation in international consortia.

**D.** Ensuring healthcare professionals are supported and encouraged to pursue research in rare diseases and provided with the dedicated time to do so.

## Recommendation 8

In relation to Orphan Medicines, the Steering Group recommends that ongoing efforts should be leveraged to maximise access and availability to treatments and technologies for people living with a rare disease and should be explored in full. This includes:

**A.** Reviewing options for earlier reimbursement of certain treatments;

**B.** Consideration of early access schemes for rare diseases, with a view to facilitating sustainable and consistent access to new medicines;

**C.** Enhancing the State's engagement with international initiatives and Health Technology Assessment (HTA) collaboration aimed at improving people's access to medicines.

## Recommendation 9

### In relation to Public Information and Awareness Campaigns, the Steering Group recommends:

**A.** The development of a Rare Disease Communication and Awareness Plan, including input from PLWRD, to raise awareness of rare diseases among people/families living with rare diseases, the general public, healthcare professionals and policy makers.

**B.** Supporting and educating people living with rare diseases, their families, and carers about the specific rare disease they are dealing with, and the availability of national/international peer-to-peer supports. It is also important that these individuals help design the information, education, and support resources to ensure they meet their needs.

**C.** Development of accessible rare disease information, resources, and educational packages for people/families living with rare diseases and healthcare professionals to increase rare disease awareness and literacy, in collaboration with PLWRD.

## Recommendation 10

### In relation to Rare Disease Education and Training, the Steering group recommends enhanced collaboration with the education sector and training bodies, and the upskilling and development of the rare disease multidisciplinary workforce, this should include:

**A.** Adding and improving rare disease content in the core curricula of college and university courses for Healthcare Professionals at all levels, to enhance standards, including undergraduate, graduate entry, and postgraduate levels.

**B.** Ensuring ongoing training and professional development opportunities for Healthcare Professionals, such as short courses or flexible learning options (like micro-credentials[2]). This should include raising awareness about these opportunities so that more professionals take them up.

**C.** Greater ongoing promotion and utilisation of education and training resources and initiatives, especially those provided by the ERNs, across all primary and acute services settings in the Health Regions.

2. In health and social care, micro-credentials are short, focused courses designed to demonstrate specific knowledge, skills, and experience in a particular area. They are often flexible, part-time, and can be delivered online or through a blended approach. Micro-credentials are intended to provide upskilling and reskilling opportunities, allowing individuals to gain new competencies in a faster and more flexible way.

## Recommendation 11

**In relation to Implementation and Monitoring, the Steering Group recommends an Implementation Oversight Group (IOG) be established on publication of this Strategy by the Department of Health with appropriately led and resourced workstreams/project teams to support optimal implementation and monitoring of national strategic recommendations:**

**A.** The membership of the IOG will include senior responsible and accountable personnel across the Department of Health, HSE, patient partners and appropriate links to other stakeholders and bodies as required.

**B.** The HSE will lead on the implementation of the Strategy, with strong policy oversight by the Department of Health. This will be supported by thematic working groups as required.

**C.** The HSE will develop an implementation plan outlining the actions required for the implementation of this Strategy's recommendations and the associated timelines for agreement with the IOG, inclusive of key performance indicators and progress updates.

# 1

# Introduction

**Rare Diseases in Ireland** — A 'rare disease' is defined within the European Union as a life-threatening or chronically debilitating disease affecting no more than 5 people per 10,000[3]. Rare diseases are primarily genetic in origin, and due to the low prevalence of each disease medical expertise and scientific information on best practice and management is often limited. Additionally, diagnostics and identifying the optimal point of effective intervention can be a challenge.

Rare diseases can also be associated with multiple difficulties including cognitive, developmental, intellectual, mental, physical and sensory, or a combination of these. Rare diseases are a constellation of conditions, each of rare incidence but which have significant levels of need within a population attributable basis. These conditions are typically chronic, progressive, degenerative and often life-limiting, and often require a spectrum of care and support across the life course. There are between 6,000 – 8,000 known rare diseases affecting up to 6% of the total EU population (at least 30 million Europeans). This means that, approximately 300,000 people in Ireland are living with a rare disease[4]. This underlines the fact that rare diseases are individually rare, but collectively common and present difficulties for people living with rare diseases (PLWRD), their families and caregivers.

Rare diseases can impact both children and adults but are more likely to be first diagnosed in children and adolescents. Wakap & Lambert *et al.* (2020), when examining Orphanet[5] descriptions of 6,172 clinically unique rare diseases found that 72% (4,440) were described as genetic in origin. Where age of onset was defined (5,018), 70% (3,510) were exclusively paediatric onset, 18% (908) had onset spanning both childhood and adult groups and 12% (600) were exclusively adult onset. It was also found that of 3,585 rare diseases examined, 85% (3,031) had a prevalence of less than 1/1,000,000 at that point in time[6]. Applying these prevalence rates to Ireland would suggest that for many individual rare diseases there could be five or less people in Ireland living with that specific disease. These low prevalence rates for individual rare diseases can make the accumulation of knowledge and expertise on individual rare diseases very difficult and underscore the importance of international collaboration.

3. European Commission (2023). Rare Diseases and European Reference Networks. [online] Available at: https://health.ec.europa.eu/rare-diseases-and-european-reference-networks/rare-diseases_en.
4. Health Service Executive (n.d.). NRDO Patients and Families. [online] Available at: https://www.hse.ie/eng/services/list/5/rarediseases/patientsfamilies.html#:~:text=What%20is%20a%20rare%20disease,at%20least%2030%20million%20Europeans.
5. Orphanet is the European reference portal for rare conditions and orphan drugs. Established in France in 1997, Orphanet has grown to include 41 countries across Europe and the Globe. Orphanet provides reliable, high-quality information with the aim of improving diagnosis, care and treatment of people with rare conditions
6. Nguengang Wakap, S., Lambert, D.M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., Murphy, D., Le Cam, Y. and Rath, A. (2019). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. European Journal of Human Genetics, [online] 28(28). Available at: https://doi.org/10.1038/s41431-019-0508-0.

Gunne & Mc Garvey *et al.* (2020)[7], when examining the contribution of rare diseases to paediatric mortality in Ireland, found that 59% (2,368) of the 4,044 deaths registered in those aged < 15 years in the 11-year period 2006-2016, had an underlying rare disease. Stratifying by age group; 55.6% (1,140/2,050) of neonatal deaths had a rare disease, 57.8% (450/778) post-neonatal, and 64% (778/1,216) of children aged 1-14 years.

Gunne & Lambert *et al.* (2022)[8] in a study seeking to estimate the cumulative paediatric prevalence of rare diseases in children born in Ireland in the year 2000 found that, out of a total of 54,789 livebirths in the year 2000, 4.2% (2,283) were diagnosed with a rare disease by 17 years of age, with a mortality rate of 12% (270) before reaching 18 years of age. A significant proportion of the approximately 2,000 people with a rare disease alive at age 18 had chronic conditions that required adult care and support services. It is reported that the children diagnosed with rare diseases used 52% of paediatric bed days over the 18 years, and 60% of teenage bed days, despite representing only 4.2% of total number of children born in the year 2000. Children with rare diseases used 25 times more hospital bed-days as compared to their peers.

In 2022, Rare Diseases Ireland (RDI) published the Rare Reality research report[9] following administration of an online survey, with 111 eligible respondents. It found that, on average for each individual, five separate areas of health were affected by their rare disease. It found that 34% of respondents regularly attended four or more specialist clinics for their care. It also found that care is typically divided between specialist services and community services, with 30% of respondents reporting that they engage with hospital and/or GP services more than ten times per year and 23% use community services more than 10 times per year.

Due to the rarity and complexity of many rare diseases, people often experience delays in receiving an accurate diagnosis. Whilst numbers are necessarily small, nonetheless, preliminary data from RDI's Rare Reality Report found that 37% of PLWRD in Ireland were waiting over five years for a diagnosis. Difficulties in discovering a diagnosis meant that 53% reported being assessed and/or treated for three or more diseases on their journey to diagnosis and 73% reported seeing three or more consultants in different specialties on their journey to a diagnosis. This can lead to prolonged uncertainty and frustration, as well as missed opportunities for early intervention and treatment. It can also result in fragmented healthcare services, with people having to navigate multiple specialists and treatment centres, leading to challenges in coordinating and accessing comprehensive hospital and community supports.

7. Gunne, E., McGarvey, C., Hamilton, K., Treacy, E., Lambert, D.M. and Lynch, S.A. (2020). A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland. Orphanet Journal of Rare Diseases, 15(1). Available at: https://doi.org/10.1186/s13023-020-01574-7.
8. Gunne, E., Lambert, D.M., Ward, A.J., Murphy, D.N., Treacy, E.P. and Lynch, S.A. (2022). An estimate of the cumulative paediatric prevalence of rare diseases in Ireland and comment on the literature. European Journal of Human Genetics, 30(11), pp.1211–1215. Available at: https://doi.org/10.1038/s41431-022-01144-4.
9. Rare Diseases Ireland (2022). Rare Reality: Living with a Rare Disease in Ireland. [online] Available at: https://rdi.ie/wp-content/uploads/2022/02/Rare-Reality-Healthcare-Experiences-Jan-2022.pdf [Accessed 26 Mar. 2025].

It was also reported that a challenge for PLWRD is obtaining health and social care from an expert-led multi-disciplinary team that meets their needs, with 62% reporting that they receive care from a specialist consultant with expertise in their condition and 44% reporting that care is provided through an expert led multidisciplinary care centre. Almost one third (31%) of respondents indicate that there is no specialist in Ireland that has expertise in their rare disease.

More recent analysis of Irish data (n=105)[10] from research conducted via EURORDIS-Rare Diseases Europe Rare Barometer program[11] reported an average of 6.1 years for diagnosis in Ireland, with 83% reporting at least one misdiagnosis and almost half (49%) receiving inappropriate treatment. Over half (58%) were never referred to an expert centre during their diagnostic journey while one-third (34%) saw eight or more healthcare professionals on their journey to diagnosis.

The role of General Practice cannot be underestimated in providing care for people living with a rare disease, particularly in the Irish healthcare system where the General Practitioner (GP) often acts as the gateway to care and access in general is means-tested. GP's play a critical role in the early recognition and diagnosis of rare diseases, care coordination, management of comorbidities and education and advocacy. Equitable access to primary and community care networks, as well as hospital care is key in the context of expanded care and support for children and adults. For PLWRD and families, cross-sectoral interdisciplinary supports are required across the care and support continuum to improve diagnostics and to ensure that all people living with a rare disease are supported to reach their full potential and to live their best lives.

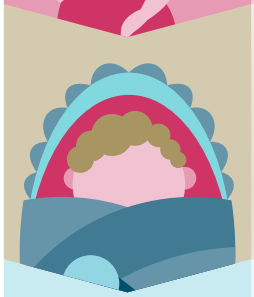
10. Arnott, C., Mcgrath, V. and Lynch, S. (n.d.). 'Comparing the Rare Disease Diagnostic Journey in the Republic of Ireland to the Rest of Europe – How Do We Compare?' [online] Available at: https://rdi.ie/wp-content/uploads/2025/02/RARE-DISEASE-DIAGNOSTIC-JOURNEY-Ireland-vs-Europe.pdf [Accessed 26 Mar. 2025].
11. EURORDIS (2024). The diagnosis odyssey of people living with a rare disease – EURORDIS. [online] EURORDIS. Available at: https://www.eurordis.org/publications/rb-diagnosis-odyssey/ [Accessed 26 Mar. 2025].

# A life course approach to rare diseases

## Preconception & Pregnancy

- Genetic Testing & Counselling
- Informed Family Planning & Reproductive Options
- Pre-natal Screening/Diagnostics

1



## Childhood

- Newborn Screening
- Genetic Testing & Counselling
- Multidisciplinary Care Coordination

2

## Adolescence

- Multidisciplinary Coordinated Care
- Transitional Care Planning
- Educational & Psychological Supports

3

## Adulthood

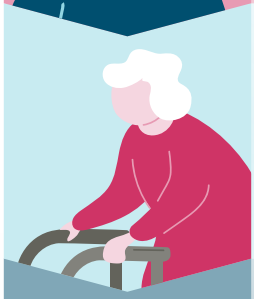
- Multidisciplinary Care Coordination
- Employment & Financial Supports
- Access to Clinical Trials
- Education & Advocacy Supports

4

## Older Persons

- Multidisciplinary Care Coordination
- Disease & Comorbidity Management
- Cognitive & Psychological Support

5



## Rehabilitation

- Multidisciplinary Rehabilitative Care
- Psychological Supports
- Assistive Technologies & Devices

6

## Palliative & End-of-Life Care

- Symptom Management
- Emotional & Psychological Supports
- Dignified and Respectful care in Flexible Care Settings

7

2

# Vision Statement

Our vision for this Strategy is to ensure that "all people living with a rare disease, and their families, have access to equitable, inclusive and cross-sectoral care throughout their life journey that will enable them to reach their full potential and to live their best lives".

We must be ambitious in seeking that every person living with a rare disease receives timely, equitable, high-quality, safe care and support throughout their life, while fostering innovation to deliver better diagnostics, treatments, and cures, where possible. This strategy strives to set Ireland on a course to become a global leader in addressing the needs of people with rare diseases, creating a transformative environment that ensures equitable access to care and supports, improves quality of life and accelerates advancements for individuals living with rare diseases. Given the complex challenges rare diseases present, the Strategy's vision and ambition reflect a commitment to addressing these issues holistically and sustainably.

This Strategy is first and foremost about people and building trust between all stakeholders through engagement, communication and partnership. Where possible, it is about PLWRD and their circle of support including caregivers and their representative patient advocacy organisations working in partnership[12] with:

- **healthcare professionals** in the community, in hospitals and in the Health Regions across Ireland to drive person-centred care and better outcomes for PLWRD;
- **researchers and healthcare service innovators** to deliver on the promise of new technologies and health system innovations that meet the needs of PLWRD; and
- **health service leadership, policy makers and Government** to adopt and put into action national and international policies that support implementation of this strategy in full and position Ireland as a global leader in rare disease innovation, policy, collaboration, health and social care equity, and supporting people living with rare diseases.

This Strategy should seek to harness the expertise, knowledge and enthusiasm that exists across the rare disease community and health and social care providers to deliver on our ambition – preventing rare diseases, enabling earlier diagnosis of rare diseases and providing optimal care and support across the life course to PLWRD to enable them to reach their full potential and to live their best lives.

12. Health Service Executive (2023). Our Vision for Partnership Across 'Our HSE' HSE National Patients' Forum. [online] Available at: https://www.hse.ie/eng/about/who/national-services/partnering-with-patients/national-patient-forum/our-vision-for-patient-partnership-across-our-hse.pdf.

## Key elements of our ambition for this National Rare Disease Strategy are:

| | |
|---|---|
| 1 | **Early diagnosis:** Through improved public and professional awareness, education, training and access to screening and diagnostic pathways our ambition is to reduce the average time to diagnosis for rare diseases significantly over the lifetime of this Strategy. Additionally, in line with the EURODIS call to action[13] we will aim to ensure that all people living with a rare disease known in the medical literature will be diagnosed in a timely manner once coming to medical attention, and those that are currently undiagnosable have the opportunity to access a relevant undiagnosed research pathway. |
| 2 | **Timely access:** Our ambition is to ensure that the principles of Sláintecare are realised for every person living with a rare disease within the lifetime of this Strategy, such that they receive holistic, person-centred, coordinated, multidisciplinary and expert care, at the right time and in the right place irrespective of diagnosis, geographic location or their ability to pay. |
| 3 | **Fostering an environment of innovation:** Our ambition is to create governance structures that support safe innovation, harnesses the power of data, research, clinical trials and the early adoption of technology across the health service. Central to this will be the establishment of a national rare disease registry, utilising a defined minimum dataset[14] with ORPHAcodes[15], that will provide a vital resource for service development, improved care and research. Our aim is to ensure that all people living with a rare disease have equitable access to the most effective and innovative diagnostics, treatments, therapies, and clinical trial opportunities, both in Ireland and internationally. |
| 4 | **Implement robust and transparent leadership, governance and accountability:** Establishing strong and transparent leadership, governance and accountability will be integral to achieving the overarching goals of this Strategy. Our ambition is to establish swiftly and support the necessary leadership, governance and accountability structures that will enable continuous health service development and innovation to become the norm. The establishment of an Implementation Oversight Group and HSE implementation structures will drive implementation of this Strategy in full across all services and settings, enabling regular monitoring and evaluation of this Strategy's impact. |

The four elements outlined above will be central to achieving our vision and ensuring PLWRD are supported and enabled to live healthier, fulfilled lives for longer. To translate the Strategy's vision, ambitions and the recommendations into meaningful and measurable goals, a suite of SMART goals, outcome measures and KPIs will be developed as part of implementation planning and iterated as required over the lifetime of the Strategy.

13. https://www.eurordis.org/our-priorities/diagnosis/
14. European Commission (2019). Set of common data elements for Rare Diseases Registration. [online] Available at: https://eu-rd-platform.jrc.ec.europa.eu/system/files/public/CDS/EU_RD_Platform_CDS_Final.pdf [Accessed 26 Mar. 2025].
15. ORPHAcodes are unique identifiers used in the Orphanet database. Each rare disease or condition listed in Orphanet is assigned a specific ORPHAcode, which helps to standardise and facilitate retrieval of information about that particular disease.

3

# Strategic Context and Approach to Development

## Strategic Context

The first National Rare Disease Plan for Ireland 2014-2018[16] contained 46 recommendations focused on improving diagnosis, identifying centres of excellence, and establishing dedicated rare disease pathways. While significant progress was made through the establishment of the National Rare Diseases Office in the HSE, publication of a Model of Care for Rare Diseases and Ireland joining 18 out of 24 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.

It is now timely that we renew our focus on rare diseases and update our vision and aims to ensure that this Strategy meets the needs of people living with rare diseases, and their circle of support including families and healthcare workers, over the next five years.

## International Context

The *Rare 2030 Foresight Study*[17] and the UN Resolution on *"Addressing the challenges of persons living with a rare disease and their families"*[18] are closely aligned in their overarching goals and vision. Both aim to address the systemic challenges faced by individuals with rare diseases and their families by advocating for the integration of rare diseases into public health and social care frameworks.

*Rare 2030* is a foresight study that gathered the input of a large group of people with rare diseases, practitioners and key opinion leaders to propose policy recommendations that will lead to improved policy and a better future for people living with a rare disease in Europe. It outlines key recommendations for shaping policies, such as advancing research, ensuring equitable access to diagnosis and treatment, fostering international cooperation, and prioritising empowerment and inclusion. Similarly, the UN Resolution emphasizes the importance of awareness, universal health coverage, and global collaboration to tackle barriers such as stigma, limited access to care and support, and insufficient data.

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[19]. 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. Further themes and priorities included access to orphan medicines and innovative health technologies, health information systems and registries; access to mental health and psychology services; and health workforce and training.[20]

16. Department of Health (2014). National Rare Disease Plan for Ireland. [online] Available at: https://assets.gov.ie/37342/da70fc6fadd24425b98311e679f4406b.pdf.
17. Kole, A., Hedley, V. and *et al.* (2021). Recommendations from the Rare 2030 Foresight Study: The future of rare diseases starts today. [online] Available at: https://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf.
18. United Nations (2021). UN Resolution on Addressing the challenges of persons living with a rare disease and their families. [online] Available at: https://documents.un.org/doc/undoc/ltd/n21/331/49/pdf/n2133149.pdf [Accessed 26 Mar. 2025].
19. Austria, Australia, Denmark, England, Finland, France, Germany, the Netherlands, Northern Ireland, Portugal, Scotland and Wales.
20. Health Information and Quality Authority (2024). Review of national rare disease strategies in selected countries. [online] Available at: https://www.hiqa.ie/sites/default/files/2024-02/Review-of%20national-rare-disease-strategies-in-selected-countries.pdf [Accessed 26 Mar. 2025].

Outcomes or measures related to strategy evaluation were outlined in four countries. There was variation between countries in governance and organisational structures through which Strategies were developed, implemented, monitored and evaluated. Seven out of 12 countries also outlined that funding was allocated to their Strategy implementation. While Ireland shared a number of similarities with other countries on the governance and organisational models outlined in the 2014 Plan, there was no specified funding model.

## Approach to Development

In February 2023, the Minister for Health announced an intention to develop a new National Rare Disease Strategy for Ireland. In April 2023, a dedicated policy unit was established within the Office of the Chief Medical Officer (CMO) to commence this 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*.

## National Rare Disease Steering Group

In December 2023, the Chief Medical Officer established the National Rare Disease Steering Group, chaired by Professor Cecily Kelleher. The Steering Group's Terms of Reference were as follows: **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.**

**In developing the National Strategy, the Steering group will:**

- 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.
- Have regard for Sláintecare, 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.
- Consider how best to integrate European Reference Networks into the national health system.
- 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.
- 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.
- Consider how to best promote rare disease awareness, including education among healthcare professionals, policy makers, and the public.
- Consider how to best promote and support participation in national and international research.
- 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 a number of 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

**The Steering Group also had broad regard for the underpinning principles of Sláintecare by considering:**

- The integration of rare disease services and the European Reference Networks into our national system to support the delivery of care in the right place and by leveraging expertise from across the European union and reducing the need for individuals to travel for specialised and complex care
- Ensuring PLWRD get the right care by further developing the highly specialised services they require.
- Ensuring integrated coordinated health and social care services in the community meet the needs of PLWRD and their family members, including psychological and mental health supports,
- Supporting people living with rare diseases in getting the care they need at the right time by supporting the creation of a more efficient and integrated system.

## National Rare Disease Patient Forum – Hearing the Lived Experience of PLWRD

The Steering Group recognised the important role that PLWRD, their families and their circle of support, have in bringing their expertise by lived experience of using health and social care services, together with their specific knowledge of a disease, illness, or disability, along with their wider experience of the health and social care system to the development of a responsive and person-centred Strategy. In February 2024, the Minister for Health established the National Rare Disease Patient Forum to ensure the voices and lived experience of people living with rare diseases and their families was central to the development of this Strategy.

**The Patient Forum was co-chaired by representatives from Rare Diseases Ireland (RDI) and The Irish Platform for Patient Organisations, Science & Industry, (IPPOSI) both of whom sat on the Steering Group to ensure that the input collected through the Patient Forum fed directly into the work of the Steering Group in the development of this Strategy.**

An Expression of Interest (EOI) sought applications from those interested in participating in the Patient Forum and those interested in becoming a Member of the National Rare Disease Steering Group. In total, 119 responses to the EOI were received, including 38 applications to become a Member of the Steering Group. Following a short listing process the Steering Group selected and invited two applicants to join the Steering Group. All applicants who expressed an interest in being members of the Patient Forum were invited to the first meeting of the Forum on 14th February 2024 where the co-chairs provided an overview of the proposed input of the Forum in the development of this Strategy.

**Subsequently, three online small group workshops were held to discuss and provide input on the topics of:**

- Integrated co-ordinated care across all settings (hospitals, clinics & community);
- Care for life – transitioning from paediatric to adult care, end of life care, and supported living; and
- Accessing the right expertise locally, nationally and/or internationally.

An in-person day-long workshop was also held in the Department of Health on 24th April 2024, to bring together members of the Rare Disease Patient Forum and to discuss the priority areas for the new Strategy, specific areas that need consideration, and how involvement of people living with rare disease can be incorporated into the implementation of the new Strategy.

Feedback from these meetings was provided to the Steering Group and a substantive Report, which outlined their key priorities raised over the small group workshops and in-person event, was presented to the Steering Groups to inform their work in developing this Strategy.

**Patient Partnership, Representation and Empowerment was identified as a high priority for Forum Members. The Patient Forum Report highlights the importance of involving people in all aspects of decision making, service delivery and continuous evaluation of this strategy and the subsequent implementation of same to measure success.**

Many participants highlighted the burden that advocating for oneself can become over time, with many raising the issue of consultation fatigue. The importance of receiving effective and efficient care coordination throughout their life course was highlighted as a potential improvement that could be implemented to mitigate this fatigue. Forum members further emphasised the importance of community and empowering others, seeking the development of, and greater access to, peer-to-peer support opportunities. Living with a rare disease can feel isolating, and Forum members raised the importance of improving a sense of belonging and empowering PLWRD in the wider community.

The overarching message from Forum members was that when PLWRD are empowered, they can contribute unique and valuable insights that help to shape policies and practices for the better.

## Public Consultation

A public consultation was held, primarily online, which sought input from a diverse range of stakeholders, including but not limited to the rare disease community, advocacy groups, industry, and healthcare providers. Its goal was to identify the strategic priorities for each of the known core stakeholder groups: people living with a rare disease, their carers/family, researchers, industry professionals and health care providers. Respondents answered a series of questions which gathered basic demographic and background information, as well as their views across a number of key domains. Each group was also provided with a selection of challenges most relevant to their self-selected category and asked to select the three most important to them. A free text option to capture additional challenges, possible solutions, and what is currently done well was provided to all participants.

## Key Findings from the Public Consultation

### Demographics

The Public Consultation received 601 total responses, 84% of response were from members of the Public and 16% were from organisations (16%). Of those members of the public who responded, 85% were female, 15% were male. When Public respondents were asked to select which of the six HSE Health Regions they were situated in, the largest group of respondents were situated in HSE Dublin and Northeast (25%) with the lowest number of responses from HSE Mid-West (8%). There were also a number of respondents from Northern Ireland (2%). These figures align with the population density of each region as detailed in the *HSE's Regional Population Profiles* in April 2024[21], once responses from Northern Ireland were excluded.

21. Health Service Executive (2024). Regional Population Profiles National Comparative Report. [online] Available at: https://www.hse.ie/eng/about/who/healthwellbeing/knowledge-management/health-intelligence-files/national-comparative-report-regional-population-profiles.pdf.

| HSE Health Region | Public Consultation Response % | Regional Population Profile % | Variance |
|---|---|---|---|
| HSE Dublin & Northeast | 25.70% | 23.10% | +2.60% |
| HSE Dublin & Midlands | 20.60% | 20.90% | -0.30% |
| HSE Dublin & Southeast | 21.00% | 18.90% | +2.10% |
| HSE Southwest | 12.10% | 14.40% | -2.30% |
| HSE Mid-West | 8.50% | 8.00% | +0.50% |
| West & Northwest | 11.90% | 14.80% | -2.90% |

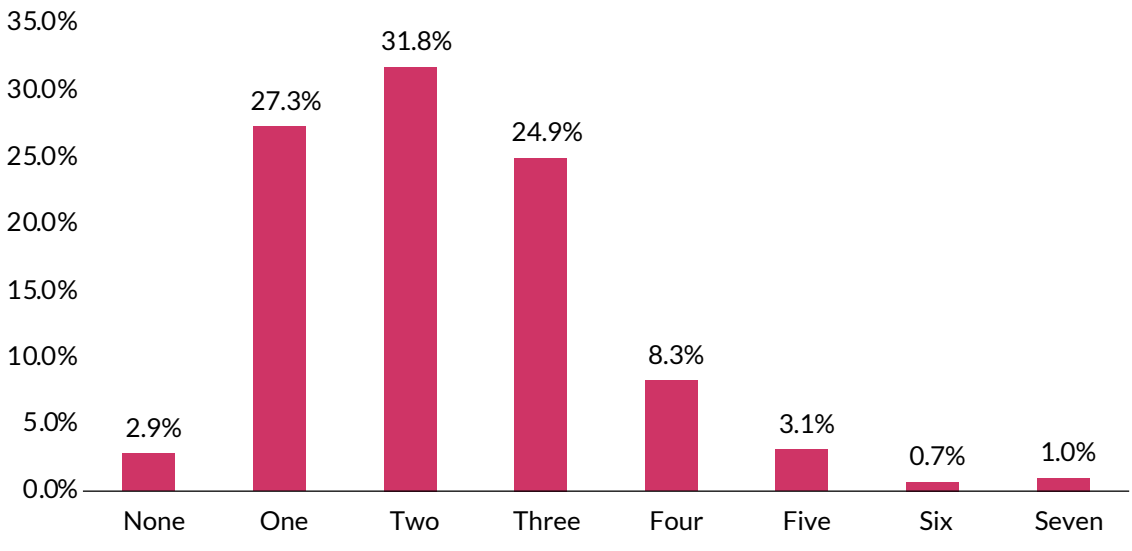
Of the 506 members of the public who responded, the highest category of responders identified themselves as a 'Person living with a rare disease' (49%) and 'Parent/relative/friend/caregiver of PLWRD' (45%). There were 95 responses from several types of organisations, 48% were from 'An advocacy group for people living with rare diseases or Non-Governmental Organisation (NGO), 36% from a 'Medical Organisation' or "public health body" and 16% from "industry or commercial lobby group".

## Diagnosis and Management of Rare Diseases

In relation to diagnosis, 93% of the 506 members of the public who responded said that they had a definitive diagnosis. Most survey participants had one rare condition (69%), however, a significant percentage reported having two to four conditions (30%). When asked to classify their condition as rare or ultra rare, 34% stated that their condition was rare (incidence of less than 1 in 2,000 people), 30% stated that their condition was ultra rare (1 in 100,000 people), and most significantly, 42.6% stated that they did not know if their disease was rare or ultra rare. There were over 250 distinct rare diseases reported.

When asked how many hospitals do you, your family member, or person you care for attend, 27% attended one hospital for treatment, while 70% attended more than 1 hospital for treatment. It was noticeable that 3.4% of participants stated that they did not attend any hospital for treatment for their condition.

Number of hospitals attended by members of the public who responded to the Public Consultation



Respondents were also given the option to list the services which they accessed. Of those who provided details of specific services accessed, the most common services being accessed were Cardiology, Dermatology, Haematology, Neurology, Occupational Therapy, Orthopaedics, Paediatrician, Physiotherapy, Psychiatry, Rheumatology, and Urology.

## Challenges Identified

All respondents were asked to select the top three challenges faced by people living with rare diseases. The top three challenges identified by members of the public were 'Awareness of rare disease amongst healthcare professionals'; 'Time to diagnosis/getting the right diagnosis (including access to genetic testing)'; and 'Access to specialist medical care and treatment'. Health care professionals, researcher and industry respondents varied in the areas they identified as challenges, these are outlined in the table below.

| Rank | Public Respondents[22] | Healthcare Professional | Researcher/Clinical Academics | Industry |
|---|---|---|---|---|
| 1 | Awareness of rare disease amongst healthcare professionals | Coordination of care across primary care and acute hospital services | Access to relevant data sets and registries | Technology appraisals and evaluations |
| 2 | Time to diagnosis/ getting the right diagnosis | Approval of funding for specialist care pathways at your centre in HSE service planning | Ethics approval, bureaucratic processes, administrative issues | Pricing and HSE affordability |
| 3 | Access to specialist medical care and treatment | Diagnosis, access to genetics/ genomic testing or reimbursement | Difficulty in getting support or funding for clinical research | Access to relevant outcome data sets and registries |
| 4 | Coordination of care between GP and hospital care; different hospital specialists; and health and social care services | Transition from child to adult services | _ | Evidence base requirements |

22. Public respondents include: Person living with a rare disease; Parent of a child living with a rare disease (under age 18); Relative or friend of a person living with a rare disease; Person caring for someone living with a rare disease; and Person at risk of a rare disease

When asked to provide information on how the identified challenges could be overcome, 26% of those who provided a response stated that public awareness campaigns and greater training for healthcare professionals would help overcome these challenges, while 12% felt that increased staffing and funding across rare disease services would help

Of significant note was that when asked…

**"what Ireland does well for people living with a rare disease" 52% of respondents living with or supporting a person with a rare disease provided answers that could be broadly categorised as negative, with many providing responses similar to "Little/Nothing/Not a lot".**

For those who responded positively, the largest proportion of total responses noted that the care, support and compassion shown by healthcare staff is a very positive (17.5%) aspect of rare disease services. While the support of the charity and voluntary sector was highlighted as a key resource for people living with a rare disease (10%).

**In summary, the results of the Public Consultation further highlighted that more work needs to be done to ensure those living with a rare disease, their families and carers, are adequately represented and empowered.**

4

# Leadership, Accountability and Governance

## Health Service Reform and the Rare Disease Strategy

The Department of Health and the HSE will collaborate to support the continued enhancement of optimal care for all people living with rare diseases and their families by ensuring that rare disease services and the implementation of this Strategy are:

- appropriately funded and resourced,
- guided by the needs of people living with rare diseases,
- delivered by a skilled workforce aligned to Sláintecare principles and
- underpinned by strong and effective leadership and governance

### Department of Health

The Department of Health has responsibility for setting health policy and strategy, leading on the development of legislation and regulatory policies, and overseeing the distribution of Government funding to health and social care services. It will work in close collaboration with the HSE to ensure that clear governance and accountability structures are in place across the system to support full implementation of this Strategy. This includes establishing an Implementation Oversight Group as detailed in chapter 11.

### Health Service Executive (HSE)

Notwithstanding the collective responsibility of the HSE Senior Leadership Team, at a national level the National Rare Diseases Office will take a lead role in the implementation of this Strategy's recommendations, reporting to the Chief Clinical Officer of the HSE through the National Clinical Director for Integrated Care. Health Regions will operationalise initiatives at a local level, with the guidance and support of the NRDO, to provide optimal care for PLWRD. This approach will leverage the new HSE structures to deliver on this Strategy's recommendations at both national and regional levels.

The HSE has undergone a period of significant change to a new structure with six Health Regions established and a revised national HSE Centre. This restructuring means that the broader health system should better meet the health and social care needs of all service users in line with Sláintecare. This Strategy will seek to ensure that the specific needs of PLWRD will be met by this new health delivery structure.

**In the context of this Strategy, it is important to ensure that the roles and responsibilities of the HSE at a national level, the National Rare Diseases Office and the Health Regions are clearly defined with respect to their specific role in the care of people living with rare diseases, including in the implementation of this Strategy. Having clear governance and accountability structures in place across the system will be a key factor in the overall success of this Strategy.**

## Office of the Chief Clinical Officer & the National Clinical Director for Integrated Care

Within the Office of the Chief Clinical Officer at the HSE Centre, the Office of the National Clinical Director for Integrated Care (NCDIC) will be responsible for the governance and oversight of the NRDO and be responsible for the implementation of this Strategy. The NRDO reports directly to the National Clinical Advisor and Group Lead (NCAGL) for Children and Young People within the office of the NCDIC and will work in partnership with relevant National Clinical Programmes (e.g. National Clinical Programme for People with Disability).

**The NCDIC will:**

- Provide national oversight for the implementation of the National Rare Disease Strategy;
- Ensure that PLWRD are included in governance and leadership structures in the development of national strategies and planning of services through the HSE's National Patients and Service Users Forum[23], the proposed Patient and Service User Partnership offices across the Health Regions and the Department of Health's Patient Partnership Advisory Group;
- Provide guidance and support to the Health Regions for integrated care and optimal patient outcomes in line with Sláintecare for PLWRD and their families/caregivers;
- Advocate for and support the development and commissioning of rare disease specialised care and services;
- Put in place mechanisms to develop and monitor outcomes such that they are consistent across Health Regions, providing equitable care for PLWRD regardless of geographic location;
- Provide governance of and support to the NRDO, ensuring it is resourced and enabled to effectively drive the implementation of this Strategy;
- Lead the development of the necessary HSE implementation structures required to coordinate and drive implementation of HSE led Strategy recommendations across various HSE functions.

23. Health Service Executive (2023a). National Patient and Service User Forum. [online] HSE.ie. Available at: https://www.hse.ie/eng/about/who/national-services/partnering-with-patients/national-patient-forum/.

## National Rare Diseases Office

The National Rare Diseases Office (NRDO) was established in 2015 as a key recommendation of the *National Rare Disease Plan for Ireland 2014-2018*. The NRDO is the national rare disease 'coordination hub' and the HSE's main contact for PLWRD and service providers. The NRDO provides support, information, and empowers people affected by rare diseases, families/caregivers and healthcare professionals.

**Core functions of the NRDO include:**

- The provision of the National Rare Diseases Information Service for PLWRD, families and caregivers, healthcare professionals and other stakeholders
- Collating and curating Irish rare disease information and data on Orphanet, the European portal for rare conditions and orphan drugs.
- Leading out on the design and supporting development of integrated care pathways
- Acting as a national Co-ordination hub to support rare disease national centres of expertise and ERNs.
- Working at a European level on EU rare disease projects e.g., leading on EU4Health Joint Actions; Orphanet Data for Rare Diseases (OD4RD) and the Integration of ERNs into National Healthcare Systems (JARDIN)
- Leading the development of rare disease education and awareness initiatives.
- Contributing to national and international research in rare diseases.

The National Clinical Programme for Rare Diseases (2013-2019) and the NRDO have delivered an extensive programme of work driving and supporting progress for people living with a rare disease, patient organisations and healthcare professionals.

Under the governance of the National Clinical Director for Integrated Care, it is intended that an expanded NRDO will support and drive recommendations in key areas such as:

- ERN integration into the national healthcare system
- Rare disease workforce planning
- Development of a national rare disease registry/data system
- Implementation of ORPHAcodes into health information systems and electronic health records
- Health and Social Care pathway design and development across the life course
- Transition of Care
- Education and training

## Health Regions

The Regional Executive Officers (REOs) have regional operational responsibility, with Health Regions as the primary delivery units for the majority of health and social care services provided across Ireland. They will provide the governance and organisational arrangements to enable planning, management, and delivery of care and support for people and for communities at a local level across their region.

In the context of this Strategy, six Regional Executive Officers (REOs) will be responsible for the operational delivery of services within each of the regions. The REOs and their teams will work with the Office of the CCO to identify the resources and structures required to deliver this Strategy at a regional level. The office of the NCDIC and the NRDO will work in close collaboration with the REOs, providing guidance and support on the delivery of this Strategy's recommendations at a regional level.

### Recommendation 1

**The Steering Group recommends:**

**A.** strengthening and, where required, establishing, clear governance processes that appropriately define and describe the roles, responsibilities and function of all rare disease stakeholders, including patient partners, to ensure appropriate leadership and accountability at both national and regional levels.

**B.** that the National Rare Diseases Office, supported by the Office of the CCO and National Clinical Director for Integrated Care, is expanded and empowered with the necessary infrastructure and resources to drive implementation of this Strategy from a national perspective, and enabled to work in close collaboration with the Health Regions and other National Clinical Programmes, to provide guidance and support for the management, organisation and delivery of rare disease services.

**C.** that the office of the CCO, in collaboration with the Regional Executive Officers, develop clearly defined processes and measurable outcomes to ensure the Health Regions are provided with appropriate guidance and support to operationalise this Strategy.

5

# Patient Partnership, Representation and Empowerment

People living with rare diseases and their advocates are uniquely positioned to provide insights into the challenges and opportunities in healthcare delivery, research, and policymaking.

**The involvement of PLWRD must go beyond consultation to ensure their representation in decision-making processes that shape the design, delivery, evaluation and sustainability of services and initiatives.**

'Good Public and Patient Involvement (PPI)' in the context of this Strategy involves a commitment to inclusion, empowerment, and collaboration/partnership. It requires a structured approach to integrating the voices of PLWRD at every level of health and social care delivery and strategy implementation, supported by appropriate resources, training, and capacity-building initiatives.

Achieving this means embedding rare disease patient partners into governance structures, advisory groups, and evaluation panels across the Department of Health, the HSE and other Departments as appropriate, ensuring they are equal partners alongside healthcare professionals, policymakers, and researchers.

By embedding rare disease patient partners into the fabric of all health and social care services, Ireland can ensure that the needs and priorities of its rare disease community are at the heart of policy and service development and delivery, driving more equitable and effective healthcare outcomes.

The PPI principles underpinning the implementation of this Strategy seek to ensure:

- **Inclusivity:** Patient partners should reflect the diversity of Ireland's rare disease community, encompassing different conditions, age groups, geographic locations, and socio-economic backgrounds.
- **Capacity Building:** Providing training and support to patient partners to ensure they have the skills and confidence to effectively contribute.
- **Transparency:** Clear and accessible communication on the objectives, scope, and expected outcomes of patient partnership.
- **Evaluation:** Regular review of PPI processes to assess impact and identify areas for improvement.
- **Support:** Ensuring patient partners are accessing the financial supports available.

### The importance of patient partnership for people living with rare diseases and their family members can't be overstated.

When it comes to addressing the medical and social care needs of our population, patient partnership for our parent-led organisation 22q11 Ireland has proved invaluable to us. Like very many rare conditions there is no cure for 22q11.2 deletion syndrome. In the absence of a cure, treatment and management of 22q is essential to our quality of life as families and for us, care-coordination is a treatment.

There are thousands of rare diseases, and no health care professional can know them all. Families however live with the impact of a diagnosis on a daily basis and become experts in the 'lived experience' of the condition. Lived experience provides insights and understandings that theory of clinical practice alone cannot give. Patient partnership allows for us to draw on the combined expertise of clinicians and families thus providing care that goes beyond just medical needs. 22q11 Ireland has made great strides towards a biopsychosocial model of care that our families have called for which includes co-design of innovative healthcare solutions and online parenting and young persons programs. There is so much more that can be achieved when we work in partnership with our clinical team and so much that is transferable to all rare diseases. When it comes to any rare disease there are common threads to the needs of families and our issues are usually a variation on the theme of access to health, social care and the educational needs of our children.

An estimated 1 in 17 people will have a rare disease during their lifetime. Those affected live with their families and in all our communities worldwide. They have lived experience and patient expertise when it comes to knowledge of their conditions and as such should always be an integral part of the design and delivery of care pathways and any research studies taking place.

Patient partnership is key to these processes.

ANNE LAWLOR, Co-Chair HSE Patient Forum, Chair 22q11 Ireland

## The Future for Ongoing Patient Partnership

While the Rare Disease Patient Forum established in 2024 will have fulfilled its original purpose with the launch of the National Rare Disease Strategy, its legacy is a foundation for ongoing engagement and partnership. Regular, meaningful and inclusive engagement with PLWRD and their representatives, including engagement with under-represented or more marginalised communities is essential for developing an effective, person-centred health and social system that meets the needs of PLWRD.

> **PPI is definitely the way forward. Though all talk of improving things needs to be carried through to action and actual change**
>
> – Response from Public Consultation

To ensure meaningful involvement of PLWRD in the implementation phase and into the future, the following steps are planned:

**Oversight:** An Implementation Oversight Group (IOG) will be established inclusive of appropriately resourced workstreams/project teams to support optimal implementation and monitoring of national strategic recommendations. These will be defined based on the recommendations of this Strategy, allowing for deeper engagement on specific issues affecting subsets of the rare disease community, such as access to treatments, diagnostic pathways, or integrated care and support across the life course. It is proposed that a partnership approach be taken and that PLWRD, their caregivers and representative partners are appointed as equal members of working groups and the IOG, as appropriate.

**Advisory:** A new PLWRD Partnership Advisory Group will be established through the Department of Health to facilitate ongoing insights to the IOG on the real-world impact and lived experience of PLWRD, through the PLWRD representatives on the IOG. This new PLWRD Partnership Advisory Group should also work to connect and engage with the National Patients and Service Users Forum[24] and the proposed Patient and Service User Partnership offices across the Health Regions, to engage in a sustainable and coordinated way to identify the needs, preferences and expectations for policy development, service design, and individual care into the future. The PLWRD Partnership Advisory Group will be jointly chaired by the Department of Health and patient partners to ensure that the voice of people living with rare disease is central to both policy development and service delivery as part of this Strategy's implementation.

**Shared Learning and Communications:** Mechanisms will be established to allow patient partners to share feedback, ask questions, connect with each other, access educational supports and access updates on the Strategy's implementation. This will include hosting events that bring together PLWRD, policymakers, clinicians, and researchers to consider implementation progress and share best practices. Such events will also provide the opportunity to engage with the broader rare disease community and offer an opportunity to ensure a wide range of voices are heard.

Achieving a high level of patient partnership will also require ensuring PPI partners are accessing the financial supports available through the HSE National Policy on Reimbursement of Expenses for Patient and Service User Partners[25] to enable them to be involved without placing significant additional costs upon them for expenses incurred as part of this work.

24. Health Service Executive (2023a). National Patient and Service User Forum. [online] HSE.ie. Available at: https://www.hse.ie/eng/about/who/national-services/partnering-with-patients/national-patient-forum/.
25. Health Service Executive (2025). HSE National Policy on Reimbursement of Expenses for Patient and Service User Partners. [online] Corporate. Available at: https://www2.healthservice.hse.ie/organisation/national-pppgs/policy-on-the-reimbursement-of-expenses-for-patient-and-service-user-partners/ [Accessed 26 Mar. 2025].

Finally, to ensure that this partnership has maximum impact, the following actions will be prioritised:

- Clear Terms of Reference will be developed that outline the roles and responsibilities of patient and service user partners;
- Collaborative leadership will be fostered to the greatest extent possible, with co-chairing arrangements between patient partners, policymakers, clinicians and researchers to emphasise equality in decision-making;
- The Rare Disease Policy Unit in the Department of Health will support the operation of the PLWRD Partnership Advisory Group, to support communication activities and events, and to support capacity building;
- Ensuring PLWRD partners do not incur significant cost in line with the HSE National Policy on Reimbursement of Expenses for Patient and Service User Partners[26];
- Integration across health systems will be prioritised to establish direct links between the advisory group and relevant HSE divisions, research bodies, and service providers.

## Recommendation 2

### The Steering Group recommends:

**A.** that PLWRD (People Living with a Rare Disease) and their representative groups are equal partners in improving health and social care services. This means PLWRD acting as equal partners across all areas, including the prioritisation, design, development, monitoring and evaluation of policy, services, and research on rare diseases. This partnership approach will be implemented across the country and driven centrally from the HSE and Department of Health, with representation from all areas in line with best practice and relevant national policy, thus ensuring rare disease voices are supported, valued and impactful throughout

**B.** establishing a PLWRD Partnership Advisory Group to support the implementation of the Rare Disease Strategy and to ensure a partnership approach across the various workstreams in the prioritisation, design, development, delivery, monitoring and evaluation of the Strategy's actions.

**C.** Financially supporting PLWRD acting as partners, in line with the HSE National Policy on Reimbursement of Expenses for Patient and Service User Partners, such that they can actively participate as partners without incurring significant cost.

26. Ibid.

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# Screening and Diagnosis

People living with a rare disease in Europe can often wait years to receive an accurate diagnosis, with one study reporting that 70% of respondents waited more than one year to get a confirmed diagnosis after coming to medical attention, with the average wait time of five years for a confirmed diagnosis.[27] It is important, however, to note that this data is based on a survey where the majority of those surveyed were aged >50 years and may not have had access to today's diagnostic techniques.

Effective screening and timely diagnosis are critical to enable PLWRD and their families to access effective treatments, therapies and health and social care supports, to manage their condition appropriately before symptoms deteriorate and permanent damage occurs.

**The vast majority of rare diseases are genetic in origin and as a result, approaches such as screening and, where appropriate, genetic/genomic testing are important methods to ensure their timely identification.**

Most PLWRD receive only symptomatic treatment. An accurate diagnosis can result in better management of the disease based on evidence, identification of potential therapeutics, avoid unnecessary treatments and potentially enable access to clinical research and trials opportunities. While this chapter deals primarily with diagnosis and screening, ensuring the appropriate post diagnosis care and support is available and integrated into the health services is equally important.

## Newborn Screening

**The HSE provides two population-based screening programmes for newborn babies, the National Newborn Bloodspot Screening Programme (NNBSP) and the National Universal Newborn Hearing Screening Programme (NUNHSP).**

The primary focus of these screening programmes is to identify babies with rare but serious clinical conditions for whom the outcomes are better with the earliest access to appropriate investigations, diagnosis and interventions.[28] The NNBSP is of particular focus as this has the potential to screen for more rare conditions than it currently does.

27. Faye, F., Crocione, C., Anido de Peña, R., Bellagambi, S., Escati Peñaloza, L., Hunter, A., Jensen, L., Oosterwijk, C., Schoeters, E., de Vicente, D., Faivre, L., Wilbur, M., Le Cam, Y. and Dubief, J. (2024). Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey. European Journal of Human Genetics, [online] pp.1–11. doi:https://doi.org/10.1038/s41431-024-01604-z.

28. Health Service Executive (2024). *Newborn Screening Shortly after Birth.* [online] HSE.ie. Available at: https://www.hse.ie/eng/health/child/newbornscreening/.

> **We got our son diagnosed within three weeks of birth, although it was a difficult time for us, were very grateful that we knew from an early age.**
>
> – Responses from Public Consultation

Since the publication of the National Rare Diseases Plan for Ireland 2014-2018 the NNBSP has been further expanded and now incorporates screening for nine conditions including Phenylketonuria (PKU), Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD), Glutaric Aciduria Type 1 (GA1) and Adenosine Deaminase Deficiency Severe Combined Immunodeficiency (ADA-SCID). All nine conditions currently in scope for screening as part of the NNBSP are considered rare but have a relatively high incidence in the Irish population when compared to international incidence rates.[29]

The establishment of the Independent National Screening Advisory Committee (NSAC) in 2019 has resulted in the substantive reform of the process for changes or additions to Ireland's screening programmes. NSAC works closely with the HSE, evidence synthesis from the Health Information Quality Authority (HIQA) Health Technology Assessment team, clinicians and patient advocates in its inclusive approach to the expansion of the NNBSP, seeking clinical expertise as required and involving patient advocates in its processes. The Committee holds calls for submissions for proposals for new screening programmes that may be introduced in Ireland, as well as suggestions for potential expansion of the six existing programmes, including the NNBSP. These calls for submissions are open to everyone, from members of the public, HSE and other medical professionals, and NSAC advises the Minister on changes/additions to the NNBSP. PLWRD are encouraged to make submissions to NSAC through this open call. Additionally, work is underway to improve transparency of the process and provide clear information on the current status of proposals on the NSAC work programme.

**The NNBSP has been incrementally expanded since its inception in 1966. In 2023, the Minister for Health endorsed two recommendations from NSAC on the addition of Severe Combined Immunodeficiency (SCID) and Spinal Muscular Atrophy (SMA) to the NNBSP. Once fully implemented, this will bring the number of conditions in scope to 11, representing a 37% increase since 2022. Nevertheless, there remains significant scope for further expansion of the Programme.**

Studies have shown that heel prick screening has the potential to be utilised for up to 50 rare diseases. Italy currently screens for the highest number of conditions in the EU with 48 conditions (or more, depending on the region) in scope for its newborn screening programme, followed by Austria (31) and Portugal (30).[30] However, there is considerable variation across Europe, as well as regionally within countries, as to the number of conditions each country screens for within its programme, with some screening for as few as two conditions and more close neighbours e.g. UK, France, Germany etc. screening for 9, 13, and 21 conditions respectively.

29. Health Service Executive (2023). *Report of the HSE National Children's Screening Programmes 2020-2022 National Newborn Bloodspot Screening Programme National Universal Newborn Hearing Screening Programme*. [online] Available at: https://www.hse.ie/eng/health/child/newbornscreening/hse-national-childrens-screening-programmes-report-2020-2022.pdf.
30. Charles River Associates (2024). *A landscape assessment of newborn screening (NBS) in Europe*. [online] Crai.com. Available at: https://www.crai.com/insights-events/publications/cra-insights-a-landscape-assessment-of-newborn-screening-nbs-in-europe/.

### I was diagnosed with Retinitis Pigmentosa in 1998 after a family member was tested in the Royal Victoria Eye & Ear Hospital.

In 2013 I was asked to take part in genetic testing research program called Target 5000, funded by the patient organisation Fighting Blindness. As I had other medical problems like under active thyroid, type 2 diabetes and other medical problems that generally only older people get my consultant suspected that there was something else going on. In fact, as I was being enrolled my consultant knew what might be causing my health issues, but couldn't tell me until I had genetic testing done.

At the time the laboratory testing was being performed outside of Ireland. It could only be moved forward when Fighting Blindness got more funding, so it was done in stages. Every couple of years I had to have another blood test taken to be sent off to rule out other things.

I finally got my diagnosis of Bardet Biedl Syndrome in 2021. I was happy to get a result, but I didn't realise how hard a journey I was going to have to get an appointment with consultants with knowledge and expertise in my rare disease; consultants who could make sure that no damage had been done to my body as I was then in my forties. I had been living with my condition since I was a child but didn't know about it. If my family had known the diagnosis earlier things would have been much easier for us.

I was really frustrated as it took so long to get an answer. It took nearly eight years to get a diagnosis. It took a toll on my mental health as I was worried about what was going to come up. When I finally got my diagnosis it came in a letter, before I even got the phone call to meet with a genetic counsellor.

When I went in to get information about my diagnosis it was all very medical and I was told that there was a support group and help in the UK but nothing in Ireland. Since my diagnosis a core group of people in Ireland living with BBS and parents of children who have BBS have come together. We have had several meet ups and an information day. I find that a lot of adults don't want to use their voice to talk about it.

I have gotten involved with this group as I want to make things better for children and adults coming behind me. I finally got referred to different consultants that are unfortunately located across three different hospitals. This is very tiring and adds to the stress. The Irish group are currently trying to get a multi-disciplinary clinic set up in Ireland so that we don't have to go from hospital to hospital or travel to the UK for treatment.

I wish that genetic testing and diagnosis could happen quicker as this would take the worry and stress away from patients and their families. It would mean that people could get help with medical conditions earlier and it wouldn't be so stressful for them later in life. It would also be great if the diagnosis is given by a genetic counsellor instead of receiving it in a letter. Dr Google sometimes provides incorrect information sending you in the wrong direction altogether.

Gillian Stafford – Member Rare Disease Steering Group

The establishment of NSAC has provided a clear process to consider new conditions for inclusion in the NNBSP and other relevant potential screening programmes for rare diseases. However, there remains a need to consider suitability for many more conditions to be included as part of the NNBSP, and other screening programmes, in line with our specific population needs and the needs of the rare disease community. Efforts are ongoing in this regard, with additional resources being provided to HIQA, including the establishment of a dedicated NNBSP Health Technology Assessment team, which will enable it to undertake a greater number of evidence reviews processes and support NSAC's expansive work programme. This team will also play a key role in identifying opportunities for collaboration and information sharing internationally. Additionally, the NSAC has also established a newborn screening subgroup to provide advice to NSAC regarding the prioritisation of more than 30 candidate conditions for possible consideration. Membership of this group includes a range of relevant experts, patient representatives, and representatives from the Rare Disease Policy unit within the Department of Health.

**In order to ethically and optimally expand the NNBSP, it is imperative that the associated diagnostics and treatments are also available. It is also important that the HSE be supported and adequately resourced to continue to deliver its quality assured screening programmes for rare diseases and expand these programmes in line with NSAC and ministerial requests, as required.**

This includes working collaboratively and proactively to design and develop the necessary follow-on services ahead of time to ensure that the expansion of any screening programme is not done in isolation or in the absence of the necessary follow-on services, and vice versa.

## Diagnostic Testing

While some rare diseases are easily diagnosed and have a treatment intervention, many others do not. People living with a rare disease often face lengthy and costly diagnostic journeys, consulting multiple physicians and undergoing extensive testing. Many remain undiagnosed or misdiagnosed for years, causing additional emotional distress for patients and families.

**Diagnosing rare conditions at the earliest opportunity also allows for more immediate intervention, giving PLWRD the opportunity to access appropriate information, treatments that might be available, better evidence-based care and wrap around supports that can enable people and their families to better plan for their future care.**

The introduction of Perinatal Genetics and Genomics Service has been a significant and welcome development since the publication of the previous rare disease plan. Perinatal genetics focuses on identifying, managing, and treating prenatal abnormalities such as chromosomal anomalies, hereditary disorders, and metabolic conditions. A comprehensive perinatal genomic service includes in-pregnancy testing, pre- and post-test counselling, and the investigation of genetic causes. Retaining strong links with perinatal pathology can help to guide future clinical care. In collaboration with maternity networks, the HSE's National Women and Infants Health Programme (NWIHP) drafted a framework for a National Perinatal Genomics Service[31].

31. HSE National Women and Infants Health Programme (2022). National Women and Infants Health Programme. [online] Available at: https://www.hse.ie/eng/about/who/acute-hospitals-division/woman-infants/national-reports-on-womens-health/national-women-and-infants-health-programme-report-2022.pdf.

In the case of rare diseases that are genetic in origin, diagnosis can inform case finding within the family, and family planning. For certain disorders prenatal testing is possible, consideration could be given to development of preimplantation genetic testing services, whereby testing could be provided under certain circumstances and as deemed clinically appropriate.

The National Genetics and Genomics Office is delivering improved access to clinical genetics as part of implementing the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland. There will be improved access to genomic medicine services in each Health Region and this will improve diagnosis and supports for PLWRD and their families.

The Clinical Genetics Service at Children's Health Ireland (CHI) provides general genetic counselling to the Irish population. The service cares for and manages families with genetic conditions rather than focusing on individual patients. The CHI service is provided in CHI Crumlin with satellite clinics in Cork, Limerick, Galway, and CHI Temple St.[32]

Additionally, St James's Hospital runs a Cancer Genetic Service for individuals and families with rare hereditary cancer risks, offering risk assessments, genetic testing, and counselling to support prevention and early detection.[33]

While there have been a number of positive developments across both diagnosis and screening of rare diseases, further work is required to remove barriers to prompt diagnosis for PLWRD. This includes the development of diagnostic pathways, addressing long waiting periods and ensuring specialist services are available and appropriately integrated into the community as far as is practicable. The *National Strategy for Accelerating Genetic and Genomic Medicine in Ireland* (the Genomics Strategy) was published in 2022. The National Genetics and Genomics Office (NGGO) was subsequently established in 2023 and tasked with advancing the strategy's implementation. This will be instrumental in strengthening the infrastructure needed to make advances in genetic and genomic services and reduce waiting times for genomic medicine services, and this will improve patient outcomes in areas including rare disease.[34]

32. Royal College of Physicians Ireland (2024). Clinical Genetics Medical Workforce in Ireland 2024-2038. [online] Available at: https://www.hse.ie/eng/staff/leadership-education-development/met/plan/specialty-specific-reviews/clinical-genetics-medical-workforce-in-ireland-2024-2038.pdf [Accessed 26 Mar. 2025].
33. St. James's Hospital (2025). Cancer Genetics Service | St James's Hospital. [online] St James's Hospital. Available at: https://www.stjames.ie/cancer/yourtreatmentandcare/servicesandtreatments/cancergeneticsservice/ [Accessed 26 Mar. 2025].
34. Health Service Executive (2022). National Strategy for Accelerating Genetic and Genomic Medicine in Ireland A National Strategy for Accelerating Genetic and Genomic Medicine in Ireland. [online] Available at: https://www.hse.ie/eng/about/who/strategic-programmes-office-overview/national-strategy-for-accelerating-genetic-and-genomic-medicine-in-ireland/national-strategy-for-accelerating-genetic-and-genomic-medicine-in-ireland.pdf.

# Genetics & Genomics

## National Strategy for Accelerating Genetic and Genomic Medicine in Ireland

Genomic medicine, incorporating clinical, laboratory and bioinformatics services, has the potential to improve healthcare, allowing for more targeted treatments resulting in better patient outcomes and a shift in healthcare from a disease-oriented 'one size fits all approach' to a more personalised, tailored service for people with rare diseases. The Genomics Strategy recognises the existing challenge of coordinated care for patients and families affected by rare diseases and undiagnosed conditions that requires the input of a range of different healthcare professionals. Improving the person's experience in accessing genomic medicine services (both clinical and laboratory) is recognised as a key contributor to the delivery of high-quality coordinated healthcare for people with a rare disease.

At present, PLWRD and their families experience long wait times to access testing, consultant opinion and genetic counselling, and the impact of these experiences was recognised during the development of the Genomics Strategy. In response the Genomics Strategy sets out a comprehensive vision for Ireland's future genomic medicine service that is person and family-centred, delivered through a strong national centre and implemented across the Health Regions.

As outlined in the Genomics Strategy the use of genomics has improved rates of diagnosis for PLWRD and can reduce the extended length of time it can take for people to receive a diagnosis. However, there continues to be a large number of PLWRD who experience a 'diagnostic odyssey' remaining undiagnosed for years and experiencing challenges in accessing appropriate care and support as a result. International studies show that for 25% of people who received a genetic diagnosis, there was immediate clinical actionability. In the diagnosis of rare diseases, there is a clear economic and personal benefit in early genome sequencing.[35]

The NGGO has developed a 5-year plan to expand both clinical services and to develop genomic testing in Ireland. The NGGO will focus on creating an equitable and nationally accessible clinical genetics service. To deliver this service an increase in the numbers of Consultant Clinical Geneticists, Genetic Counsellors and Genomic Resource Associates will take place from 2025. This will lead to improved national access to genomic medicine for PLWRD and their families.

**The introduction and development of national genomic testing services in Ireland has been mapped to the Genomics Strategy's key objectives which will be implemented from 2025 – 2029. The first of these key objectives was the development and delivery of a National Genomic Test Directory for Rare and Inherited Disease.**

35. Smedley, D., Smith, K.R., Martin, A., Thomas, E.A., McDonagh, E.M., Cipriani, V., Ellingford, J.M., Arno, G., Tucci, A., Vandrovcova, J., Chan, G., Williams, H.J., Ratnaike, T., Wei, W., Stirrups, K., Ibanez, K., Moutsianas, L., Wielscher, M., Need, A. and Barnes, M.R. (2021). 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care — Preliminary Report. New England Journal of Medicine, 385(20), pp.1868–1880. doi:https://doi.org/10.1056/nejmoa2035790.

Genomics is a relatively new science, and the introduction of the Test Directory will enable the person and clinicians to receive the right test, at the right place, at the right time. The first version of the Genomic Test Directory for Rare and Inherited Disease was published on the HSE website in December 2024.

## National Genomic Test Directory for Rare and Inherited Disease

The Test Directory will:

- Deliver equitable genomic testing for eligible people to the full range of clinically appropriate genomic tests which meet the needs and prevalence of conditions in the population.
- Provide clear detail about who is eligible for testing and where in a person's care pathway testing should be performed.
- Indicate which technology should be used for testing and which clinicians can request testing.
- Standardise the national approach to testing and provide a systematic approach to introduce new genomic tests in the future in line with the latest research and evidence.

The key to success in harnessing the power of genetic and genomic testing for improving a person's outcomes lies in the supporting infrastructure to collect, test, store, process, and analyse samples and data for patient diagnoses, prognostic and predictive testing, and ongoing research applications. As part of the Genomics Strategy's implementation, the NGGO will work to develop a future-proof, scalable national model for genetic and genomic testing, bioinformatics, and data management.

> **Support for immediate and extended family with a genetic illness – information, clear pathways to testing, support during pregnancies at risk of disease, genetic counselling is needed.**
>
> – Responses from Public Consultation

## Future Progress

Progress has been made in enhancing newborn screening and diagnosis for rare diseases, and this remains a priority for the Department of Health and the HSE. The establishment of NSAC, and more recently the establishment of its newborn screening subgroup, and dedicated Health Care Technology (HTA) team, reflects the growing recognition of the need to continuously evaluate and refine the NNBSP, to ensure that newborns have the best possible start in life, and that those with rare disease are identified at the earliest possible opportunity.

**The continued roll out of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland has set a clear direction for introducing and developing genomic testing in Ireland, for improving access to clinical genetics and for integrating genomic testing into the healthcare system and these developments will ultimately improve the time to diagnosis for PLWRD.**

Additionally, the development of a test directory for rare and inherited diseases provides clarity on the growing array of available tests, helping clinicians to choose the right test for the right patient at the right time in their diagnostic journey and enabling the person and families to be better informed. Furthermore, the continued development of the perinatal genomics service will support the better identification of rare diseases before birth, offering parents timely information and more comprehensive supports.

### Recommendation 3

In relation to Screening and Diagnosis the Steering Group recommends the expansion of screening, diagnostics and genetic counselling services in line with best practice to reduce the time to diagnosis, allow timely access to treatment and research/trial opportunities, and ensure PLWRD and their families are fully informed. This includes:

**A.** Expediting the prioritisation, review and evaluation process for potential additions to the Newborn Bloodspot Screening Programme, supported by the dedicated review team within HIQA, while retaining the appropriate rigour;

**B.** Supporting and enhancing the current Newborn Bloodspot Screening Programme (NNBSP) to enable further expansion of the programme as per National Screening Advisory Group (NSAC) recommendations and Ministerial approval.

**C.** Supporting the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland as it relates to rare diseases.

**D.** Developing and implementing diagnostic pathways to reduce the time to diagnosis for all people living with a rare disease including access to research pathways for SWAN and undiagnosed people.

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# Eligibility, Access, and Integration of Healthcare Services across the Life Course

## Integrated care and support are essential in ensuring that all people living with a rare disease and their families have access to equitable, inclusive and integrated health and social care that meets people's needs throughout their life.

Only by ensuring person-centred, integrated and coordinated care and support among different healthcare professionals, services and settings, including a range of social and community services, can we enable people living with a rare disease to attain the highest standard of health and wellbeing and realise their full potential. It is also important that speciality networks of care, such as national speciality outreach services (e.g. CHI outreach clinics), which assist in the provision of care closer to home for PLWRD are supported.

Within the HSE, the new Health Region structure aims to deliver integrated care and support to the population. People will experience a comprehensive continuum of care and support that meets their needs from a health promotion and disease prevention perspective, providing diagnosis, treatment and symptom and disease management as close to home as possible. Integrated care and support improves access, quality of care and outcomes, and puts the person's experience at the centre. It is based on the principles of early identification, empowering the person, multi- and inter-disciplinary cross-service care planning and delivery, where health and social care services work together to provide a flexible network of care and support that is responsive to the changing needs of people and their families.

> **Our neurologist is excellent and staff are generally great once admitted. Access to drugs, medical card, DCA and LTI have been easy. This is partially because of our excellent and experienced local pharmacist.**
>
> – Response from Public Consultation

This means improving the way care and support are provided so that people with rare diseases, the majority of whom live with serious, chronic and highly complex health conditions, as well as with disabilities, can live healthier and more independent lives. Integrated care and support must also ensure that PLWRD are supported throughout their life, from early childhood onwards:

**Specialist Care incl. Palliative/Respite/Rehabilitation/Psychological Services**

**Neonatology/Early Childhood** → **Transitions of care across the life course**

## Achievements since the Publication of the National Rare Diseases Plan for Ireland

In 2013, the National Clinical Programme for Rare Diseases was established as a joint initiative between the HSE and the Royal College of Physicians of Ireland (RCPI). It led to the establishment of the NRDO in 2015 with an early focus on improving patient empowerment and access to reliable information on expertise in rare diseases.

**In 2019 the National Clinical Programme for Rare Diseases published the HSE 'Model of Care for Transition from Paediatric to Adult Healthcare Providers in Rare Diseases'. This document provides a number of recommendations/guiding principles for healthcare providers to ensure a smooth, safe, and effective transition of young people with a rare disease from paediatric to adult health services. Additionally, the importance of well managed and coordinated transitions across all care settings at all stages of life must be recognised.**

A Model of Care for Rare Diseases was published in 2019 to inform service design and planning. The NRDO collaborated with ERN Clinical Experts, patient representatives and wider Multidisciplinary Teams (MDT) stakeholders, to develop integrated care pathways for a number of the more common multi-systemic rare conditions. The development of rare disease care pathways supports the delivery of multidisciplinary integrated care and supports, and enhancing care coordination across specialist and community services. They support patient empowerment by increasing knowledge and awareness of rare conditions and confidence to navigate the health service. The development and implementation of rare disease care pathways is aligned with Ireland's National Rare Disease Plan, HSE Model of Care for Rare Diseases, Sláintecare and European Reference Networks (ERNs) objectives.

The introduction and development of genomic testing and access to clinical genetics will improve the rates of diagnosis for patients with rare diseases and reduce the 'diagnostic odyssey', or the extended length of time it can take for a patient to receive a diagnosis. A pilot study into the development of rare disease care pathways by the NRDO was published in 2022 in the 'Orphanet Journal of Rare Diseases'.[36] This work described the development, benefits and barriers to successful implementation and has been recognised as best practice by EURORDIS.

Furthermore, in 2020, the HSE established the National Clinical Programme for People with Disabilities. This Clinical Programme takes a rights-based approach, promoting and supporting compliance with the United Nations Convention on the Rights of People with Disability (UNCRPD) across the HSE sector and partner agencies, this includes all health and social care service for people of all ages.

36. "what Ireland does well for people living with a rare disease" 52% of respondents living with or supporting a person with a rare disease provided answers that could be broadly categorised as negative, with many providing responses similar to "Little/Nothing/Not a lot".

## Voice of People Living with a Rare Disease

This Rare Disease Strategy has been informed by the expert experience of PLWRD, their families and carers through both public consultation and feedback from the National Rare Disease Patient Forum.

**Key findings include:**

- **A diverse range of hospital-based specialist** services are required including Neurology, Rheumatology, Ophthalmology, Cardiology and Endocrinology with 68% of respondents attending two or more hospital-based services for treatment.
- **A diverse range of community-based services** are required including Physiotherapy, Occupational Therapy, Psychology, General Practitioner and Speech and Language Therapy.
- **Care pathways can be complex, fragmented and difficult to navigate** and, oftentimes, individuals can 'fall through the gaps' in care with 37.5% of respondents identifying 'Care Coordination' as one of the main challenges.

**PLWRD and their families and carers recommended the following areas for improvement:**

- establishment of care-coordinators in each region;
- multi-disciplinary clinics for specific rare diseases/ groups of diseases;
- ability to access services when required;
- establishment of care pathways that include all relevant disciplines;
- clear pathways for transition between acute and community services, as well as from paediatric to adult services;
- improved referral process to disease specialists; and
- accountability for when delays occur in treatment.

## The Role of General Practitioners

**The important role played by General Practitioners (GPs), primary and community care in the identification of people with, or suspected to have, a rare disease cannot be understated. Although GPs may not by definition be specialists in rare conditions, they are often the first point of contact for people and are crucial in the ongoing care of many people diagnosed with a rare disease. GPs are oftentimes the first to observe unusual symptoms and are important in coordinating early referrals to specialists or specialist services for diagnosis.**

The Irish College of General Practitioners (ICGP) has highlighted the difficulty many GPs have in identifying the right specialism to refer people with a suspected rare disease to, which can delay diagnosis and can mean that people are attending a range of healthcare providers before receiving a diagnosis.

Through increased access to continuing education and training, as well as signposting to specialised services, resources and ERNs, and ensuring appropriate care pathways are established, it is envisaged that GPs can, over time, be equipped with the necessary resources and support to adequately manage and refer those with a suspected rare disease to the appropriate community, acute and specialised services.

## Rehabilitation

In any system where care is effectively integrated, availability of rehabilitation is crucial. The WHO defines rehabilitation as "a set of interventions designed to optimize functioning and reduce disability in individuals with health conditions in interaction with their environment".[37]

**As outlined in the HSE Model of Care (MOC) for the provision of Specialist Rehabilitation Services in Ireland, rehabilitation is a dynamic and critical component of any modern health care systems. Rehabilitation improves health outcomes, reduces disability and improves quality of life. There is a significant and emerging body of international evidence to support the benefit and cost effectiveness of specialist rehabilitation services within a modern health service. The demand for rehabilitation services is growing and is anticipated to continue to grow with changes in populations and with the advances in health care and new interventions and technology".**[38]

37. Health Service Executive (2017). Model of Care for the Provision of Specialist Rehabilitation Services in Ireland. [online] Available at: https://www.hse.ie/eng/services/publications/clinical-strategy-and-programmes/model-of-care-for-specialist-rehab-medicine.pdf.
38. WHO (2024). Rehabilitation. [online] World Health Organisation. Available at: https://www.who.int/news-room/fact-sheets/detail/rehabilitation.

Rehabilitation is commonly understood as a recovery process for one-off conditions such as stroke or spinal cord injury, however, it also applies to any complex or progressive disability. Rehabilitation provides numerous benefits for people. For PLWRD, it enables individuals – whether children, adults or older people to be as independent as possible in daily activities and facilitates participation in education, work, recreation and meaningful life roles such as the taking care of a family.[39] Thus, rehabilitation should be seen as essential to the integrated ecosystem of care and support for PLWRD, with appropriate services developed to meet people's needs.

As such, the national approach to rare diseases must include rehabilitation needs. Rehabilitation services should be available to people of all ages, across service delivery contexts (in-patient, out-patient, community and residential) and across geographic regions to meet the needs of people living with rare diseases.

## Palliative care

Palliative care aims to enhance the quality of life by alleviating pain, managing symptoms, and providing support for both the person living with a rare disease and their family.

**Palliative care is an essential component in the treatment of rare diseases, particularly because many rare conditions are chronic, progressive, and lack effective curative treatments. These diseases often present complex symptoms that can be difficult to manage, and people may experience significant physical, emotional, and psychological distress.**

Palliative care for people with rare diseases takes a holistic approach that not only focuses on managing the medical symptoms but also addresses the emotional and social needs of people. This integrated care model ensures that individuals are supported in a comprehensive manner, helping people cope with the challenges of living with a rare disease. While palliative care does not exclusively refer to end of life care, the importance of high quality and adequately resourced end of life palliative care cannot be understated. PLWRD and their families deserve access to high quality end of life palliative care delivered in a compassionate and dignified manner, by suitably trained experts in an appropriate setting. In the context of rare diseases, where medical options may be limited, palliative care provides a compassionate and person-centred approach, ensuring that people living with a rare disease are treated with dignity and respect throughout their journey. As such, palliative care should be available for PLWRD, as is appropriate, to look after the physical, psychological and emotional demands of rare diseases.

39. WHO (2024). Rehabilitation. [online] World Health Organisation. Available at: https://www.who.int/news-room/fact-sheets/detail/rehabilitation.

## Respite

Short breaks are vital and valuable for individuals and families affected by a rare disease. It provides a break for the person with rare disease (who has additional needs due to a chronic health condition or a disability) and the family. It provides the person living with a rare disease with the opportunity to socialise; meet up with friends; or take the first steps towards independence, while also supporting family carers to balance responsibilities between work, family and personal needs and promote wellbeing for everyone in the family. While there are many types of short breaks, all forms should be flexible enough to meet the person's and family's needs at home and in the community.

**Short breaks should, therefore, be an essential part of the Model of Care for Rare Diseases and should offer support tailored to a person's specific needs, while also providing peace of mind to the family carer when out for a few hours, taking a short holiday or weekend break.**

Not only do short breaks support the physical and emotional needs of carers, but they also contributes to the overall quality of life for the person living with a rare disease, promoting a healthier caregiving environment that supports and brings renewed energy to the whole family.

## Mental Health and Wellbeing

Psychological and mental health supports should be imbedded with health and social care services supporting individuals with rare diseases, as well as for their families and caregivers. A rare disease diagnosis can often lead to significant emotional distress, anxiety, depression, and a sense of isolation for both the person diagnosed and their loved ones. Psychological and mental health supports can assist with addressing these emotional and mental health needs, offering crucial coping strategies and improving overall well-being.

For people living with a rare disease, psychological support can help individuals navigate the emotional impact of living with a chronic, often unpredictable condition. It provides a safe space to express fears, grief, and frustrations, which can alleviate feelings of hopelessness and improve mental resilience. For families and caregivers, stress can be experienced when managing supports for a person with a rare disease, navigating services and systems can be overwhelming, especially when they are unsure about how to provide care and support, or what the future holds. Psychological and mental health supports are important in ensuring that caregivers have the skills, coping strategies and supports necessary to maintain mental health and wellbeing.

Psychological and mental health supports are integral to the holistic care and support for people with rare disease and their families, and should be considered as an integral part of integrated care following a rare disease diagnosis and throughout a person's life journey. By ensuring that people living with a rare disease and their caregivers are supported and that they know where to access support, both emotionally and psychologically, we can contribute to better health outcomes and quality of life by fostering a supportive environment where people and caregivers can thrive despite the challenges posed by rare diseases.

## Health and Social Care Professionals

**Health and Social Care Professionals (HSCPs) play an important role in supporting people with the ongoing management of rare diseases across all healthcare settings. These roles are intrinsic to meeting the diverse needs of people with a rare disease in a multidisciplinary/interdisciplinary way that can have a significant impact on quality of life and health outcomes.**

HSCPs, such as Physiotherapists, Occupational Therapists, Speech and Language Therapists, Dietitians, Psychologists and Social Workers all play an important role in supporting people to manage life with a rare disease, placing the person's needs central when setting goals. For many people, their rare disease can affect daily living tasks, where skills, speech and cognition can be impaired. Health and Social Care Providers can provide targeted rehabilitation services to improve quality of life, help maintain independence and assist with adapting to physical or cognitive limitations or navigating the health and social care system and accessing necessary resources.

Only by adopting a multidisciplinary/interdisciplinary approach to providing streamlined integrated care that spans across all health and social care settings, can we ensure that all PLWRD have access to the right care and supports throughout life, which is delivered in the most appropriate setting and as close to home as possible.

> **"Research on treatment adherence that doesn't reflect the full range of treatment - both physical treatment and emotional issues and support required with a rare disease**
>
> – Response from Public Consultation

## Eligibility

Eligibility for health services in Ireland is primarily based on residency[40] and means that any person who is accepted by the HSE as being ordinarily resident in Ireland is entitled to have either full eligibility (medical card holders) or limited eligibility (all others) for health services. Full eligibility is determined primarily by reference to income limits.

**Eligibility for a Medical Card is primarily based on a financial assessment which is conducted by the HSE in accordance with the Health Act 1970 (as amended). The HSE assesses each medical card application on a qualifying financial threshold. This is the amount of money that an individual can earn a week and still qualify for a card. It is specific to the individual's own financial circumstances.**

Persons aged 70 or older are assessed under medical card income thresholds which are based on gross income. Persons under 70 are assessed under the general means tested medical card thresholds which are based on an applicant's household income after tax and the deduction of PRSI and the Universal Social Charge. Certain expenses are also taken into account.

40. The Health Service Executive normally regards a person as ordinarily resident in Ireland if they satisfy the HSE that they have been living in Ireland for at least a year or that it is their intention to remain in Ireland for a minimum period of one year.

The issue of granting medical or GP visit cards based on having a particular disease or illness was previously examined in 2014 by the HSE Expert Panel on Medical Need and Medical Card Eligibility. The Group concluded that it was not feasible, desirable, nor ethically justifiable to list medical conditions in priority order for medical card eligibility. In following the Expert Group's advice, a person's means remains the main qualifier for a medical card.

Against this background, every effort is made by the HSE, within the framework of the legislation, to support applicants in applying for a medical card and GP visit card and, in particular, to take full account of the difficult circumstances in the case of applicants who may be in excess of the income guidelines.

**The HSE may exercise discretion and grant a medical card or GP visit card, even though an applicant exceeds the income threshold where they face difficult financial circumstances, such as extra costs arising from an illness. Social and medical issues are also considered when determining whether undue hardship exists for an individual accessing GP or other medical services. In circumstances where an applicant is over the income limit for a medical card, they may be eligible for a GP Visit Card. A GP visit card allows the holder to visit a participating family doctor (GP) without charge.**

All children under 8 years of age living in Ireland are entitled to a GP visit card which covers free GP visits; assessments at age 2 and 5; out-of-hours urgent GP care; and care for children with asthma.

In context of the recommendation now being made, the Department and the HSE will have further engagement to increase awareness regarding the range and type of rare diseases and to further highlight, for the benefit of applicants, the existing capacity to specifically include relevant medical expenses for consideration within the medical card assessment process administered by the HSE. The aim of this engagement will be to consider how applicants can be informed of and supported to identify and include relevant expenses, and in doing so, to ensure that the increased financial cost of a rare disease diagnosis is fully recognised.

## Challenges

As highlighted through the public consultation, frequent healthcare appointments, often requiring visits to multiple specialists, disrupts daily life and places a significant demand on family resources and time. The financial strain associated with travel to frequent appointments, periods of in-patient care, and the need for specialised equipment can be overwhelming and place a significant strain on people with a rare disease and their families. Additionally, the psychosocial impact, including feelings of isolation, anxiety and uncertainty adds to the challenges PLWRD face. These challenges are not limited to those with a definitive diagnosis, but they also affect people that have an undiagnosed or undefined rare disease.

**To address these challenges this Strategy aims to highlight the need to improve needs-based access to coordinated multidisciplinary/interdisciplinary care and support beyond what is currently available to ensure future services are being developed in an equitable, integrated, person and family centric manner.**

Delivering real change for PLWRD will require close collaboration between primary and community care providers, ERNs and rare disease centres of expertise, to ensure that people with a rare disease receive a definitive diagnosis as early as possible, are referred promptly to appropriate acute and specialist services, and have access to long-term follow-up care and monitoring to manage disease progression, treatments and any complications that may arise.

Reform of the health service into six Health Regions which are responsible for their respective populations has the potential to improve services for people living with a rare disease in several ways such as:

- **Improved access to local services** to reduce travel time and improve access to general healthcare services and specialised care and support.
- **Facilitating better communication and coordination of care and support** between different health and social care providers, leading to enhanced collaboration to meet differing levels of need.
- **Improving pathways for diagnosis and referral** to specialist services, as appropriate, leading to more timely and accurate diagnoses.
- **The implementation and support for models of care and integrated pathways** to ensure continuous care and support that accounts for the unique nature of individual rare diseases.
- **Mapping service needs with a life course approach** where a journey begins in childhood or adulthood, or where there is an indication of a rare disease, and showing how services should align, be integrated and seamless throughout life.

**The NRDO must continue to play an important role in identifying the national frameworks required to support the delivery of care across the six Health Regions for those with a rare disease. A combination of national approach led by the NRDO, in collaboration with the new Health Regions, will ensure that there is no disparity in healthcare access for people living with a rare disease across the regions and ensure the equitable implementation of this Strategy nationally.**

While it is recognised that the operationalisation of many initiatives will be driven by the Health Regions, the NRDO and HSE Centre *must* define the vision and direction for the future development of rare disease services and monitor and provide oversight of implementation across the regions.

## Recommendation 4

**The Steering Group recommends that future services should be developed in an equitable, integrated, person and family centric manner aligned to Sláintecare and the Model of Care for Rare Diseases, specifically recommending:**

**A.** That, as part of the application process for medical card assessment, the financial cost of a rare disease diagnosis is recognised and medical expenses arising are taken into consideration.

**B.** The ongoing development and implementation of rare disease integrated care pathways including those for people with an undiagnosed rare diseases or Syndrome without a Name (SWAN).

**C.** The provision of integrated, coordinated, and multidisciplinary care and support within the community across all regions in line with individual needs, with timely referral and access to specialist care and support. This includes the development and greater utilisation of rare disease networks nationally and in other EU Member States and should be supported by understanding population-based needs.

**D.** Full implementation of the Model of Care for Transition from Childhood to Adult Healthcare Providers in Rare Diseases, the National Framework for Transition of Care and the development of associated care pathways to ensure appropriate and flexible transition arrangements for children, adolescents and young people, adults and older persons.

**E.** Access to necessary wraparound support services for PLWRD and their families throughout the life course, inclusive of rehabilitation, respite, palliative care, psychological and mental health supports, and all associated ancillary services in line with individual need.

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# Data, Health Information Systems & Registries

Availability and ease of access to information are crucial for improving the lived experience of people living with a rare disease (PLWRD). Better information-sharing practices enhance a person's outcomes, facilitate collaboration, inform policy, support research, and empower people with rare disease.

The availability of accurate rare disease data will also serve as an essential tool for service planning, delivery, evaluation and sustainability — enabling the development of guidelines and standards and assessing how Irish health services compare with international benchmarks. Moreover, robust data is critical to European Reference Networks (ERNs) and for identifying PLWRD to participate in research and clinical trials.

> **The ability to link all your information with different consultants is a challenge. All information can't be shared among different consultants meaning additional bloods have to be taken and more wait time**
>
> – Response from Public Consultation

## Legislative and policy developments

The European Health Data Space (EHDS), for example, seeks to establish a secure framework for sharing health data across EU member states, which can be transformative for PLWRD who may need specialized care abroad. The EHDS will also facilitate research by improving data availability and interoperability. Meanwhile, at a national level, the proposed **Health Information Bill** is expected to provide a modern, robust legislative framework for the governance, sharing, and protection of health information. Its alignment with the EHDS and focus on data security will be critical in enabling secure, data-driven healthcare improvements that benefit all people, including PLWRD.

**In Ireland, the Digital for Care: A Digital Health Framework for Ireland 2024–2030 outlines plans to digitally transform healthcare and improve a person's access. Its goals include ensuring that, by 2030, all individuals will have access to their own digital health records, healthcare professionals can securely view a person's information in real time, and people can receive care closer to home without needing to carry physical notes between providers.**

## Orphanet and ORPHAcodes

Orphanet is a European database that provides information on rare diseases, expert centres, healthcare professionals, patient organisations, orphan drugs, clinical trials, registries, and research projects.

**Since 2015, Ireland has been a full member of the Orphanet consortium. The National Rare Diseases Office (NRDO), acting as Orphanet Ireland, manages Irish data on Orphanet.**

Recognising the importance of mapping national Centres of Expertise, the NRDO has validated and mapped over 113 multidisciplinary centres in accordance with Orphanet's international criteria.

ORPHAcodes, Orphanet's nomenclature for rare diseases, is widely considered the optimal clinical coding system for rare diseases in Europe, as recommended by the European Commission Expert Group on Rare Diseases (CEGRD, 2014). One of the ERNs' key priorities is the development of rare disease registries, which requires the use of interoperable ORPHAcodes. This use is also mandated by the European Commission's annual ERN audit process (AMEQUIS).

## Databases and Registries

A healthcare *registry* is a systematic collection of patient data—such as diagnoses, treatments, and outcomes—designed to support surveillance, service planning, research, and treatment improvements. High-quality registries can help identify patterns of disease, measure the effectiveness of interventions, and plan more responsive services. In Ireland, certain registries (e.g., the National Cancer Registry) exemplify how comprehensive data collection can guide evidence-based policy and practice for the overall benefit of people. Ireland currently lacks a comprehensive national registry for rare diseases, and its health information systems do not adequately capture rare disease data.

At present, Orphanet hosts data on 16 patient registries and one network of registries. Ireland participates in the EUROCAT Registry (focused on surveillance of congenital anomalies) and the RARECAREnet database (which examines the epidemiology of rare cancers in Europe). Additionally, the EU Joint Research Centre (EU-JRC) has created a European Platform on Rare Diseases Registration to host registries for the 24 European Reference Networks (ERNs). Although there are several individual rare disease registries in operation in Ireland through various ERNs, they differ substantially in both development and resourcing, and no central mechanism currently exists to harness or analyse their data at the national level.

**In Ireland, several registries (e.g., the National Cancer Registry) have been established or empowered through legislation, which can mandate data collection and enforce data standards. Legislation provides a clear legal framework for collecting, storing, and using health data. This brings advantages such as: clarity and accountability, consistency and quality, and enhanced privacy and security.**

However, legislative processes can be time-consuming and may introduce complexities around patient consent or data sharing across institutions. Ultimately, legislation serves as both a shield and a framework: it protects patient data while compiling the consistent, high-quality data collection necessary for effective surveillance, research, and service improvements. The National Cancer Registry provides a model for how a high-quality registry can be developed and supported by legislation, and which should be given detailed consideration in the design and development of a rare disease registry.

Under the National Rare Disease Plan for Ireland 2014–2018 guidelines were created to standardise the coding and recording of rare diseases and to develop a national rare disease registry within Irish health data systems. Following national consultation, these guidelines were approved and incorporated into the HSE's 2019 Model of Care for Rare Diseases. These guidelines, the Digital for Care framework, EHDS requirements and examples of good practice should inform the development of a National Rare Disease Registry.

## Key Considerations for a New National Rare Disease Registry

When developing a new National Rare Disease Registry, a practical and pragmatic approach is essential. Existing ERN registries and databases should be leveraged to avoid duplication. In particular, the registry should:

- **Comply with all relevant legislation and national standards** (e.g., the Digital for Care Framework and the HSE *Digital for Care* roadmap).
- **Encompass all rare diseases** (unless a relevant registry is already established, e.g. National Cancer Registry including rare cancers) according to a commonly agreed EU definition and the recommended common data elements from the EU Joint Research Centre.
- **Utilise ORPHAcodes** as the European standard coding terminology, ensuring international alignment, facilitating data sharing and reporting, maximising support for service evaluation and planning, as well as ensuring that PLWRD can be identified by ORPHAcode and offered opportunities to engage in clinical research and trials.
- **Leverage and support existing ERN registries** and databases, further advancing each ERN's work and avoiding duplication.
- **Meet regional and national service planning needs**, ensuring a mixed methods approach to the data collected and is both actionable and beneficial for long-term health and social care improvements.
- **Provide, for instance, regular epidemiological data** on rare disease incidence, prevalence, distribution across country, etc.

> **Lack of a registry/data base/ERN access is a challenge for PLWRD**
>
> – Response from Public Consultation

By aligning national efforts with international standards, adopting robust legislative frameworks, and prioritising PLWRD in new digital health initiatives, Ireland can significantly improve rare disease data collection and service provision.

## Electronic Health Records and Coding Terminologies

**Electronic Health Records (EHRs) are digital versions of a patient's paper-based health history. They contain information such as medical history, diagnoses, medications, treatment plans, immunisation dates, allergies, radiology images, and laboratory test results. By enabling real-time and secure access to patient data, EHRs facilitate more coordinated and efficient care.**

Ireland does not currently have a unified health information system and the recommended coding nomenclature, ORPHAcodes, are currently not implemented in existing health information systems. Consequently, PLWRD often carry personal medical records across multiple healthcare settings and must repeatedly recount their health history. While these issues affect many patients, the complexity of rare diseases can make such challenges particularly acute.

**Over the next five to ten years, in alignment with the Digital for Care framework and roadmap, EHRs will be introduced across the health service, aiming for a single digital health record per patient. These EHRs are the front facing systems that healthcare professionals and patients will interact with, they are supported in the background by other systems that provide standardised information and unique codes for individual diseases, diagnoses and other medical information, known as clinical terminologies.**

The HSE plans to adopt the *"Systematized Nomenclature of Medicine—Clinical Terms"* (SNOMED CT) as the clinical terminology to support EHRs, this will allow different healthcare facilities to share standardised information seamlessly. While SNOMED CT will provide multilingual clinical terminology and standardised codes for diseases, symptoms, procedures, and more, it does not itself serve as a dedicated rare disease code set. This means that for PLWRD, their information will first be assigned a SNOMED CT code, which is then subsequently mapped to corresponding ORPHAcodes. This mapping should occur automatically and seamlessly within the system, thanks to collaboration between INSERM (developer of ORPHAcodes) and SNOMED International.

Once the HSE implements EHRs that use SNOMED CT, these mappings will make it possible to extract ORPHAcode data as needed. While relying on SNOMED CT in the first instance with subsequent mapping to ORPHAcodes is not an ideal permanent solution from a rare disease perspective, it does represent a pragmatic improvement over current practices and allows Ireland's health system to remain aligned with global standards for clinical terminology. However, it is essential that people with rare diseases are not "left behind" and that their specific needs are prioritised as these digital initiatives, including the unique patient identifier which facilitates linkage across the public system, move forward.

The development of a Bioinformatics and Genetic Data Infrastructure Roadmap as part of implementing the Genomics Strategy will see genomic data being available from the introduction of genomic testing aligned with the development of EHRs. The Roadmap will also set out access to this data by researchers.

## Recommendation 5

### The Steering Group recommends:

**A.** The establishment of a national rare disease registry, underpinned by legislation, as necessary, 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 surveillance and data reporting is required across health and social care sectors.

**B.** A core project team, with the necessary supports, should be established 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.

**C.** The Steering Group recommends that the longer-term vision for the development of EHRs and associated IT systems should include the full integration of ORPHAcodes, as the gold standard nomenclature for rare diseases. A stepwise and an incremental approach to the integration of ORPHAcodes may be required to achieve this. In the interim, the capabilities of existing IT systems (i.e. SNOMed CT) should be leveraged to allow relevant data and information to be extracted for utilisation within a registry, surveillance, and service planning.

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# International Cooperation and European Reference Networks

International cooperation is crucial for rare diseases to ensure that Ireland, and other countries, can address the challenges posed by rare diseases in an equitable way that leads to better outcomes for PLWRD both in Ireland and internationally. Collaboration on a North-South, EU, and Global level is important to drive innovation, enable early diagnosis and improve management, treatments, and outcomes for PLWRD.

As many rare diseases often affect small patient populations, the research and development of treatments can be limited if countries work on it in isolation. By sharing knowledge, data, reports, expertise and resources across borders, countries can pool their resources and increase the chances of developing effective therapies and sharing experiences and best practices with others, such as ERN consensus care guidelines.

Developing Ireland's profile on the international stage is also an important factor in both attracting and securing research funding and in developing appropriate relationships with industry. While this collaboration can have real benefits with regard to opening up opportunities for PLWRD to participate in international clinical trials, it can also strengthen advocacy for rare diseases at an EU-level, helping raise awareness and pushing for policies that support PLWRD. While this chapter focuses heavily on the international cooperation of European Reference Networks and the potential to build on this, it is equally important that international cooperation is undertaken across all domains for the benefit of PLWRD in Ireland. Health Technology Assessments (HTA), particularly with regard to screening, diagnostics and medicines is one example of an area where international collaboration could be leveraged to the greatest extent possible.

The EU HTA regulations (HTAR) is designed to improve the availability, for EU patients, of innovative technologies in the area of health, such as medicines and certain medical devices. The regulation provides a transparent and inclusive framework by establishing a Coordination Group of HTA national or regional authorities. This Regulation replaces the previous voluntary networks and structures with a permanent framework for joint work. The HTAR will also reduce duplication of efforts for national HTA authorities and industry, facilitate business predictability and ensure the long-term sustainability of EU HTA cooperation. HIQA is an active participant in HTAR-related work.

## An All-Island Approach

Collaboration across the island of Ireland and establishing an all-island approach to rare diseases will benefit PLWRD across the entire island. Both Ireland and Northern Ireland face similar challenges in treating rare diseases, with limited resources and comparatively small populations. By working together, both Ireland and Northern Ireland can pool expertise, share best practices, and overcome the challenges presented by rare diseases more effectively. By working on a cross-border basis, both jurisdictions can explore areas for collaboration, including the possibility of all-island networks of care, that can increase access to essential services for PLWRD, as well as ensuring equity of access for all PLWRD on the island of Ireland and for families who travel between both jurisdictions for care.

## European Reference Networks

**European Reference Networks (ERNs) are, as the name suggests, networks of healthcare providers across Europe that collaborate and share knowledge and expertise on rare diseases. There are 24 ERNs that cover the main clusters of rare, complex, and low-prevalence diseases. The ERNs include over 1,600 specialised centres located in 382 hospitals across 27 EU Member States and Norway.**

Since January 2022, Ireland has become a member to 18 of the 24 ERNs, which has further enhanced and expanded patients' rights to appropriate assessment and treatment. This is a collaboration of over 40 Irish centres of expertise, across five academic teaching hospitals, three universities and the NRDO as the ERN co-ordination hub.

ERN membership facilitates mobility of expertise, the sharing of information, knowledge and best practice and fosters developments in the diagnosis, research and multidisciplinary treatment of rare diseases. This is a significant achievement for people in Ireland affected by rare diseases in cases of rare or low-prevalence diseases which affect the daily lives of around 6% of the Irish population, given Ireland's small population size and capacity.

Through the ERNs, a doctor can now access existing expert knowledge from another country on a patient's disease. To review a patient's diagnosis and treatment, ERN coordinators have the ability convene 'virtual' advisory panels of medical specialists across different disciplines, using a dedicated IT platform: Client Patient Management System (CPMS) and telemedicine tools.

The work of ERNs is diverse and there are several key enablers which seek to advance the overall aim of ERNs in facilitating discussion and cross-border collaboration on complex or rare diseases and conditions that require highly specialised treatment. These include virtual expert discussions, the development of clinical practice guidelines, training and education activities, and the development of ERN-specific registries.

- **Virtual Expert Discussions:** ERNs provide direct guidance to clinicians through cross-border discussions of rare clinical cases, using a dedicated, secure, and online IT platform, known as the Clinical Patient Management System (CPMS). CPMS, which was launched in 2017, facilitates collaboration between healthcare professionals across Europe and supports the diagnosis and treatment of rare, low-prevalence, and complex diseases.
- **Clinical Practice Guidelines:** ERNs also have responsibility for developing, updating and appraising Clinical Practice Guidelines. This helps to bring forward evidence based and unbiased information on rare diseases and to help standardise best practices. Ultimately, this aims to ensure high quality and effective care for rare disease patients.
- **Education and Training:** ERNs are also dedicated to improving the quality of care provided to rare disease patients through the development and sharing of educational resources and training opportunities for healthcare professionals. These education and training resources are developed by individual ERNs and made available through their respective websites.
- **Registries:** Through the recognition that research on rare diseases can only take place if data is available and that registries are the key repositories of health data, ERN-specific registries seek to enable the identification of contemporary cohorts of patients with rare diseases for clinical research, enable monitoring of diagnostic and treatment procedures across healthcare providers (HCPs), and enable research into the diagnosis, treatment and outcome or prognosis of rare diseases.

ERN sites in Ireland are essential pillars in the field of rare diseases, fostering collaboration, knowledge exchange, and innovation that benefit both PLWRD in Ireland and the wider rare disease community across Europe.

**The National Rare Diseases Office in the HSE, as the 'coordination hub', provides information to stakeholders and PLWRD regarding the location of national expert sites and linked ERNs.**

## Challenges faced currently

European Reference Networks (ERNs) were founded on the principle that many rare disease issues are pan-European, and any single Member State cannot solve them alone. There are a number of significant challenges faced by ERN sites in Ireland, which are not uniquely Irish, and which are faced by ERN sites across the EU. These include insufficient funding and resources being allocated to ERNs and a lack of ERN integration into national healthcare systems.

Integrating ERNs into the national healthcare system of EU Member States has been identified by the EU as a key challenge that is impacting on their effectiveness across the EU. To address this challenge, the European Commission, in 2024, launched a new EU4Health Joint Action, JARDIN, to support the integration of European Reference Networks (ERNs) into national health systems. This three-year project is designed to enhance the impact of the ERNs, and improve integration into healthcare systems and includes work packages on:

- National governance and quality assurance models
- National care pathways and ERN referral systems
- National reference networks (NRN) and undiagnosed disease programmes or equivalent strategies interlinked with ERNs
- Data management and national support options for ERN-HCPs

**Ireland is playing a leading role in this Joint Action. The Department of Health nominated the HSE as the Joint Action's Competent Authority and on behalf of the HSE, the NRDO is the coordination centre.**

The HSE through the NRDO is co-leading on Work Package 6: National care pathways and ERN referral systems and is a key partner in other work packages under this Joint Action. However, sustained funding is also required to support and resource ERNs particularly in the areas of data management and care co-ordination. As part of this Strategy, it is recommended that the HSE, working with ERN sites, develop and implement a roadmap for the integration and operationalisation of ERNs into the national healthcare system, to include the minimum or necessary operating/staffing model for a functional ERN.

By working to support ERN sites, and ensuring that they are fully resourced, Ireland can play a leading role at a European level to highlight the impact that ERNs can have in the further development of services for patients and families with a rare disease diagnosis in Ireland.

## Recommendation 6

**The Steering Group recognises that international and cross-border cooperation, including the integration of the European Reference Networks (ERNs) into the Irish healthcare system are essential to ensure access to expertise and the delivery of high-quality care and support to people living with a rare disease and recommends that:**

**A.** International and cross border collaboration is not limited to ERN activity but rather, is actively promoted across the board, inclusive of policy development, clinical practice, training and education, and research

**B.** The HSE establish an ERN Integration Working Group of key stakeholders to develop and implement a roadmap for the integration and operationalisation of ERNs into the national healthcare system.

**C.** The NRDO in conjunction with the ERN Clinical Leads determine the necessary operating/staffing model for a functional ERN, such that they can deliver across all elements of the Strategy, including data and research.

**D.** ERNs are fully supported and resourced to ensure effective and sustainable integration into the national healthcare system.

**E.** To ensure the long-term sustainability of ERNs, the DOH and the HSE take into account the recommendations and outcomes and outputs from the EU4Health Joint Action on the Integration of ERNs into National Healthcare Systems (JARDIN) when available.

**F.** The Department of Health and HSE actively encourage and support applications to join the remaining ERNs at the earliest available opportunity.

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# Innovation and Rare Disease Research

## Importance of rare diseases research within our healthcare system

Rare Diseases research is crucial for advancing our understanding and the treatment of conditions that often go unnoticed due to their low prevalence. The role of rare disease research is to engage and involve PLWRD, to develop a better understanding of the mechanisms of disease enabling early diagnosis, earlier treatment and overall better health outcomes for people. Greater knowledge of rare diseases is important for the development of novel therapeutics, treatment strategies and facilitation of more responsive and appropriate services for PLWRD, their families and circle of support.

**Rare Disease research supports the recruitment, retention and motivation of healthcare personnel who will drive the development of quality services. Research is multi-disciplinary and engages professionals from a variety of backgrounds, from basic science through to translational, clinical and health services research. It requires healthcare system infrastructure and supports appropriate within different environments and contexts.**

## The Environment for Rare Disease Research

Substantial advancements have been made to embed research within Ireland's healthcare system. The Department of Health has ensured that research is a key aspect of strategies and frameworks, for example the development of the National Mental Health Research Strategy as part of the overall national mental health policy *'Sharing the Vision – A Mental Health Policy for Everyone 2020–2030'* supported by key partners such as the Health Research Board (HRB).

In line with the recent commitment in the Programme for Government, the Department also facilitates the work of the National Clinical Trials Oversight Group (NCTOG) which has been tasked with increasing clinical trial activity in Ireland.

The development of a foundational framework to support research – *'HSE National Framework for the Governance, Management and Support of Research 2021'* and the progression of the Sláintecare regional reform programme has embed research into wider HSE structures.

## Funding for Rare Disease Research

**In Ireland, several organisations have their own specific roles in contributing towards the advancement of rare diseases research. Organisations such as the Health Research Board (HRB), Health Research Charities Ireland (HRCI) and Taighde Éireann (Research Ireland) will need to play a central role in funding and supporting research initiatives related to rare diseases through the provision of grant schemes, co-funding of research projects and programmes and partnerships that prioritise rare diseases, direct investment in clinical research, population health and health services and investment in research infrastructure.**

The HRB has funded specific initiatives to ensure Ireland is well positioned to participate in, and conduct, research to tackle rare diseases. The HRB have invested €17 million in research over the past 10 years on behalf of the Department of Health. These initiatives aim to deliver better quality care and treatment for people with rare diseases.

Research in Ireland is further supported by the involvement of patient advocacy groups such as Health Research Charities Ireland (HRCI), Irish Platform for Patients' Organisations, Science and Industry (IPPOSI), and Rare Diseases Ireland (RDI) who, through ensuring that research is aligned with patient priorities, advocate and provide a voice for the needs of people with rare diseases and ensure that their perspectives are included in research agendas while also promoting civil society involvement in the design and operation of research and clinical trials. Some rare disease representative organisations also fund and develop their own disease specific research.

Finally, industry partners, including pharmaceutical and biotechnology companies play an integral role in the development of new therapies for rare diseases through investing in research projects and conducting clinical trials to test potential therapies for safety and efficacy and through their collaborations with other partners and initiatives.

Improved engagement, collaboration, and partnership both nationally and internationally with industry, Government, representative organisations, healthcare professionals and academic partners will help to drive strong research and innovation for all aspects of living with rare diseases. As many rare diseases often affect small populations, the research and development of treatments can be limited if countries were to work in isolation. Thus, the field of rare diseases research, due to low prevalence and scarcity of expertise and practice, makes research collaboration essential. International collaboration connects healthcare professionals and researchers and increases the number of patients available for each study which in turn potentially accelerates the development of new diagnostics and treatments. By sharing knowledge, data, and resources across borders, countries can pool resources and increase the chances of developing effective therapies and sharing experiences and best practices with others.

The All-Ireland Rare Disease Interdisciplinary Research Network (RAiN), for example, was established in 2023 with funding from the Department of the Taoiseach's Shared Island strand of the Irish Research Council's 'New Foundations' awards. This is a person-centred project which aims to directly impact policy through the development of Patient Report Outcome Measures (PROMs), with a focus on children with rare diseases and their specific care needs across the island of Ireland. RAiN will also feed into a global rare disease research project, enhancing the profile of our all-island rare disease research.

### Why is rare disease research important for PLWRD:

Meaningful research on rare diseases can give hope for the future to people living with rare diseases and their families. The opposite is a feeling of hopelessness which some patients and families often feel.

Basic research which may include translational research studies that can lead to the development of new treatments are very important as is research which aims to improve quality of life.

Ireland must make much bigger efforts with research on rare diseases and for me as a patient family member there should be dedicated rare disease research grant opportunities in Ireland. No rare disease research proposal should be discriminated against based on disease prevalence.

Ireland is currently a smaller player in rare disease research versus other countries that have dedicated rare disease grant opportunities. New rare disease grant opportunities in Ireland should allow for some of the funds to go outside of Ireland as expertise in many cases may be elsewhere, this would be welcome as it may maximise impact. Ideally there should be a co-applicant based in Ireland which would give strong opportunities to expand rare disease research and expertise into Ireland. International collaboration is often key to make an impact on rare diseases. Dedicated rare disease research grants in Ireland will create sustainability, growth and continuity.

When patients and families affected by rare diseases know that meaningful research is moving along on their disease this can be comforting to many as it can mean that there is hope for a brighter future.

Alan Finglas – Member Rare Disease Steering Group

## Advances in rare diseases research in Ireland to date

### Rare Diseases Clinical Trials Network (RDCTN) 2022

- Following an open and competitive call, a five-year investment of €1 million, funded through the HRB, was awarded to the RDCTN in April 2022. The objective of the Clinical Trials Network in Rare Diseases is to act as a collaborative hub for trials in rare diseases, support the conduct of trials in rare diseases and increase the opportunities for patients to access high-quality clinical trials. The RDCTN continues to work with the European Reference Networks for Rare Diseases to increase the availability of clinical trials for rare disease patients.

National Rare Disease Strategy 2025-2030

### Research and Innovation Catalyst Awards (RDCat) 2023

- The Rare Disease Research and Innovation Catalyst award (RDCat), funded through the HRB, is aimed at boosting rare diseases research capacity in Ireland through a once-off award. This award was intended to provide targeted research support for clinical sites in Ireland as they transition to active members of European Reference Networks, and to maximise preparedness for the planned European Rare Diseases Partnership. This award brings together all relevant stakeholders to ensure that Ireland is better placed to engage in rare disease research activities. A three-year once off investment of €3 million was awarded to the Rare Disease Research Catalyst Consortium in December 2023. The consortium is hosted by UCD and is made up of a group of Irish-based healthcare workers, researchers and patients. This project aims to:
  - increase the influence of rare diseases patients in rare diseases research and policy,
  - increase Irish engagement with national and international networks including the RDCTN and ERNs,
  - build rare diseases research activity and excellence in Ireland, enabling engagement with the European Rare Disease Partnership and
  - open and extend communications within Ireland and with international stakeholders to further these aims.

### Participation in the European Rare Diseases Alliance (ERDERA) 2024

- In January 2024, the European Commission approved €56 million for the European Rare Diseases Research Alliance (ERDERA) for seven years. Throughout 2023 the HRB actively engaged with both the rare disease research community in Ireland and the NRDO to participate in an application for the ERDERA proposal. Ireland will now participate in ERDERA working alongside 171 international partners from 35 countries, including 38 funders, 115 research performing organisations, 12 patients' organisations, 3 research infrastructures, and 27 private for-profit partners. ERDERA aims to deliver a robust rare diseases research ecosystem supporting patient need-led research, developing new diagnostic methods and pathways, spearheading the digital transformational change, and connecting the dots between care, patient data and research, while ensuring strong alignment of strategies in rare disease research across countries and regions.

### European and International Cooperation

- Rare diseases research benefits strongly from coordination at a European and international level to achieve scale and to overcome fragmentation, leading to better use of data and resources, faster scientific progress and competitiveness, and deliver better patient care. In recognition of this, Ireland participates in several international initiatives including the International Consortium in Personalised Medicine (ICPerMed), ERANET Cofund (ERAPerMed), the European Partnership for Personalised Medicine (EPPerMed), and the European Rare Diseases Research Alliance (ERDERA). The HRB, as the main health funding body supports new and established researchers to relevant calls for proposals and funding opportunities through Joint Transnational Calls associated with these European initiatives.

### Genomic Research Infrastructure

- Genomic technologies and big data analytics have revolutionised the understanding and diagnosis of rare diseases. As part of the European Union's Digital Transformation of Health and Care and in line with the European Health Data Space, the European Commission launched the 1+ Million Genomes initiative, which Ireland officially joined in 2021. Arising from the 1+MG initiatives the Genome of Europe (GoE) project, to establish a unique pan-European reference database, representing Europe's diverse populations, integrating existing genomic and new genomic datasets of distinct national populations is being established. To enable the 1+MG project, the Genomic Data Infrastructure (GDI) consortium of 20 EU Member States was established with the aim to enable access to genomic data across Europe through a secure data infrastructure.

## Rare Disease Clinical Trials

For many people living with a rare disease, participation in a clinical trial is a potential method of accessing innovative treatments. Whilst participation in research is entirely voluntary, subject to informed consent and the nature of trials involves random allocation to treatment with no guarantee of access to the novel intervention, ethically conducted research is of prime importance to future care improvements.

**Ireland became a member of the European Clinical Research Infrastructure Network (ECRIN) in 2018 which enables increased access for Irish patients to multi-national clinical trials and makes it easier for Irish researchers to extend their own trials internationally and help improve patient safety and quality of care. ECRIN, which represents a population of 300 million, also gives access to much larger trials than could be delivered in Ireland and provides real opportunities for trials in rare conditions.**

Rare disease research is crucial to advancing understanding of conditions which often go unnoticed. Research into rare diseases must span fundamental, translational, clinical, population health and health services areas to address existing gaps aligned to the needs of PLWRD. Research also needs to inform evidence-based policy across all settings and systems, extending beyond acute care to incorporate areas such as disability, social care, mental health and education.

> **"Access to clinical trials should be fast tracked, there is no support available right now. We email the teams abroad ourselves**
>
> – Response from Public Consultation

Research should create tangible and positive changes at individual, societal, service, and policy levels. This framework recommends that engagement takes place between stakeholders, policymakers and funders to support collaboration between researchers, senior health service managers, and policymakers in rare disease research in order to facilitate proactive and timely research grants and calls to increase the evidence base for national-level policymaking and service delivery.

There are high-level priorities identified within this framework which funding organisations and agencies can use to determine their own specific rare disease research objectives and formulate research questions aligned with these areas.

**Some of the key questions established in this framework include:**

- How to evaluate and map the existing rare disease research being conducted in Ireland and through Ireland's involvement in international initiatives and identify potential opportunities for growth, development and collaboration.
- Taking action to integrate health economics assessments into rare disease research to support sustainable and cost-effective healthcare planning.
- Aligning national research initiatives with Horizon Europe and other international funding opportunities such as ERDERA.

## Empowering European Reference Network (ERN) Sites and promoting international Collaboration

Given the small patient populations associated with rare diseases, international collaboration is vital to achieving meaningful research outcomes in adequately designed and statistically powered studies.

**The European Reference Networks (ERNs) serve as a model for effective collaboration. Established to connect healthcare providers across Europe, ERNs have made significant contributions to data sharing and joint research and are well-positioned to play a central role in rare disease diagnostics and treatment.**

However, to fully leverage the potential of ERN sites in Ireland to maximise their research potential and properly support international collaboration the necessary supports and enablers should be considered.

This includes:

- Research funders should engage on the **optimum model for investing in capacity-building** to ensure that ERN sites have research capabilities to compliment the clinical expertise;
- **Enhancing data systems** to facilitate the collection, sharing, and analysis of rare disease data, including genomic data;
- **Introducing and developing infrastructure for genomic testing** to facilitate access to and sharing of genomic data for clinical and research purposes;
- **Supports to strengthen participation** in European and global research consortia; and
- **Aligning national research initiatives with Horizon Europe** and other international funding opportunities such as ERDERA

## Enhancing Clinical Research Capacity

One of the fundamental barriers to rare disease research is the limited capacity of clinicians to engage in research. Many healthcare professionals face significant time and resource constraints, making it difficult to participate in research initiatives. Strengthening the link between research and the health and social care system is essential to overcoming this challenge.

**This barrier could be unlocked by:**

- **Ensuring workforce planning considers research and innovation** in rare diseases research beyond acute care to areas such as quality of life, survivorship and palliative care,
- **Ensuring protected research time for clinicians**, with a research interest, to engage in research activities without impacting their service delivery responsibilities.
- **Providing funding and infrastructure support** to enable clinicians with a research interest to lead and participate in projects that directly benefit patient care.
- **Expanding research fellowships and placements** to develop a workforce skilled in both clinical care and research.

## Access to high quality clinical trials

Access to clinical trials is an important vehicle for PLWRD to potentially access novel and innovative therapies. Findability of patients for these clinical trials is a significant challenge which could be overcome through the development of a rare disease registry to allow suitable participants to be identified and offered the opportunity to participate in trials relevant to them. Rare disease clinical trials often impose higher demands on participants than prevalent disease trials. The number of experienced trial sites is limited due to low levels of expertise in rare diseases across health systems, often requiring PLWRD their families and carers to travel longer distances to participate in clinical research.

Thus, rare diseases clinical trials require a patient centred approach with effective participation support strategies and sufficient resources at sites to succeed. Involving PLWRD in clinical trial design is key to ensuring more relevant studies that incorporate the lived experience and quality of life measures, with streamlined recruitment strategies to ensure success. Work underway in ERDERA and the RDCTN aims to ensure that Ireland is “clinical trial ready” with sites that prioritise the needs of PLWRD with tailored support, logistical solutions and strong partnerships.

## Innovation and Readiness

**Research and innovation** are profoundly transforming healthcare systems worldwide, leading to improvements in mortality, morbidity, and quality of life. This is particularly so in the case of rare diseases, where advances in rare disease research and innovation have led to the development of new diagnostic tools, devices, treatments and therapies to address the unmet needs of often small, rare disease populations, transforming the lives of those affected.

**Early diagnosis** through genetic screening has reduced infant mortality associated with rare diseases and novel treatments and therapies have significantly improved survival rates for previously fatal conditions. Innovations in genomics and biotechnology have enabled treatments tailored to individual genetic profiles, improving efficacy and reducing side effects, while disease-modifying therapies have reduced the progression and complications of chronic rare diseases, enabling better physical and mental health outcomes. Furthermore, innovations in the development of patient-centred support services such as telemedicine and virtual clinics, integrated care models, and care coordinators can help individuals navigate the healthcare system, ensuring they get the right treatments as close to home as possible, while adapting to the individual needs of people living with a rare disease.

**Treatment advances**, diagnostic innovation, and device and digital developments have led to advances in earlier diagnosis, providing hope for conditions previously deemed untreatable, facilitating coordinated care and improving long-term outcomes for people living with a rare disease. However, there are barriers that can give rise to the slow adoption of innovation in the health service such as insufficient resourcing and infrastructure, as well as a lack of data to support enrolment in clinical trials.

**In recent years there have been initiatives which have sought to help overcome some of these barriers, such as collaboration with other EU countries, the establishment of the Rare Disease Technology Review Committee (RD-TRC) and the initiation of the EU-level Joint Clinical Assessments for advanced therapy medicinal product (ATMPs). Ensuring that innovative practices are adopted and rolled out across our health service to the ultimate benefit of patients requires a concerted effort and forward planning.**

Furthermore, the HSE's Technology and Transformation Office is advancing the eHealth Ireland Strategy which will bring improved population wellbeing, health service efficiencies, and economic opportunity using technology enabled solutions. One of the current aims is to advance Ireland's electronic health records, streamlining patient care and improve efficiency of health records management, and at the patient side the HSE Health App will allow patients to securely access their health information, find services and advice, and manage hospital appointments.

The HSE is advancing in its implementation of new health technologies, through its Action Plan for Health Research 2019 – 2029, the eHealth Ireland strategy, as well as the wide participation of HSE facilities in conducting clinical trials and research.

## Strengthening Leadership and Coordination

A critical step toward advancing rare disease research in Ireland is the need for stronger leadership and coordination, both nationally and within the HSE. The alignment of key stakeholders is essential to fostering a cohesive research environment. By streamlining collaboration and establishing stronger relationships between stakeholders through the development of a clear and coordinated approach, research efforts can be better integrated into the health system and have greater impact on patient outcomes.

This Strategy offers an opportunity to enhance strategic alignment across all relevant actors. By establishing dedicated leadership and oversight mechanisms, Ireland can ensure that research priorities align with the needs of PLWRD and clinicians, while also fostering a culture of innovation that permeates all aspects of service design and delivery will ensure innovations in healthcare and the delivery of services are identified as early as possible, facilitating operational readiness and speedy adoption.

## Recommendation 7

**The Steering Group recommends that a coordinated, and strategic approach to rare disease research across the island of Ireland be put in place. This Includes:**

**A.** Establishing a National Rare Diseases Research Group, including PLWRD acting as partners, to provide a structured, coordinated, multi-actor and strategic approach to ensuring more high-quality rare diseases research in Ireland. This should aim to build research capacity and capability from basic research to the conducting of clinical research (including clinical trials) within the health services (including ERN hospital sites) and universities

**B.** Increasing the opportunities for PLWRD to access, as potential participants, high-quality clinical trials and healthcare interventions by supporting the RD Clinical Trial Network (HRB) and through Ireland's participation in the European Rare Diseases Research Alliance (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 will support this recommendation.

**C.** Increase opportunities for rare disease specific funded research calls targeting both indigenous research and greater participation in international consortia.

**D.** Ensuring healthcare professionals are supported and encouraged to pursue research in rare diseases and provided with the dedicated time to do so.

# 11

# Orphan Medicines

Genetic testing has enabled faster diagnosis and advances in clinical based research have illuminated the causes of many conditions, identifying new biological targets for drug development.

However, where breakthroughs do occur, ensuring that people with rare diseases have equitable and affordable access to new medicines remains both a priority and challenge. Only an estimated 5% of rare disease types have a treatment available[41], and only some of these treatments can be considered transformative.

**Rare diseases were historically overlooked by the pharmaceutical industry due to the high costs and low profitability associated with the commercialisation of research for conditions with small population numbers. The classification of Orphan Medicinal Product (OMP) was introduced to the European Union by Regulation (EC) No 141/2000 (Orphan Drug Regulation) with the goal of fostering an attractive market landscape through incentives for companies developing treatments for rare diseases.**

By recognising the need for viable commercial models to bring OMPs to market, these supports have unlocked innovation and offer a vital tool in addressing the unmet medical needs of patients with rare diseases. As of 2024, the European Commission[42] has granted a Marketing Authorisation (MA) to more than 250 OMPs, with over 2000 active OMP designations (not withdrawn or expired) granted by the European Medicines Agency (EMA).

OMPs have the potential to offer hope where, previously, little to none existed. Public policy in Ireland, must continue to play a central role in changing the way the drug development process works, improving patient access to life-changing medicines by harnessing the full potential of OMPs within our national health and care services.

41. Orphanet (2024). Medicinal products for rare diseases in Europe. [online] Available at: https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Medicinal_products_for_rare_diseases_in_Europe.pdf.
42. European Commission (2025). Orphan medicinal products. [online] health.ec.europa.eu. Available at: https://health.ec.europa.eu/medicinal-products/orphan-medicinal-products_en.

## European Regulatory Approval

The European Medicines Agency (EMA) is responsible for reviewing applications by the sponsors of a new medicine for orphan designation within the European Union. To qualify for designation as an OMP:

- the **medicines must be intended for the treatment, prevention or diagnosis** of a disease that is life-threatening or chronically debilitating;
- the **prevalence of the condition in the EU must not be more than 5 in 10,000** or it must be unlikely that marketing of the medicine would generate sufficient returns to justify the investment needed for its development; and
- no satisfactory method of diagnosis, prevention or treatment of the condition concerned can be authorised, or, if such a method exists, the **medicine must be of significant benefit to those affected by the condition**.

Applications for designation as an OMP are reviewed by the EMA's Committee for Orphan Medicinal Products (COMP), which includes patient representatives. The COMP evaluation process does not consider a medicine's efficacy or safety. Designation as an OMP occurs early in the development life cycle of a medicine and while 66% of applications for OMP designation have been approved, only 8% have gone on to successfully receive marketing authorisation

After a new medicine has been designated as an OMP, the developer may then submit a marketing authorisation application to the EMA's Committee for Human Medicinal Products (CHMP). It is compulsory for all OMPs to undergo the EMA's centralised procedure. This ensures that newly authorised OMPs are authorised for sale throughout the European Union and participating countries in the European Economic Area.

The CHMP evaluation process considers evidence of a medicine's risk-benefit ratio for a specific indication. A marketing authorisation is not a general endorsement of a new medicine, but rather an expert opinion that its benefits outweigh its health risks. Health Technology Assessments (HTAs) by national authorities compare new medicines to other health technologies once they have been authorised to assess comparative clinical superiority and cost-effectiveness.

## Incentives

**A range of regulatory incentives are offered in the European Union (EU) for medicines that have been granted an orphan designation. These include**

- 10 years market exclusivity (extendable to 12 years in certain circumstances), a single EU authorisation system, and reduced fees for several EMA regulatory activities including applying for marketing authorisation.
- Where a new medicine has the potential to provide a significant benefit from unmet medical need (i.e. one where no suitable treatment is available), the developer can apply for the accelerated authorisation support scheme PRIME. This can accelerate the assessment by the CHMP from 210 to only 150 days.
- The European Commission also provides research grant funding. For example, Horizon Europe and the E-Rare ERA-NET support research programmes on rare diseases.

These incentives have led to an increase in the number of orphan drugs developed and approved in recent years. To date, the European Commission has already authorised more than 250 orphan medicines for the benefit of people living with rare diseases.

## A Changing Environment

**Following the publication of the Pharmaceutical Strategy for Europe in 2020, the regulatory environment is being reshaped by new European legislative proposals governing all human medicines. The framework for OMPs will be particularly impacted by these changes as stricter criteria and conditions in relation to orphan designation are proposed in the revised Pharmaceutical Regulation.**

This will ensure that incentives are used to best effect by rewarding those innovative technologies with the highest impact. The emphasis on 'first-in-class' therapies will encourage the development of gene and cell therapies with direct benefits for previously untreated rare diseases.

The Health Technology Assessment Regulation[43] formally entered into force on 01 January 2025. The clinical efficacy element of national Health Technology Assessments will now be conducted jointly with other EU Member States. This will be broadened in 2028 to include all OMPs as well, reducing duplication of effort between national reimbursement processes and is intended to speed up the national reimbursement process. It will ensure the most extensive and up-to-date information is available to inform national decision-makers. Ireland's National Experts have played an instrumental role behind the Regulation and should continue to take full advantage of this new opportunity.

43. European Parliament and Council of the European Union (2021). Regulation – 2021/2282 – EN – EUR-Lex. [online] Europa.eu. Available at: http://data.europa.eu/eli/reg/2021/2282/oj.

## Ireland's Medicines Pricing & Reimbursement Process

Once marketing authorisation has been granted by the European Commission, a medicine may be legally sold in Ireland.

**The Health (Pricing and Supply of Medical Goods) Act 2013 provides the statutory framework for the reimbursement of medicines in Ireland. It sets out a clear legislative basis for the supply and reimbursement of items to patients under the GMS and community drugs schemes.**

It also sets out criteria[44] which the HSE must take into account when making reimbursement decisions. It is important to reflect that unless a company submits an application to the HSE, it cannot reimburse that medicine through the Community Drugs Schemes or in hospitals. OMPs may be unaffordable for many patients out-of-pocket, and this can impact the equity of patient access. Once an application has been received, the HSE can commission the National Centre for Pharmacoeconomics (NCPE) to conduct a rapid review of the comparative clinic efficacy and cost-effectiveness of the new medicine. The NCPE publishes regular updates on this process on its website.[45]

The **Rapid Review** assessment report consists of the following information:

- An overview of current clinical practice and treatment options for the disease
- An overview of how well the drug works compared to current clinical practice
- An overview of the drug cost and likely budget impact
- Information on cost-effectiveness assessments in other countries.

If the Rapid Review report finds that the medicine is cost-effective, the NCPE will recommend that it be reimbursed. In the absence of definitive evidence of cost-effectiveness, the NCPE will advise the HSE of the need for a full HTA. Products with a high-cost relative to potential comparators and/or those with a substantial net impact on the drugs budget require a full HTA. Similarly, products where there is a query in relation to the comparative clinical efficacy will also be selected for formal HTA.

44. (1) The health needs of the public; (2) The cost effectiveness of meeting health needs by supplying the item concerned rather than providing other health services; (3) The availability and suitability of items for supply or reimbursement; (4) The proposed costs, benefits, and risks of the item or listed item relative to therapeutically similar items or listed items provided in other health service settings and the level of certainty in relation to the evidence of those costs, benefits and risks; (5) The potential or actual budget impact of the item or listed item; (6) The clinical need for the item or listed item; (7) The appropriate level of clinical supervision required in relation to the item to ensure patient safety; (8) The efficacy (performance in trial), effectiveness (performance in real situations) and added therapeutic benefit against existing standards of treatment (how much better it treats a condition than existing therapies) and (9) The resources available to the HSE

45. National Centre for Pharmacoeconomics, Ireland (2025). NCPE Ireland. [online] www.ncpe.ie. Available at: https://www.ncpe.ie/.

**Full Health Technology Assessment:** This is a systematic assessment of the clinical and cost-effectiveness of a medicine. The full HTA report consists of the following information:

- Disease background and epidemiology
- Detailed description of current clinical practice and treatment options
- Detailed description of the intervention (drug) under assessment
- Detailed review of the clinical and comparative efficacy of the drug under assessment
- Detailed review of the safety and comparative safety of the drug under assessment
- Detailed review of the cost-effectiveness of the drug under assessment
- Detailed review of the budget impact of the drug under assessment.

Upon completion of a HTA, the NCPE will issue its recommendation to the HSE Drugs Group.

The Drugs Group is the national committee which the HSE has in place to make recommendations on the pricing and reimbursement of medicines to the HSE Senior Leadership Team (SLT). The membership of the HSE Drugs Group includes public interest members. The HSE Drugs Group reviews the pharmacoeconomic report of the NCPE but also considers other factors, including clinical need and patient submissions, in reaching its recommendation.

When the medicine under consideration is an OMP, the Drugs Group can also request a report from the Rare Diseases Technology Review Committee (RDTRC). The RDTRC membership includes consultant clinicians specialising in rare diseases, the Clinical Lead for the National Clinical Programme for Rare Diseases, pharmacists, and a minimum of two patient representatives from a selected panel of three. The RDTRC was a recommendation of the National Rare Diseases Plan 2014-2018 and was established in 2018.

Final decision-making authority in the HSE rests with the SLT. The SLT decides on the basis of all the demands it is faced with (across all services), whether it can fund a new medicine, or new use of an existing medicine, from the resources that have been provided to it, in line with the Health (Pricing and Supply of Medical Goods) Act 2013.

Greater transparency and insight in the reimbursement process is now available to the public through the reimbursement applications tracker which was established in December 2024.[46] While this applications tracker is a recent development, the HSE is committed to expanding the scope of the tracker over time, and in 2025, as set out in its National Service Plan, the HSE aims to include indicative timelines for each application on the tracker, which will further enhance transparency.

46. HSE Pricing and Reimbursement Application Tracker (2025). HSE – PCRS. [online] Sspcrs.ie. Available at: https://www.sspcrs.ie/portal/rdt/pub/application/home [Accessed 26 Mar. 2025].

The industry-funded annual Waiting to Access Innovative Therapies (WAIT) Survey ranks European countries in order of the number of EMA-authorised medicines available for reimbursement, and the time between EMA marketing authorisation and an OMP's addition to the reimbursement list. In the WAIT Surveys, Ireland has performed less well than other European Countries. The timing of company applications for new medicines reimbursement in different countries can vary for several reasons, not least the available market share in each country. Ireland, by virtue of its size and market share, may not always be prioritised by a company in the first stages of marketing a new product. The reimbursement assessment process cannot begin until an application is received. Ireland encourages all developers of medicines to submit their applications promptly following marketing authorisation.

**In February 2023, the Minister for Health published a Review of the Governance Arrangements and the Resources currently in place to support the Health Service Executive reimbursement and pricing decision-making process. The Review found that the HSE reimbursement process is operating in line with the legislation and that it is delivering results in keeping with international norms.**

However, it made a range of recommendations to support improvements to the process. On foot of this review, the Government provided substantial funding for investment in resources to enhance the HSE's medicines pricing and reimbursement process. This investment has delivered 34 new staff across the system to enable the HSE to operate to its full potential and provide predictability for applicant companies. This investment in capacity will enable them to conduct timely and efficient evaluations of medicines for reimbursement. It will also maximise the use of this substantial public investment to support access to more medicines for more people. Given the specialised skillset required for these roles, recruitment was a complex endeavour which only reached completion in the second half of 2024. As a result, their full impact will only become evident in the coming months.

## Progressing the National Rare Disease Plan 2014-2018

The establishment of the RDTRC in line with the National Rare Disease Plan for Ireland 2014-2018 was a significant advance, enhancing the voice of patients in the national reimbursement process. The Committee has contributed to the development of clinical guidelines for OMPs and supports the implementation of guidelines in conjunction with the HSE's National Drugs Management Programme Office where applicable.

**In June 2018, Ireland joined the Beneluxa Initiative, a collaboration between Belgium, The Netherlands, Luxembourg and Austria. Its aim is to work towards sustainable access to, and appropriate use of, medicines in member countries.**

Ireland successfully worked with its partners in Beneluxa to jointly assess and negotiate access to two OMPs, Zolgensma, a treatment for Spinal Muscular Atrophy (2021), and Libmeldy, a treatment for Metachromatic Leukodystrophy (2024). Initiatives, such as Beneluxa provide opportunities, including by providing greater bargaining power in joint negotiations and the leveraging of a bigger market to encourage pharmaceutical companies to launch products in Ireland at the earliest opportunity.

One of the recommendations of the NRDP 2014-2018 was to enhance horizon scanning between pharmaceutical companies and the HSE to improve information available regarding orphan medicines in the pipeline, and this has been another major benefit of Ireland's participation in international initiatives. The International Horizon Scanning Institute (IHSI), launched by Beneluxa members in October 2018 but open to all countries, has provided unprecedented predictive insights into upcoming developments in the clinical and regulatory space. Ireland will continue to actively support and engage with Beneluxa and the IHSI to ensure that the reimbursement system can anticipate and prepare for new treatments.

The HSE Medicines Management Programme (MMP), established in 2016, has supported clinicians with prescribing guidelines and Managed Access Protocols (MAPs). MAPs ensure that OMPs are utilised to greatest potential benefit for patients and optimise the allocation of resources within the HSE.

## Future Progress

New technologies, gene therapies, cell-based treatments, and advanced diagnostics are offering new hope for patients with rare diseases. As the global awareness of rare diseases grows, there is increasing collaboration between health services, patients, advocacy groups, researchers, and pharmaceutical companies.

**The new Programme for Government 2025 – Securing Ireland's Future, commits to "investigating new methods for earlier reimbursement of certain treatments, including early access schemes for rare diseases".**

However, there are still many challenges ahead. The cost of orphan drugs remains a barrier for patients and healthcare budgets, especially when cost-effectiveness is difficult to demonstrate from the small-cohort studies inherent to small-population rare diseases. There is a need for more robust data collection and patient registries to better understand rare diseases and track treatment outcomes over time. The input of patient groups at an early stage of the HTA process and during the development of patient registries is valuable and should be harnessed further.

Ireland continues to be at the centre of technological advances by co-ordinating its existing resources. Leadership on research has the potential to bring more clinical trials to Ireland and promote dynamic solutions to the problems faced by patients with rare diseases. The Health Research Board has invested more than €150m over the last decade and a half in clinical trials infrastructure and research support services to ensure Ireland's clinicians and researchers have the enabling environment to form partnerships and collaborations with other investigators and industry for the benefit of patients, the health system and the economy. Ireland must now encourage the pharmaceutical industry and other stakeholders to engage with this infrastructure to mutual benefit.

There is a growing trend in the EU towards offering reimbursed access to medicines prior to marketing authorisation by the European Medicines Agency or later, prior to a final reimbursement list decision by the relevant national authority. This is often contingent on the collection of real-world data (RWD) by the participating company and is a complex, rapidly evolving area of public health policy. There are benefits for patients through system changes to deliver a more flexible, partnership approach between the pharmaceutical industry and national health systems.

Ireland must embrace change to ensure its health and care services integrate and champion the latest and most advanced technologies. The provision of a shared care records platform may provide future opportunities to support rare diseases. The future of orphan medicinal products is promising, and with continued support from regulators, researchers, and healthcare systems, more rare disease patients can look forward to a brighter future.

## Recommendation 8

The Steering Group recommends that ongoing efforts should be leveraged to maximise access and availability to treatments and technologies for people living with a rare disease and should be explored in full. This includes:

**A.** Reviewing options for earlier reimbursement of certain treatments;

**B.** Consideration of early access schemes for rare diseases, with a view to facilitating sustainable and consistent access to new medicines;

**C.** Enhancing the State's engagement with international initiatives and Health Technology Assessment (HTA) collaboration aimed at improving people's access to medicines.

12

# Public Information and Awareness Campaigns

Public Information and Awareness Campaigns should be aimed at educating the public, healthcare providers and policymakers about rare diseases to increase understanding, reduce isolation, and improve people's outcomes.

**We will also seek to raise awareness amongst the rare disease community of the resources and supports that are available to them. All stakeholders and partners have a role in improving awareness of rare disease across society.**

The general public may have never heard the name of most rare diseases, which can lead to misunderstanding and isolation for people affected by these diseases. Awareness campaigns will work to inform the public about the nature of living with rare diseases from a rights-based perspective, promoting equity and inclusion. By increasing general awareness across society, we will support communities and society as a whole to become a more understanding and inclusive place for PLWRD.

Campaigns targeting healthcare providers, developed in conjunction with people with lived experience of rare diseases, will focus on increasing knowledge and general awareness about rare diseases, and the supports that exist to improve health equity for PLWRD when accessing services. This, in conjunction with increased educational opportunities for healthcare personnel will improve the overall awareness and knowledge, skills and attitudes of healthcare personnel and positively impact quality of life for PLWRD.

**Raising awareness of rare diseases as a public health priority requiring investment and resources is critical to the successful implementation of this Strategy. By informing policymakers and healthcare professionals on the challenges PLWRD face – such as day to day challenges, delayed diagnosis, fragmented care coordination, limited treatment options, and high healthcare costs, – we can encourage the creation of supportive policies to improve outcomes and quality of life. Increased policy maker understanding and engagement should strengthen their support in the implementation of this Strategy and drive improvements in outcomes for the individuals affected, their families and caregivers, and society as a whole.**

> **Being given a diagnosis with no information available on the condition and without supports organised has been very challenging, and has left us to navigate the life changes with no support or guidance.**
>
> – Response from Public Consultation

Raising awareness of rare disease supports and resources (and developing additional resources where needed), such as accessible information and resources, patient organisations, support groups, psychological and social care supports, specialised healthcare providers and research opportunities will bridge the information gap and guide individuals and families toward additional sources of help and potentially provide hope for the future. Giving individuals, family members and caregivers access to information resources and support networks will empower them as they navigate the challenges of living with a rare disease to make informed decisions about their health care.

To achieve this, a partnership approach must be taken to the design of public information and awareness campaigns, with PLWRD and their representative groups co-designing information and awareness campaigns informed by lived experiences. This will require the development of a communications and awareness plan, and a range of accessible resources.

## Recommendation 9

### The Steering Group recommends:

**A.** The development of a Rare Disease Communication and Awareness Plan, including input from PLWRD, to raise awareness of rare diseases among people/families living with rare diseases, the general public, healthcare professionals and policy makers.

**B.** Supporting and educating people living with rare diseases, their families, and carers about the specific rare disease they are dealing with, and the availability of national/international peer-to-peer supports. It is also important that these individuals help design the information, education, and support resources to ensure they meet their needs.

**C.** Development of accessible rare disease information, resources, and educational packages for people/families living with rare diseases and healthcare professionals to increase rare disease awareness and literacy, in collaboration with PLWRD.

13

# Rare Disease Education & Training

Education and training of healthcare professionals (pre and post registration) in rare diseases is crucial in improving diagnosis, treatment, care and support for people living with a rare disease. Since rare diseases are often complex and poorly understood, informed and trained professionals can recognise symptoms earlier, offer a more timely and accurate diagnosis, and provide effective intervention processes including treatment and management of rare conditions.

Ongoing education and training will also support healthcare providers to stay updated on the latest research, therapies, innovative technologies and clinical trials, ensuring that PLWRD receive the best possible care, leading to better outcomes.

Moreover, well-trained and informed healthcare providers can better advocate for people living with a rare disease, navigate the challenges of coordinating specialised supports, and collaborate with multidisciplinary/interdisciplinary teams to improve overall health and social care outcomes.

## Graduate and Postgraduate Health and Social Care Programmes

While recognising the work done by the higher education sector to raise awareness of rare diseases and support students to develop the competencies needed to identify a potential rare disease, it remains important that increased education on rare diseases is incorporated into the core curricula of both graduate and postgraduate healthcare and social care courses in line with enhanced standards of best practice. This will ensure that healthcare professionals are provided with the knowledge and skills to recognise and appropriately triage any person who may present with symptoms that could be linked to a rare disease.

Rare diseases modules or short specialised education programmes should be incorporated into both graduate and postgraduate curricula, these should address genetics, pathology, clinical practice, diagnostics, daily living, physical, psychological and mental health and wellbeing.

Workshops focused on rare diseases should also be offered as part of continued professional development (CPD) and tailored to specific providers to ensure that health and social care professionals are well equipped to understand rare diseases when they present through clinical and/or health and social care practice.

> **Limited understanding amongst healthcare professionals regarding the impact on quality of life and other family members has been an ongoing challenge**
>
> – Response from Public Consultation

**To progress this work, it is important that the Implementation Oversight Group engage with the relevant educational and training bodies, including the third level institutions and umbrella entities such as the Forum of Irish Postgraduate Medical Training bodies which seek to support the alignment of medical education and training in partnership with other major stakeholders in promoting excellence in healthcare provision for the Irish population. The universities and third level institutions offering CAO programmes in Health Sciences similarly should be engaged in discussion about ongoing formative curriculum development for undergraduate and graduate entry programmes, along with the principal regulatory and accreditation bodies.**

## Education and Training Activities provided by European Reference Networks

European Reference Networks (ERNs) play a pivotal role in improving the diagnosis, treatment, and care of patients with rare and complex diseases. To achieve these goals, ERNs provide a wide range of training and educational activities designed to enhance the knowledge and skills of healthcare professionals, researchers, and participants.

All ERNs have developed comprehensive online learning platforms that offer various e-learning courses, webinars, and interactive modules. These platforms are accessible to healthcare professionals across Europe, enabling them to stay updated on the latest advancements in rare disease management.

In addition to online learning platforms, ERNs have established ERN Academies, which offer a structured curriculum designed to provide in-depth training on various aspects of rare disease management. These academies feature a blend of theoretical and practical learning, combining online modules with in-person sessions led by renowned experts and week-long secondments to other ERN sites. These academies fund the activities for the participants including their travel and international visit, and award certificates for their programmes and some include postgraduate diplomas being awarded. ERN Academies aim to cultivate a highly skilled workforce capable of addressing the unique challenges associated with rare diseases.

Additionally, regularly organised workshops and seminars provide hands-on training and foster collaboration among experts from different medical disciplines through ERNs. These events cover a broad spectrum of topics, from diagnostic techniques to innovative treatments, ensuring that participants gain practical insights and skills. This includes annual conferences and symposia that bring together leading specialists, researchers, and policymakers. These gatherings serve as platforms for sharing innovative research, discussing clinical guidelines, and exploring new therapeutic approaches. Attendees benefit from networking opportunities and the exchange of best practices.

The educational activities delivered through ERNs are not limited to healthcare professionals; PLWRD, their families and representative groups may also benefit from tailored programmes. ERNs provide resources, workshops, and support groups that empower patients with the knowledge needed to manage their conditions effectively.

## Role of the National Rare Diseases Office

The NRDO is the coordination hub for rare disease related activity within the Irish healthcare system. Rare disease education, training and awareness is a key function of the office. The NRDO is currently operating the following related initiatives:

- **Third level education and information sessions**
- **Development of information/educational material** to increase knowledge and awareness about rare diseases and the resources and supports available to HSCPs and PLWRD and their families
- **Development of eLearning modules** for healthcare professionals, including GPs, and educational videos for GPs and members of the public
- **NRDO website** which provides information on the National Rare Disease Information Service, Care Pathways for rare diseases, ERNs, national social care supports, research projects and educational resources.
- As part of its rare disease awareness and educational programmes, the NRDO regularly **collaborates with stakeholder groups** to deliver educational presentations/ lectures.
- **HRB / NRDO collaborative workshops** and education / research dissemination to Clinical Leads and patient representative groups (on request)

It is important that the work already being undertaken by the NRDO is supported to ensure that these important initiatives are continued and, where applicable, expanded to ensure ongoing collaboration and knowledge sharing for healthcare professionals with the support of all stakeholder groups, including PLWRD and their families.

Through the public consultation for this Strategy, respondents noted that the biggest challenge facing people living with a rare disease was **"Awareness of rare disease amongst healthcare professionals. This represents a significant barrier for people living with a rare disease, who often must become experts in their own condition and who feel that they are constantly explaining their rare disease, treatment and management to healthcare professionals due to a lack of awareness or knowledge of their condition."**

> **Once diagnosed, I've received excellent care and received a lot of information**
>
> – Response from Public Consultation

By ensuring that appropriate modules and training on rare diseases are integrated into the curricula and training programmes of medical professional and carer disciplines, we can ensure that our health workforce are equipped with the knowledge, skills and awareness to provide better care for people living with a rare disease, their families, and carers.

The important education and training resources provided through ERNs and the National Rare Diseases Office should also be harnessed and promoted in a sustained and co-ordinated manner to ensure that continuous education and training is provided to healthcare providers at all career stages, medical students and researchers, to ensure that there is continued awareness of the latest research, treatment options, and clinical guidelines for rare diseases. Good promotion will result in a greater uptake of opportunities and a more knowledgeable health workforce.

## Recommendation 10

**The Steering group recommends enhanced collaboration with the education sector and training bodies, and the upskilling and development of the rare disease multidisciplinary workforce, this should include:**

**A.** Adding and improving rare disease content in the core curricula of college and university courses for Healthcare Professionals at all levels, to enhance standards, including undergraduate, graduate entry, and postgraduate levels.

**B.** Ensuring ongoing training and professional development opportunities for Healthcare Professionals, such as short courses or flexible learning options (like micro-credentials[47]). This should include raising awareness about these opportunities so that more professionals take them up.

**C.** Greater ongoing promotion and utilisation of education and training resources and initiatives, especially those provided by the ERNs, across all primary and acute services settings in the Health Regions.

47. In health and social care, micro-credentials are short, focused courses designed to demonstrate specific knowledge, skills, and experience in a particular area. They are often flexible, part-time, and can be delivered online or through a blended approach. Micro-credentials are intended to provide upskilling and reskilling opportunities, allowing individuals to gain new competencies in a faster and more flexible way.

# 14

# Implementation and Monitoring

The successful implementation of Ireland's new National Rare Disease Strategy requires a comprehensive, coordinated, and sustainable approach to implementation and monitoring. This strategy and its implementation represent a critical opportunity to transform care and support for individuals and families affected by rare diseases.

## Guiding Principles

Full Implementation of this Strategy will ensure that the vision of equitable, inclusive, and cross-sectoral care for people living with rare diseases is fully realised. The guiding principles that will underpin good implementation are:

## Governance and Accountability

Good governance and clear accountability will be integral to delivering on this Strategy's recommendations. The structures below will be established as a priority to drive implementation and deliver on the Strategy's recommendations. Acknowledging the recent programme of reform undertaken within the HSE, the ever-evolving nature of healthcare services and the current pace of innovation, we must remain flexible and agile in our approach to implementation over the lifetime of this Strategy.

### Implementation Oversight Group

The establishment of the Implementation Oversight Group (IOG) will ensure oversight and direction at the highest level. The IOG will oversee the implementation of the Strategy, providing strategic guidance and monitoring progress at a national level. The IOG will monitor progress against and deliver on the commitments made within this Strategy. It will work with stakeholders to define and evaluate performance against key performance indicators and monitor overall progress across all recommendations. Membership of the IOG will be fully representative of those stakeholders principally involved in ensuring effective delivery of the Strategy's recommendations, including strong patient representation and partnership. It **will** include senior responsible and accountable personnel across the Department of Health, HSE, patient partners and appropriate links to other stakeholders and bodies as required. **A diverse membership will ensure that decision-making is informed by a broad range of perspectives and expertise.** The IOG will meet regularly and publish progress reports on the implementation status of the Strategy's recommendations.

### HSE Implementation Structures

To ensure ongoing operational implementation of this Strategy's recommendations, it is proposed that the HSE establish a structure to assist the IOG and to drive implementation over the life of the Strategy. The HSE structure will report to and participate in the IOG, it will ensure delivery of HSE led strategy recommendations in line with an implementation plan to be developed and agreed with the IOG, inclusive of key performance indicators and progress updates. This structure should oversee and drive implementation of HSE-led Strategy recommendations, bringing together necessary stakeholders across HSE functions and patient representatives, and will establish the necessary thematic workstreams as required.

### Implementation planning

The above HSE structure, in conjunction with the IOG, will develop an Implementation Plan that will include a robust framework for monitoring and evaluation, with measurable goals and performance indicators to track progress against the Strategy's vision, ambition and recommendations. Regular progress and status updates will be provided to the IOG.

## Monitoring, Evaluation, and Continuous Improvement

As outlined above the IOG will oversee implementation and monitor progress against the Strategy's recommendations at a national level. Three Implementation Reports will be published at regular intervals. This will offer an opportunity to recalibrate as needed. These regular evaluation cycles will ensure that the Strategy remains dynamic and responsive. Insights gained from monitoring, evaluation and patients' lived experience will inform continuous improvement, helping to refine approaches and maximise impact.

Workstreams will focus on specific thematic areas of the Strategy and implementation plan, ensuring that actions are addressed in a coordinated and timely manner. This structure will provide clarity, accountability, and flexibility to respond to emerging challenges. Policy oversight will be provided by the Department of Health, ensuring alignment with national health priorities and legislation as required. The HSE will lead operational delivery, leveraging its capacity to integrate rare disease services within the broader healthcare system. Implementation reports will provide an opportunity to robustly assess progress, identify challenges, and refine approaches as needed.

## Stakeholder Engagement and Inclusivity

Achieving broad stakeholder buy-in will be essential to the Strategy's success. People Living with Rare Diseases, families, health and social care providers, researchers, policymakers and community organisations all have a role to play in its implementation. Mechanisms for meaningful engagement, such as regular consultation forums (e.g., HSE's National Patients and Service Users Forum[48] and the proposed Patient and Service User Partnership offices across the Health Regions) and advisory panels, will foster collaboration and ensure that diverse voices are heard. Understanding the lived experience of PLWRD will provide invaluable insights, helping to shape actions and ensure that the Strategy remains person-centred.

48. Health Service Executive (2023a). National Patient and Service User Forum. [online] HSE.ie. Available at: https://www.hse.ie/eng/about/who/national-services/partnering-with-patients/national-patient-forum/.

Inclusivity will be prioritised to ensure that all voices, particularly those of individuals and families affected by rare diseases, are heard and considered. The Strategy's implementation will aim to address the diverse needs of people living with rare diseases, ensuring that no one is left behind. Patient Partnership will be embedded throughout the implementation process to ensure that all actions are shaped by the voices of those most affected. Good partnership will ensure that people with lived experience play an active role in shaping and evaluating the Strategy's implementation.

## Capacity Building and Sustainability

For the Strategy to succeed, the healthcare system must be equipped with the capacity to deliver the required services and adopt innovation practices and technologies. This will involve investment in workforce development, infrastructure, and technology. Multidisciplinary/interdisciplinary teams in the Health Regions across primary, community and acute health and social care settings must be adequately resourced to provide integrated and person-centred care, while ongoing education and training will ensure that healthcare professionals are equipped to meet the needs of people with rare diseases. Horizon scanning and investment in infrastructure will be required to support continuous improvement of services and develop operational readiness to keep pace with advancements in practice and treatment. Adequate and sustained funding will be essential to support the implementation of actions over the long term.

### Recommendation 11

The Steering Group recommends an Implementation Oversight Group (IOG) be established on publication of this Strategy by the Department of Health with appropriately led and resourced workstreams/project teams to support optimal implementation and monitoring of national strategic recommendations:

**A.** The membership of the IOG will include senior responsible and accountable personnel across the Department of Health, HSE, patient partners and appropriate links to other stakeholders and bodies as required.

**B.** The HSE will lead on the implementation of the Strategy, with strong policy oversight by the Department of Health. This will be supported by thematic working groups as required.

**C.** The HSE will develop an implementation plan outlining the actions required for the implementation of this Strategy's recommendations and the associated timelines for agreement with the IOG, inclusive of key performance indicators and progress updates.

# Appendices

# Appendix I – Recommendations

It is important to note that the timelines provided against each of the below are indicative and subject to funding, sufficient resources being available and recruitment. As such timelines may be subject to amendment as part of detailed implementation planning, inclusive of engagement with patient partners and all relevant stakeholders on the prioritisation and operationalisation of the Strategy's recommendations.

| Recommendation 1: Governance, Leadership & Accountability | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Leadership, Accountability and Governance, the Steering Group recommends:<br>a) strengthening and, where required, establishing, clear governance processes that appropriately define and describe the roles, responsibilities and function of all HSE rare disease stakeholders, including patient partners, to ensure appropriate leadership and accountability at both national and regional levels.<br>b) that the National Rare Diseases Office, supported by the Office of the CCO and National Clinical Director for Integrated Care, is expanded and empowered with the necessary infrastructure and resources to drive implementation of this Strategy from a national perspective, and enabled to work in close collaboration with the Health Regions and other National Clinical Programmes, to provide guidance and support for the management, organisation and delivery of rare disease services.<br>c) that the office of the CCO, in collaboration with the Regional Executive Officers, develop clearly defined processes and measurable outcomes to ensure the Health Regions are provided with appropriate guidance and support to operationalise this Strategy. | HSE | a) Commenced immediately and developed as a priority action<br>b) Work is already under way and will require a sustained focus over the lifetime of this strategy.<br>c) Commenced within 6-9 months and iterated as required |

| Recommendation 2: Patient Partnership | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Patient Partnership, Representation and Empowerment, the Steering Group recommends:<br>a) that PLWRD (People Living with a Rare Disease) and their representative groups are equal partners in improving health and social care services. This means PLWRD should act as equal partners across all areas, including the prioritisation, design, development, monitoring and evaluation of policy, services, and research on rare diseases. This partnership approach will be implemented across the country and driven centrally from the HSE and Department of Health, with representation from all areas in line with best practice and relevant national policy, thus ensuring rare disease voices are supported, valued and impactful throughout<br>b) establishing a PLWRD Partnership Advisory Group to review implementation progress and provide input from a lived experience perspective, ensuring a partnership approach across the various workstreams in the prioritisation, design, development, delivery, monitoring and evaluation of the Strategy's actions.<br>c) Financially supporting PLWRD acting as partners, in line with the HSE National Policy on Reimbursement of Expenses for Patient and Service User Partners, such that they can actively participate as partners without incurring significant cost. | DOH/HSE/ Patient Organisations | a) From the outset and continued throughout the lifetime of the strategy<br>b) Established Within 3-6 Months<br>c) Throughout the lifetime of the Strategy |

| Recommendation 3: Screening & Diagnosis | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Screening and Diagnosis the Steering Group recommends the expansion of screening, diagnostics and genetic counselling services in line with best practice to reduce the time to diagnosis, allow timely access to treatment and research/trial opportunities, and ensure PLWRD and their families are fully informed. This includes:<br>a) Expediting the prioritisation, review and evaluation process for potential additions to the Newborn Bloodspot Screening Programme, supported by the dedicated review team in HIQA, while retaining the appropriate rigour<br>b) Supporting and enhancing the current Newborn Bloodspot Screening Programme (NNBSP) to enable further expansion of the programme as per National Screening Advisory Group (NSAC) recommendations and Ministerial approval<br>c) Supporting the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland as it relates to rare diseases.<br>d) Developing and implementing diagnostic pathways to reduce the time to diagnosis for all people living with a rare disease including access to research pathways for SWAN and undiagnosed people. | DoH/HSE<br>Child Health<br>Public Health/<br>NGGO | a) Work to commence immediately<br>b) Work to commence within 12months<br>c) throughout the lifetime of the Strategy<br>d) developed and improved on an ongoing basis over the lifetime of the Strategy |

| Recommendation 4: Eligibility, Access and Integration | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Eligibility, Access, and Integration of Healthcare Services, the Steering Group recommends that future services should be developed in an equitable, integrated, person and family centric manner aligned to Sláintecare and the Model of Care for Rare Diseases, specifically recommending:<br>a) That, as part of the application process for medical card assessment, the financial cost of a rare disease diagnosis is recognised and medical expenses arising are taken into consideration.<br>b) The ongoing development and implementation of rare disease integrated care pathways including those for people with an undiagnosed rare diseases or Syndrome without a Name (SWAN).<br>c) The provision of integrated, coordinated, and multidisciplinary care and support within the community across all regions in line with individual needs, with timely referral and access to specialist care and support. This includes the development and greater utilisation of rare disease networks nationally and in other EU Member States and should be supported by understanding population-based needs.<br>d) Full implementation of the Model of Care for Transition from Childhood to Adult Healthcare Providers in Rare Diseases, the National Framework for Transition of Care and the development of associated care pathways to ensure appropriate and flexible transition arrangements for children, adolescents and young people, adults and older persons.<br>e) Access to necessary wraparound support services for PLWRD and their families throughout the life course, inclusive of rehabilitation, respite, palliative care, psychological and mental health supports, and all associated ancillary services in line with individual need. | HSE/NRDO/ DOH | a) Within 2 years<br>b) Work commenced within 18 Months<br>c) Delivered within 5 years, and targeting incremental improvements over the lifetime of the strategy.<br>d) Delivered within 5 years, and targeting incremental improvements over the lifetime of the strategy.<br>e) Delivered within 5 years, and targeting incremental improvements over the lifetime of the strategy. |

| Recommendation 5: Data, Health Information Systems & Registries | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Data, Health Information Systems and Registries, the Steering Group recommends:<br>a) The establishment of a national rare disease registry, underpinned by legislation, as necessary, that that 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 surveillance and data reporting is required across health and social care sectors.<br>b) A core project team, with the necessary supports, should be established 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.<br>c) The Steering Group recommends that the longer-term vision for the development of EHRs and associated IT systems should include the full integration of ORPHAcodes, as the gold standard nomenclature for rare diseases. A stepwise and an incremental approach to the integration of ORPHAcodes may be required to achieve this. In the interim, the capabilities of existing IT systems (i.e. SNOMED CTSNOMED CT) should be leveraged to allow relevant data and information to be extracted for utilisation within a registry, surveillance, and service planning. | HSE Child Health Public Health/DOH/ NGGO | a) Aim to be operational within 3 years<br>b) Core Project Team to be established within 6 months<br>c) Planning for the utilisation of existing data to commence within 12 months. Greater embedding of ORPHAcodes within 5 years |

| Recommendation 6: International Cooperation & European Reference Networks | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to International Cooperation and European Reference Networks, the Steering Group recognises that international and cross-border cooperation, including the integration of the European Reference Networks (ERNs) into the Irish healthcare system are essential to ensure access to expertise and the delivery of high-quality care and support to people living with a rare disease and recommends that:<br>a) International and cross border collaboration is not limited to ERN activity but rather, is actively promoted across the board, inclusive of policy development, clinical practice, training and education, and research<br>b) The HSE establish an ERN Integration Working Group of key stakeholders to develop and implement a roadmap for the integration and operationalisation of ERNs into the national healthcare system.<br>c) The NRDO in conjunction with ERN Clinical Leads determine the necessary operating/staffing model for a functional ERN, such that they can deliver across all elements of the strategy, including data and research.<br>d) ERNs are fully supported and resourced to ensure effective and sustainable integration into the national healthcare system.<br>e) To ensure the long-term sustainability of ERNs, the DOH and the HSE take into account the recommendations and outcomes and outputs from the EU4Health Joint Action on the Integration of ERNs into National Healthcare Systems (JARDIN) when available.<br>f) The Department of Health and HSE actively encourage and support applications to join the remaining ERNs at the earliest available opportunity. | NRDO/ERNs/ REOs | a) A process considered and developed within 2 years<br>b) working group to be established within 1 year<br>c) Work to commence within 6-12 months<br>d) Over 5 years – targeting incremental improvements over the lifetime of the strategy.<br>e) when available<br>f) pending open calls for membership |

| Recommendation 7: Innovation & Rare Disease Research | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Rare Disease Research and Innovation, the Steering Group recommends that a coordinated, and strategic approach to rare disease research across the island of Ireland be put in place. This Includes:<br>a) Establishing a National Rare Diseases Research Group, including PLWRD acting as partners, to provide a structured, coordinated, multi-actor and strategic approach to ensuring more high-quality Rare Diseases research in Ireland. This should aim to build research capacity and capability from basic research to the conducting of clinical research (including clinical trials) within the health services (including ERN hospital sites) and universities<br>b) Increasing the opportunities for PLWRD to access, as potential participants, high-quality clinical trials and healthcare interventions by supporting the RD Clinical Trial Network (HRB) and through Irelands participation in the European Rare Diseases Research Alliance (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 will support this recommendation.<br>c) Increase opportunities for rare disease specific funded research calls targeting both indigenous research and greater participation in international consortia.<br>d) Ensuring healthcare professionals are supported and encouraged to pursue research in rare diseases and provided with the dedicated time to do so. | HSE/HRB/ HCRI/ Research Ireland | a) established within 12 months<br>b) over the lifetime of the Strategy<br>c) over the lifetime of the Strategy<br>d) over the lifetime of the Strategy |

| Recommendation 8: Orphan Medicines | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Orphan Medicines, the Steering Group recommends that ongoing efforts should be leveraged to maximise access and availability to treatments and technologies for people living with a rare disease and should be explored in full. This includes:<br>a) Reviewing options for earlier reimbursement of certain treatments;<br>b) Consideration of early access schemes for rare diseases, with a view to facilitating sustainable and consistent access to new medicines;<br>c) Enhancing the State's engagement with international initiatives and Health Technology Assessment (HTA) collaboration aimed at improving people's access to medicines. | HSE/DoH | a-c) Subject to the progression of Programme for Government commitments |

| Recommendation 9: Public Information & Awareness Campaigns | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Public Information and Awareness Campaigns, the Steering Group recommends:<br>a) The development of a Rare Disease Communication and Awareness Plan, including input from PLWRD, to raise awareness of rare diseases among people/families living with rare diseases, the general public, healthcare professionals and policy makers.<br>b) Supporting and educating people living with rare diseases, their families, and carers about the specific rare disease they are dealing with, and the availability of national/ international peer-to-peer supports. It is also important that these individuals help design the information, education, and support resources to ensure they meet their needs.<br>c) Development of accessible rare disease information, resources, and educational packages for people/families living with rare diseases and healthcare professionals to increase rare disease awareness and literacy, in collaboration with PLWRD. | NRDO/ HSE/Patient Organisations | a) Within 12 months<br>b) Over 5 years – targeting multiple initiatives over the lifetime of the strategy.<br>c) Over 5 years – targeting multiple initiatives over the lifetime of the strategy. |

| Recommendation 10: Rare Disease Education & Training | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Rare Disease Education and Training, the Steering group recommends enhanced collaboration with the education sector and training bodies, and the upskilling and development of the rare disease multidisciplinary workforce, this should include:<br>a) Adding and improving rare disease content in the core curricula of college and university courses for Healthcare Professionals at all levels, to enhance standards, including undergraduate, graduate entry, and postgraduate levels.<br>b) Ensuring ongoing training and professional development opportunities for Healthcare Professionals, such as short courses or flexible learning options (like micro-credentials[49]). This should include raising awareness about these opportunities so that more professionals take them up.<br>c) Greater ongoing promotion and utilisation of education and training resources and initiatives, especially those provided by the ERNs, across all primary and acute services settings in the health regions. | 3rd Level Institutions/ HSE/NRDO/ ERNs | a-c) Ongoing over 5 years – targeting multiple initiatives over the course of the strategy |

49. In health and social care, micro-credentials are short, focused courses designed to demonstrate specific knowledge, skills, and experience in a particular area. They are often flexible, part-time, and can be delivered online or through a blended approach. Micro-credentials are intended to provide upskilling and reskilling opportunities, allowing individuals to gain new competencies in a faster and more flexible way.

| Recommendation 11: Implementation & Monitoring | Suggested Lead Agency | Indicative Timeline |
|---|---|---|
| In relation to Implementation and Monitoring, the Steering Group recommends an Implementation Oversight Group (IOG) be established on publication of this Strategy by the Department of Health with appropriately led and resourced workstreams/project teams to support optimal implementation and monitoring of national strategic recommendations:<br>a) The membership of the IOG will include senior responsible and accountable personnel across the Department of Health, HSE, patient partners and appropriate links to other stakeholders and bodies as required.<br>b) The HSE will lead on the implementation of the Strategy, with strong policy oversight by the Department of Health. This will be supported by thematic working groups as required.<br>c) The HSE will develop an implementation plan outlining the actions required for the implementation of this Strategy's recommendations and the associated timelines for agreement with the IOG, inclusive of key performance indicators and progress updates. | DOH/HSE | Established following publication of this Strategy with ongoing oversight and monitoring of implementation and delivery of recommendations within 5 years. |

# Appendix II – Glossary of Abbreviations

| Abbreviation | Definition |
|---|---|
| ADA-SCID | Adenosine Deaminase Deficiency Severe Combined Immunodeficiency |
| AMEQUIS | Assessment, Monitoring, Evaluation and Quality Improvement System |
| ATMPs | Advanced Therapy Medicinal Products |
| CAO | Central Applications Office |
| CCO | Chief Clinical Officer |
| CHI | Children's Health Ireland |
| CHMP | EMA's Committee for Human Medicinal Products |
| CMO | Chief Medical Officer |
| COMP | EMA's Committee for Orphan Medicinal Products |
| CPD | Continuous Professional Development |
| CPMS | Client Patient Management System |
| DoH | Department of Health |
| ECRIN | European Clinical Research Network |
| EHDS | European Health Data Space |
| EHR | Electronic Health Record |
| EMA | European Medicines Agency |
| EOI | Expression of interest |
| EPPerMed | The European Partnership for Personalised Medicine |
| ERAPerMed | European Research Area Networks Cofund of Personalised Medicine |
| ERDERA | European Rare Disease Research Alliance |
| ERN | European Reference Network |
| EU | European Union |
| EUROCAT | European network of population-based registries |
| EURODIS | European Organisation for Rare Diseases |
| GA1 | Glutaric Aciduria Type 1 |
| GDI | Genomic Data Infrastructure Ireland |
| GMS | General Medical Service |
| GOE | Genome of Europe |
| GP | General Practitioner |

| | |
|---|---|
| HCP | Health Care Provider |
| HIQA | Health Information Quality Authority |
| HRB | Health Research Board |
| HRCI | Health Research Charities Ireland |
| HSCPs | Health and Social Care Providers |
| HSE | Health Service Executive |
| HTA | Health Technology Assessment |
| ICGP | Irish Congress of General Practitioners |
| ICPerMed | International Consortium in Personalised Medicine |
| IHSI | International Horizon Scanning Institute |
| INSERM | Institut National de la Sante et de la Recherche Medicale |
| IOG | Implementation Oversight Group |
| IPPOSI | The Irish Platform for Patient Organisations, Science and Industry |
| IT | Information Technology |
| JARDIN | EU Joint Action on the integration of ERNs into National Healthcare Systems. |
| MA | Marketing Authorisation |
| MAPs | Managed Access Protocols |
| MCADD | Medium chain Acyl CoA Dehydrogenase Deficiency |
| MDT | Multidisciplinary Teams |
| MMP | HSE Medicine Management Programme |
| MOC | Model of Care |
| NCAGL | National Clinical Advisor and Group Lead |
| NCDIC | HSE National Clinical Director for integrated Care |
| NCPE | National Centre for Pharmacoeconomics |
| NCTOG | National Clinical Trials Oversight Group |
| NGGO | National Genetics and Genomics Office |
| NGO | Non-Governmental Organisation |
| NNBSP | National Newborn Bloodspot Screening Programme |
| NRDO | National Rare Diseases Office |
| NRN | National Reference Networks |
| NSAC | National Screening Advisory Committee |
| NUNHSP | National University Newborn Hearing Screening Programme |

| | |
|---|---|
| NWIHP | National Women and Infants Health Programme |
| OD4RD | Orphanet Data for Rare Disease |
| OMP | Orphan Medicinal Products |
| ORPHAcodes | System for clinical coding of rare diseases in Europe |
| Orphanet | Database of rare diseases and orphan drugs |
| PKU | Phenylketonuria |
| PLWRD | Person living with a rare disease |
| PPI | Public and Patient Involvement |
| PRIME | EMA Priority Medicines Scheme |
| PROMs | Patient Report Outcome Measures |
| RAiN | All-Ireland Rare Disease Interdisciplinary Research Network |
| RCPI | Royal College of Physicians Ireland |
| RDCat | Rare Diseases Research and Innovation Catalyst Call |
| RD-CTN | Rare Disease Clinical Trials Network |
| RDI | Rare Diseases Ireland |
| RD-TRC | Rare Disease Technology Review Committee |
| REO | Regional Executive Officers |
| RWD | Real-World Data |
| SCID | Severe Combined Immunodeficiency |
| SLT | Senior Leadership Team |
| SMA | Spinal Muscular Atrophy |
| SWAN | Syndromes Without a Name |
| UN | United Nations |
| UNCRPD | United Nations Convention on the Rights of Persons with Disabilities |
| WAIT | Waiting to Access Innovative Therapies Survey |
| WHO | World Health Organisation |

# Appendix III – Steering Group Membership

| Name | Position |
|---|---|
| Prof. Cecily Kelleher | Chairperson |
| Dr Atif Awan | Paediatric European Reference Networks Representative |
| Dr Abigail Collins | Public Health Medicine for Children |
| Mr Alan Finglas | Patient Representative |
| Dr Rosie Gowran | National Clinical Programme for People with Disability |
| Ms Dervela Gray | HSE Clinical Design & Innovation |
| Dr Keith Lyons | Rare Disease Policy, Department of Health |
| Dr Ciara Martin | HSE Clinical Design & Innovation |
| Dr Emma McCann | National Genetics & Genomics Office |
| Dr Cormac McCarthy | Adult European Reference Networks Representative |
| Ms Vicky McGrath | Patient Forum Co-Chair (RDI) |
| Mr Derick Mitchell | Patient Forum Co-Chair (IPPOSI) |
| Dr Ciarán Murphy | Rare Disease Policy, Department of Health |
| Ms Gillian Stafford | Patient Representative |
| Ms Geraldine Sweeney | National Rare Diseases Office (Sept. 2024 – Present) |
| Prof. Eileen Treacy | National Rare Diseases Office (Dec. 2023 – Sept. 2024) |
| Ms Alana Ward | National Rare Diseases Office (Sept. 2024 – Present) |
| Ms Oonagh Ward | Health Research Board |

# Appendix IV – Terms of Reference for the National Rare Disease Steering Group

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.

**In developing the National Strategy, the Steering group will:**

- 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.
- Have regard for Sláintecare, 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.
- Consider how best to integrate European Reference Networks into the national health system.
- 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.
- 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.
- Consider how to best promote rare disease awareness, including education among healthcare professionals, policy makers, and the public.
- Consider how to best promote and support participation in national and international research.
- Consider the governance and funding implication of any of the recommendations arising out of the new National Rare Disease Strategy.

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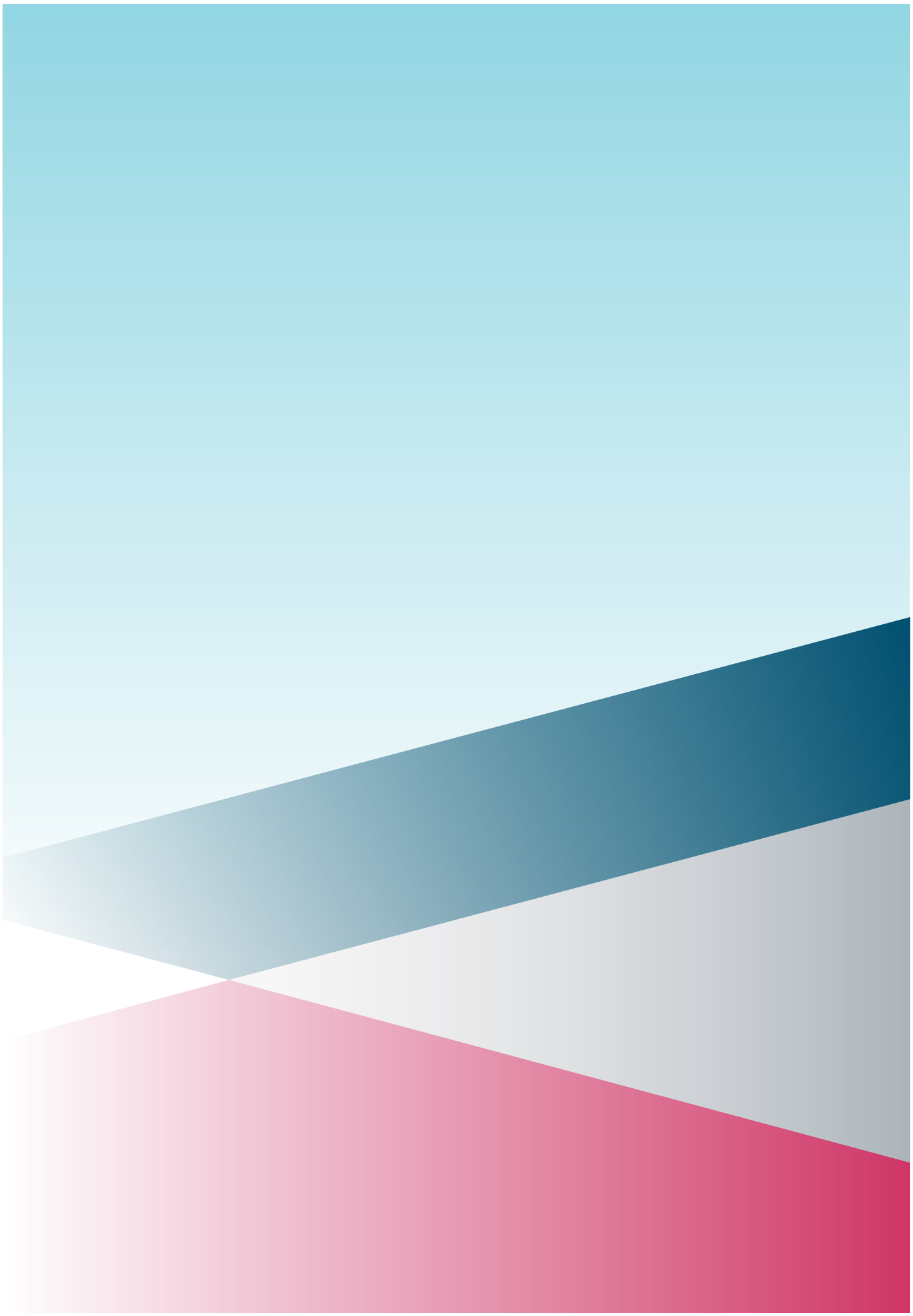

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