NORTHERN IRELAND RARE DISEASE ACTION PLAN

PROGRESS REPORT 2024-25

Contents

Chair’s Foreword

In March 2022, the former Health Minister Robin Swann published the Northern Ireland Rare Disease Action Plan. This Plan set out a roadmap, through 14 high level actions, for improving how services for our rare disease community in Northern Ireland are delivered.

Five Working Groups are now firmly established, led by members of the Northern Ireland Rare Disease Implementation Group (NIRDIG), which I chair, with representation from the Northern Ireland Rare Disease Partnership (NIRDP), as well as stakeholders from across clinical practice, academia, commissioners, research and policy. Progress made by these Working Groups during 2024–25, across a range of critical areas, included:

- Establishment of the LifeArc Centre for Acceleration of Rare Disease Trials that will improve access to specialist care, diagnosis and treatments, and increasing participation in rare disease research;
- Improved access to rare disease specialists and care/treatments through the development of rare disease clinical pathways;
- A dedicated mavacamten clinical service for the treatment of symptomatic obstructive hypertrophic cardiomyopathy, as well as approval to deliver NICE-approved treatment for sapropterin, for people with phenylketonuria (PKU);
- Collaboration with the Northern Ireland Education Authority on awareness sessions for schools, and the development of a Rare Disease Education Toolkit;
- Establishment of a doctoral training program for rare diseases, helping to train the next generation of scientists, data analysts, and healthcare professionals;
- A series of consultation events across Northern Ireland that have reinforced the importance of community engagement, as well as informing future iterations of the Rare Disease Action Plan.

In addition, 2024/25 saw the completion of the roll-out of Encompass, Northern Ireland's digital healthcare record for every citizen, across all five Health and Social Care Trusts, providing all patients with the ability to view and update their health information online through the My Care patient portal. Patients with rare diseases were among those consulted in the design of the portal and are now beginning to experience the benefits of this innovation.

We have also continued to engage regularly with our counterparts across the UK and Ireland with responsibility for rare diseases policy, and with the Assembly All-Party Group on Rare Diseases, ensuring that our priority actions remain appropriately aligned and that opportunities to raise the profile of rare diseases are taken.

Despite the very challenging financial constraints facing the Health and Social Care system, much has been achieved through collaboration and leadership within and across the five Working Groups. There is, however, much more to do as well as a great deal of learning that can be gleaned from progress to date. As we continue our work during 2025-26 and beyond, we must continue to deliver together, building on the firm foundations that have been set.

Finally, a sincere thank you to the NIRDP, Queen's University Belfast, Ulster University, University College Dublin, Rare Diseases Ireland, and all other colleagues, stakeholders and organisations who have continued to provide invaluable assistance since the inception of the Action Plan.

I am delighted to share with you this third Progress Report that provides a summary of the extensive activity undertaken during 2024-25.

Professor Ian Young

Chair, NI Rare Disease Implementation Group

Introduction

The UK Rare Diseases Framework was jointly published in January 2021 by all four UK Health Ministers. The Framework was based on the outcomes of the *National Conversation on Rare Diseases*, launched in 2019. This survey gathered views from across the rare disease community in the UK on the major challenges faced by people affected by rare conditions.

Based on the evidence gathered by the National Conversation on the issues that matter most to people living with a rare condition, the Framework outlines the UK's priorities for rare diseases over five years. The Priorities are:

- Helping patients get a final diagnosis faster
- Increasing awareness of rare diseases among healthcare professionals
- Better coordination of care
- Improving access to specialist care, treatments and drugs

The Framework's priorities are underpinned by five important themes:

- Patient Voice
- Collaboration
- Research
- Data and Technology
- Wider Policy Alignment

Whilst the Framework remains a UK-wide document, each of the four UK nations have developed their own Rare Diseases Action Plan, commensurate with local needs and available resources. Each devolved nation has its own delivery or implementation group responsible for developing, implementing and monitoring nation-specific action plans, while working together through the UK Rare Diseases Framework Board to guarantee as much alignment across the UK as possible. The implementation group for Northern Ireland (NI) is the NI Rare Diseases Implementation Group (NIRDIG).

Northern Ireland Rare Diseases Action Plan

NI’s Rare Diseases Action Plan was developed throughout 2021 by NIRDIG, informed by extensive stakeholder engagement and events with the rare disease community in NI. The group continues to oversee and coordinate delivery of the Action Plan. NIRDIG is chaired by Professor Ian Young, Chief Scientific Advisor (CSA) for NI, and its membership comprises stakeholders from across the Department of Health (DoH) and Health and Social Care (HSC), including policy officials, commissioners, public health, the Public Health Agency’s Research and Development (R&D) Division, clinical input from the HSC Trusts, academia (rare disease educators and researchers from Queen’s University Belfast and Ulster University), as well as the rare disease patient voice represented by the NI Rare Diseases Partnership (NIRDP). The group met on four occasions during 2024–25.

The NIRDP is a private-sector umbrella organisation that brings together those living with a rare disease and organisations representing them; clinicians and other health professionals; researchers and producers of specialist medicines and equipment; health policymakers; and academics. NIRDP was heavily involved in the design of the NI Rare Diseases Action Plan, working closely with the Department to ensure that the patient voice was front and centre in rare disease policy development and agreement of actions and priorities.

The Action Plan was published by former Health Minister Robin Swann in March 2022 and sets out 14 high-level actions, with an overall vision to improve the lives of people living with a rare condition in Northern Ireland. Better coordination of care, improved access to clinical trials and research, increased education and training, and collaboration among specialist teams across these islands were — and remain — the key elements of the Action Plan, which can be accessed on the DoH website: [Northern Ireland Rare Diseases - Action Plan 2022/23 and reports | Department of Health]

Five Working Groups (WGs) were established to take forward groups of actions within available resources, with representation on each WG from the NIRDP, as well as wider stakeholders from across clinical practice, academia, commissioning, research, and policy. WGs report on progress through NIRDIG, and the progress made in the first two years of the Action Plan (2022–23 and 2023–24) has been set out in the Department's annual Rare Diseases Action Plan Progress Reports, available on the DoH website at the link above.

Further progress has been made throughout 2024–25, with many actions nearing completion. This progress is fully attributable to the collective efforts, commitment, and diligence of the WGs in driving forward the actions, despite various challenges faced, and the Department offers its sincere thanks to NIRDIG members for all the hard work they have undertaken and all they continue to do for rare diseases across NI.

While good progress continues, there is still much to do, and full implementation within the five-year timeframe remains subject to available resources.

Funding

Since the publication of the Action Plan in 2022, sustained financial pressures across the HSC budget have significantly constrained the availability of dedicated government funding to progress these actions. A non-recurrent, in-year allocation of £50k was secured in 2023–24 to support implementation of the Action Plan, as outlined in last year's Progress Report. However, no additional departmental funding was available in 2024–25 to help progress specific actions.

NIRDIG members have sought funding from various external sources such as the Medical Research Council, LifeArc, and the National Lottery. Where successful, these funding awards have been key to enabling work that underpins the Action Plan's priorities, such as establishing a Rare Disease Information Hub and improving access to clinical trials for people living with a rare disease. The LifeArc funding award was a notable achievement of 2023–24 and, with the project formally commencing in September 2024, it is expected that this investment will start to be fully realised throughout 2025–26.

In addition, there is existing funding within the wider HSC system that supports the diagnosis and treatment of rare diseases, including recurrent funding for adult and paediatric rare disease clinical leadership posts and for genomic sequencing to improve rare disease diagnosis. Expenditure is also ring-fenced for specialist medicines, much of which is used for rare disease treatments and therapies. Investment in major strategic programmes of work, such as Encompass (with capital investment expected to be £300m over ten years), further enables and bolsters the progression of actions within the Action Plan (more detail on Encompass below).

However, funding is only one part of the jigsaw and, as referenced above, a key strength of the Action Plan is the collective commitment and dedication of NIRDIG members.

Summary of Key Achievements over 2024-25

LifeArc Centre for Acceleration of Rare Disease Trials

Improving access to specialist care, diagnosis, and treatments – as well as increasing participation in rare disease research - are key priorities for the NI Rare Diseases Action Plan. The £12m LifeArc Centre for Acceleration of Rare Disease Trials brings together a consortium of three universities (Queen’s University Belfast, Newcastle University, and the University of Birmingham) to boost the capacity and efficiency of rare disease trials across the UK.

The Centre will harness rare disease expertise from across the UK to improve the design of translational research studies, facilitate the delivery of clinical trials, build much-needed local capacity, and improve access to innovative and specialist treatments for patients. This award also establishes a doctoral training programme for rare diseases, helping to train the next generation of scientists, data analysts, and healthcare professionals.

We know that NI is underrepresented in clinical trials, in clinical research involving children, in rare disease research funding, and in rare disease clinical centres of expertise. The LifeArc Centre will help improve research opportunities and facilitate access to treatment for people living with rare diseases in NI and across the UK.

Clinical Pathways and Treatments

Good progress has been made in improving access to rare disease specialists and care/treatments through the development of rare disease clinical pathways by the HSC Rare Disease Clinical Leads and Specialist Services Managers. The Leads are responsible for providing clinical leadership, advice, and input into NIRDIG, including contributing to its work and helping achieve its objectives.

They also work with healthcare professionals to raise the profile and awareness of rare diseases within NI, including identifying the need for educational and information resources and signposting potential specialist treatment services.

A dedicated mavacamten clinical service for the treatment of symptomatic obstructive hypertrophic cardiomyopathy has been established, following a pilot that began in May 2024. Mavacamten is the first treatment that specifically targets the condition of symptomatic obstructive hypertrophic cardiomyopathy rather than just its symptoms. Approval was also secured to deliver NICE-approved treatment for sapropterin for people with phenylketonuria (PKU).

Collaboration continues to strengthen access to specialist drugs and treatments, including the development of relevant service level agreements (SLAs) for NI patients requiring specialist care from GB (for example, the paediatric mitochondrial service).

Education

An education survey was carried out to gather feedback from parents and guardians about their experiences of the education system. The results and views shared by parents have been documented in a Rare Disease Education Toolkit, available here: RD Education Toolkit

These findings were also presented to the Education Authority and have helped shape ongoing collaboration, including delivering awareness sessions for school staff, providing practical tools through the Toolkit, hosting listening sessions with families, and offering signposting to supportive resources.

Patient Portal (Through Encompass)

With the rolling implementation of Encompass throughout the region in 2024–25, NI patients have been gaining access to the MyCare patient portal app, which allows people to view and update their health information online via their digital medical record, empowering patients to take an active role in their healthcare and wellbeing.

The patient portal enables seamless communication with multiple consultants across different medical specialties - a common requirement for those with rare conditions - and helps ease the burden on patients by reducing the need to repeatedly share their medical history and hospital interactions with each health professional they encounter.

Research

A doctoral training program has been established for rare diseases, helping train the next generation of scientists, data analysts, and healthcare professionals - 12 PhD students at Queen’s University Belfast, alongside 10 students in England. Several research projects are being co-led with patients and families living with rare diseases, in partnership with the charities that support them.

Public Engagement Events

“Power of Connection” consultation events were held across NI in February and March 2025 that reinforced the importance of community engagement, as well as informing future iterations of the NI Rare Diseases Action Plan.

2024-25 Progress by Working Group

Working Group 1: Expert Centre, Information Hub, Patient Portal

Led by: Professor AJ McKnight (QUB)

Actions: Information Hub (Action 1), Expert Centre (Action 7) and Patient Portal (Action 10)

Membership of Working Group 1 (WG1) includes stakeholders from organisations such as Queens University (QUB), Ulster University, Department of Health (DoH), Belfast HSC Trust and the Northern Ireland Rare Disease Partnership (NIRDP).

Information Hub

- The action to establish an online Information Hub for Northern Ireland is progressing well. During 2024–25, three years of research funding was secured to develop the RareDiseaseNI.info website. Since then, the Information Hub has continued to evolve, with expanded links to resources, including the NI-specific carers platform (caringwithrare.ni) and the UK-wide Rare Disease Research Network (RDRN) resource led by the Cambridge Rare Disease Network (CamRARE) team. User evaluation sessions are planned for summer 2025 and the intention is that the websites will go live during 2025-26.

- The appointment of an Information and Communications Lead for rare disease marked an important step in strengthening accessibility efforts for people living with a rare condition. This role will ensure the publication of updates and rare disease information on NI Direct, giving patients and carers access to accurate, up-to-date information.

Expert Centre

- Work on this action is ongoing. A scoping review for the Expert Centre was finalised in 2024-25 and public consultation with service users was carried out to ensure alignment with patient needs and research priorities.

Patient Portal through Encompass

- Encompass is a HSC programme that has created a single digital care record for every citizen in NI who receives health and social care. It aims to provide patients and service users with the safest, highest quality care possible and gives them the ability to view and update their health information online, wherever and whenever they like, via MyChart, the patient portal. Encompass also makes it easier for HSC staff to access important information about patients and service users, both in clinical settings and while working in the community.

- As of March 2025, Encompass had been implemented in the South Eastern, Belfast, and Northern HSC Trusts. (Since then, it has been rolled out across the remaining two Trusts — Southern and Western.) An important resource for all citizens, however the patient portal via Encompass is particularly valuable for rare disease patients because it enables seamless communication with multiple consultants across different medical specialties (a common feature of rare conditions), providing fast access to test results, and helping people stay informed about their ongoing care.

- Coordination of care was a key priority identified by the rare disease community in Northern Ireland (and across the UK). The implementation of a patient portal (via the MyChart app) addresses this by empowering patients to take control of how they access and interact with their health information. Every NI patient now has their health information digitally stored in one place, so doctors treating them in a secondary care setting can access their records no matter where they are across NI — leading to more informed and coordinated decisions around diagnosis, treatment, and care. A patient commented:

  > *"The MyChart app has been transformational. I can see test results quickly and directly, hospital letters are easily available, and appointments that help me manage my rare disease alongside that of my family for whom I care – particularly important as we see lots of doctors across different hospitals."*

Looking Ahead

- Discussions are underway to explore the expansion of WG1's scope to include additional initiatives, such as scoping the potential for a Northern Ireland Syndrome Without A Name (SWAN) Clinic, with learning from the establishment of successful SWAN clinics in Wales and England.

- Work will continue to further develop the carers online support tool and website that will help unpaid carers navigate the different stages of their caring journey.

- Steps will be taken to include "rare disease" as an option within the selection box in the patient portal (MyChart) in encompass, enabling patients to explicitly opt in to participate in rare disease research.

Working Group 2: Registry/Pathways/Access to Drugs & Treatments

Led by: Prof AJ McKnight and Dr Alison Muir

Actions: Rare Diseases Registry (Action 4), Pathways (Action 8); Improve access to drugs & treatments (Action 11) and Improve access to rare disease specialist teams (Action 12).

Pathways

- Much progress has been made on the action to improve access to rare disease specialists and care/treatments through the development of the rare disease clinical pathways by the HSC Rare Disease Clinical Leads and Specialist Services Managers. The pathways have been developed in line with NHS England service specifications and also look to the National Model of Care for Paediatric Healthcare Services in the Republic of Ireland for examples of best practice.

- These pathways define current services for rare disease specialties, identify areas for improvement and support a roadmap towards equitable care across NI, while accommodating local healthcare needs through supplementary sections.

- Areas of pathways work include, on the adult side, Inherited Cardiac Conditions (ICC), Inherited Metabolic Disorders (IMD), Immunology, Haematology, Red Cell Disorders (RCD), and Cystic Fibrosis (CF); and on the paediatric side, IMD, CF, Nephrology, Oncology, Haematology, Cardiology, and Chronic Pain.

- Collaboration with GB teams continues to strengthen access to specialist drugs and treatments, including development of relevant service level agreements (SLA) for NI patients requiring specialist care from GB, including for example the paediatric mitochondrial service.

- Work to establish a Northern Ireland Virtual Amyloid Centre is progressing steadily, supporting improved diagnosis and treatment of amyloidosis. The recent approval of tafamidis and additional other specialist amyloid therapies by NICE will further shape this pathway.

- A dedicated mavacamten clinical service for the treatment of symptomatic obstructive hypertrophic cardiomyopathy has been established, following a pilot that began in May 2024. Mavacamten is the first treatment that specifically treats the condition of symptomatic obstructive hypertrophic cardiomyopathy rather than the symptoms and it is now available to patients in Northern Ireland. Many patients have experienced dramatic improvements, with one remarking:

  > *"…mavacamten has given me my life back", and*
  > *"…friends and family cannot believe the change in me."*

- Also of note is the development of a Fabry service, which links with the Royal Free London in a multi-disciplinary team (MDT) after each clinic in NI, to help build resilience within the service in the Belfast Trust.

- A service specification and clinical pathway have been developed to reflect recent policy changes for sapropterin therapy. Sapropterin is used for patients with phenylketonuria (PKU), a rare genetic disorder where the body cannot properly break down phenylalanine (Phe). Untreated, high Phe levels can cause serious health issues. While a strict low-protein diet has been the standard treatment, sapropterin offers an additional option for responsive patients. Parents have commented:

  > *"As a whole, sapropterin has been life changing for us…"*
  > *"Because of sapropterin my daughter was able to attend her first ever team bonding event for her football team…"*

> *"As a mum, I am incredibly thankful for how life-changing sapropterin has been…"*

- A new pathway has also been developed for patients transitioning from children's PKU services to adult services.

Looking Ahead

- A prioritisation process for service specifications and patient pathways will be designed and developed as a structured approach to determine which rare disease services should be developed or reviewed next.

- Paediatric and adult rheumatology will be added to the respective workplans and will commence during 2025-26.

- Work will begin on developing specifications for paediatric nephrology and paediatric cardiology.

- Following approval of business cases, fully funded services for mavacamten and sapropterin will be rolled out. Educational meetings with cardiology colleagues will also be established to share the roll-out strategy and encourage referrals to the mavacamten service.

- The next group of patients is scheduled to start treatment in November 2025, with plans to start a further 17 patients by Spring 2026.

Rare Disease Registry

- Work is ongoing to develop a NI Rare Disease and Congenital Anomalies Register, primarily using secured research funding, in the absence of government funding to date. The Registry's aim is to guarantee the capture and utilisation of patient data to enhance research and treatment accessibility.

- There is established incidence and / or prevalence data for several rare conditions and congenital anomalies in NI, including trisomy 13, trisomy 18, trisomy 21, rare cancers, and selected rare diseases. With support from charity partners, research funding is supporting a focus on congenital anomalies, Fragile X syndrome, neuromuscular, inherited cardiac, and rare kidney conditions over the next few years, connecting patient demographics with multisystem phenotype information and genomic data.

- Diverse stakeholder meetings have been held exploring the consent process for national disease registration of rare diseases, registries, patient and family perceptions on registries and data sharing, options for data collection, and the use of artificial intelligence to support clinical decision making.

- Digital developments are also supported in NI through the recent LifeArc award that focuses on accelerating access to clinical trials for rare diseases. The LifeArc Centre for the Acceleration of Rare Disease Trials explicitly brings together NHS England data from the National Congenital Anomaly and Rare Diseases Registration Services (NCARDRS) with rare disease registry Leads in NI (Northern Ireland RAre Diseases & Congenital Abnormalities Registry - NIRADCAR), the Congenital Conditions and Rare Diseases Registration and Information Service for Scotland (CARDRISS), and the Congenital Anomaly Register and Information Service (CARIS) in Wales (CARIS) to explore options for UK-wide/4-nation research and clinical information in 2025. NI have implemented a scalable, openEHR approach that should enable federation of data where appropriate, resources and governance permitting.

- Registry Leads across the devolved nations continue to meet regularly to discuss best practice, novel ideas, and opportunities to work together. The Leads also input to the blood spot taskforce as part of the work of the National Screening Committee, with a focus on newborn screening.

- Research funding has been secured to develop a population-wide pre-cancer registry using genomic data to identify individuals at risk of developing inherited cancers, including people who may be predisposed to developing certain cancers because of a rare condition e.g. TP53. This project will build digital infrastructure that facilitates streamlined data validation and enhanced health surveillance of people at increased risk, while also enabling benchmarking and audit. By identifying and monitoring at-risk patients, the project seeks to ensure timely and effective interventions, improved patient outcomes and the transformation of the landscape of cancer care.
- Quality Statements for Rare Diseases are being developed on a four-nation basis to define excellence in care, incorporating patient, carer, and clinical perspectives.

Looking Ahead:

- The registry teams from NI, Scotland, Wales, and England will continue to meet regularly, with Wales, Scotland and NI planning to publish a “registry resources” paper noting metadata available alongside accessibility / barriers to accessing data in each devolved nation.

Access to Medicines

- In April 2024, the Department commenced a whole-system review of the NI medicines access landscape, supported by the 2024 Voluntary Scheme for Branded Medicines Pricing, Access and Growth (VPAG) investment programme.

- The aim was to review policy and processes and make recommendations for improvement to ensure that the NI population can continue to benefit from access to clinically and cost-effective new medicines on an equitable and sustainable basis.

- The review sought the views and experiences of a range of stakeholders involved in access to new medicines, including HSC clinicians, the pharmaceutical industry, patient groups, and the Northern Ireland Rare Disease Partnership.

- This engagement has informed a detailed assessment of the current medicines access landscape in Northern Ireland. It sets out future challenges and provides recommendations for change across three emerging themes: policy and guidance, leadership and governance, and systems and processes.

Looking Ahead:

- The Department intends to publish the final Access to Medicines report by the end of September 2025 and will consider how best to take forward the recommendations to ensure that individuals with rare diseases can benefit from clinically and cost-effective innovative medicines in a timely manner.

Working Group 3: Education and Training

Led by: Dr Stephanie Duguez (Ulster University)

Actions: Education & Training (Action 5) and Awareness Raising (Action 6)

- Fostering knowledge and awareness among healthcare professionals and educators remains a key priority. The NIRDP Education Action Team, led by Dr Stephanie Duguez, has met with the Special Educational Needs (SEN) lead at the NI Education Authority to explore ways of improving support for children and young people with rare conditions in Northern Ireland schools.

- The **Rare Disease Education Toolkit** was created during 2024–25 to help schools and educators understand and support pupils with a rare condition.

- As part of this work, an anonymous education survey was carried out to gather valuable feedback from parents and guardians about their experiences of the education system. The results and views shared by parents are documented in the Rare Disease Education Toolkit, which is freely available on the NIRDP website at this link: RD Education Toolkit.

- These findings were also presented to the Education Authority and have helped shape ongoing collaboration, including delivering awareness sessions for school staff, providing practical tools through the Toolkit, hosting listening sessions with families, and offering signposting to supportive resources. NIRDP is now listed as a useful resource for medical needs on the Education Authority's website, further increasing visibility and access to support services.

- During 2024–25, the Rare Disease Society at Ulster University secured funding for awareness events to increase student engagement with the rare disease community. Planned events will further integrate rare disease education into medical training.

Looking Ahead

- A comprehensive rare disease teaching framework is under development, aiming to synchronise curricula across various health disciplines and create a unified approach to rare disease education.

- A Rare Disease Education Passport initiative is underway, designed to provide structured guidance for schools and healthcare providers in supporting children affected by rare diseases.

- Work will continue in collaboration with the Education Authority to raise awareness of the impact of rare diseases.

Working Group 4: Research

Led by: Claire Cleland (PHA) and Professor AJ McKnight (QUB)

Actions: Research (Action 13)

Strengthening rare disease research capabilities and expanding funding opportunities continues to be a major priority for the Action Plan:

- Queen's University Belfast (QUB) co-leads the LifeArc Centre for the Acceleration of Rare Disease Trials, which brings together a consortium of three UK universities — Queen's University Belfast, Newcastle University, and the University of Birmingham — to boost the capacity and efficiency of rare disease trials. This £12m Centre will focus on helping patients access appropriate treatments earlier, delivering trials of new treatments using "one-stop," patient-friendly models, and facilitating equitable recruitment. It will accelerate the delivery of clinical trials for people with rare diseases and enable more rapid approval of new therapies for use in the NHS.
- NI also participates in the LifeArc–Kidney Research UK Centre for Rare Diseases, a major EU-funded initiative aiming to transform rare disease research across Europe. It provides significant collaborative research opportunities for NI and connects 13 paediatric kidney centres across the UK, with an ambition to align care, bridge datasets, connect scientists, and develop treatments.
- The European Rare Diseases Research Alliance (ERDERA) will be officially launched in September 2025. This major EU-funded initiative aims to transform

rare disease research across Europe and provide significant collaborative research opportunities for NI.

- Several key research roles have been funded, including project management, clinical trial facilitator, and patient logistics and experience officer positions, ensuring continued progress in NI's rare disease research landscape.
- Funding bids for an All-Ireland Rare Disease Research Centre are progressing, fostering greater cross-border collaboration. While competition is high, continued efforts aim to position NI as a leader in rare disease innovation.
- A Five-Nation strategy for rare disease clinical trials is being explored to ensure improved access to innovative therapies.
- Views on barriers to research participation were sought from people living with rare diseases as part of the Rare Diseases Patient and Carer Survey, to inform future activities that could make research studies more accessible, inclusive, and available.
- A doctoral training program has been established for rare diseases, helping to train the next generation of scientists, data analysts, and healthcare professionals. Resources are in place to support 12 PhD students at Queen's University Belfast, alongside 10 students in England. Several research projects are co-led with patients and families living with rare diseases, in partnership with charities such as the Northern Ireland Rare Disease Partnership, the Northern Ireland Kidney Research Fund, and the Fragile X Society.
- Investments in digital registry developments continue, including alignment with the developing UK rare disease clinical trial recruitment platform through the LifeArc award, charity investment, national screening initiatives, and the Cancer Strategy via the Small Business Research Initiative (SBRI).

- The All-Ireland Rare Disease Interdisciplinary Research Network (RAiN) continues to host events that strengthen connections between researchers, industry, government, local authorities, charities, and patient and caregiver partners across the island of Ireland.

Looking Ahead

- An in-person event formally launching the NI Rare Disease Clinical and Academic Network (RaDCAN) is planned for Autumn 2025, followed by a series of engagement opportunities.
- A rare disease research event, with a focus on clinical trials, is planned for Winter 2025–26.
- Scoping for an “undiagnosed clinic” is ongoing, focusing on practical logistics and funding.
- A concept paper is planned to establish an All-Ireland Rare Disease Research and Innovation Hub, bringing together a wide range of stakeholders and international collaborators to address a significant gap in current rare disease activities.

Working Group 5: Community Engagement & Patient/Carer Voice

Led by: NIRDP/DoH

- Active patient and carer involvement underpins the NI Rare Disease Action Plan. Key to this is mutual respect and recognition between all partners - patients, carers and families; advocates; researchers; and policymakers. Continued and strengthened collaboration between NIRDIG and NIRDP ensures that patient voices are heard and integrated into policy and action planning.

- Several successful “Power of Connection” consultation events were held by NIRDP throughout February and March 2025. These events reinforced the importance of community engagement and informed future iterations of the Action Plan (see below for more detail).
- Upcoming initiatives aim to strengthen engagement, including funding applications to support ongoing dialogue with the rare disease community.

“Power of Connection” Events: February-March 2025

- A series of rare disease engagement “Power of Connection” events were organised across Northern Ireland in February and March 2025 to coincide with Rare Disease Day. These events, each focused on different aspects of the Action Plan, facilitated vital conversations, shared powerful patient stories, and gathered feedback on the Action Plan and its progress from patients, carers, family members, academics, researchers, students, health professionals, policymakers (civil service and elected representatives), industry representatives, clinicians, allied health professionals, and other community organisations supporting people with rare diseases in Northern Ireland.
- The events were very well attended, with around 300 participants across all sessions - some of whom had never attended a rare disease-focused event before - highlighting strong public support for improving rare disease services across Northern Ireland. Each event (apart from the Stormont Long Gallery Reception) included a facilitated workshop using a “Think-Pair-Share” format, allowing several common themes and potential solutions to emerge.

Looking Ahead

- A report will be prepared based on the engagement events to update the Minister and help inform the next iteration of the NI Rare Disease Action Plan.
- Work will begin to take forward the key findings, such as the importance of peer support and local community connections, by intensifying efforts at the local level.

Rare Disease Day 2025

On 26 February 2025, the NIRDP, Queen's University Belfast, Ulster University, Rare Diseases Ireland, and the Departments of Health in Northern Ireland and the Republic of Ireland attended an all-island event in the Long Gallery, Parliament Buildings, to celebrate Rare Disease Day. The event was organised by NIRDP, and the programme included discussions on advancements and strategic planning in rare diseases.

Health Minister Mike Nesbitt (Northern Ireland) and RoI Minister for Health Jennifer Carroll MacNeill provided opening remarks via video link, setting the strategic context for the event. There were in-depth presentations on the Northern Ireland Rare Disease Action Plan and the National Strategy for Rare Disease in the Republic of Ireland.

A panel discussion titled "More Than You Can Imagine: The Power of Connection and Resilience" was facilitated by Prof AJ McKnight and Dr Suja Somanadhan and featured diverse perspectives and insights into the strengths and challenges faced within the rare disease community — as parents, patients, carers, and researchers.

Christine Collins MBE provided closing remarks. This annual event underscored the importance of collaboration, strategic action, the resilience of individuals and families affected by rare diseases, and the spirit of unity in marking Rare Disease Day across the island.

Other Related Actions

Mental Health

Action 22 of the NI Mental Health Strategy commits to ensuring that those with physical health problems leading to mental ill health receive the care and treatment they need. This does not mean providing dedicated mental health resources within physical health services, but rather creating effective pathways to allow individuals to access specialist support — ensuring patients get the care and treatment they need when they need it.

Although Action 22 has not yet been formally commenced, good progress was made during 2024–25 across a range of Mental Health Strategy actions that will help improve access to mental health services for all, including those with physical health conditions such as rare diseases.

These actions include:

- Implementation of a Regional Mental Health Service and establishment of a Collaborative Board;
- Commencing the implementation of recommendations from the review of the Mental Health workforce to inform a future workforce model for mental health services;
- Implementation of an Outcomes Framework through Encompass for Mental Health Services to underpin and drive improvements in service delivery;
- Development of an Early Intervention and Prevention Action Plan to promote mental health across the whole lifespan; and
- Implementation of the Regional Mental Health Crisis Service.

Genomics

Action 27 of the Northern Ireland Cancer Strategy commits to delivering genetic and genomic testing in line with NICE recommendations.

Cancer Strategy funding was secured for a three-year business case to expand the current provision of solid tumour genomic profiling by the Regional Molecular Diagnostic Service (RMDS), based in BHSCT, with investment starting in 2023–24.

The funding has enabled RMDS to develop a pathway to increase the number of patients accessing high-quality, comprehensive, and timely molecular characterisation of their tumours to inform therapeutics and clinical management. An additional 1,000 patients each year will now be offered access, bringing the annual total to approximately 3,500 patients.

Of particular significance for rare disease is that this new pathway will also identify individuals at risk of developing inherited cancers, including those predisposed to certain cancers because of a rare condition (e.g., TP53). By identifying inherited cancer risk early, effective screening, prevention, and early intervention measures can be implemented to diagnose cancer as early as possible, provide cascade testing to family members, offer genetic counselling, and improve patient outcomes.

Northern Ireland was the first region in the UK to undertake this revolutionary pathway at a population scale, enabled by investment from the Northern Ireland Cancer Strategy. However, further expansion of the service is subject to available funding.

Looking Ahead: 2025-26

NI will continue to ensure that the needs of rare disease patients are recognised in wider policy development, for example, across ongoing IT developments. The completed roll-out of the encompass solution will transform rare disease patient and service user journeys, care provision, and information flows across the entire HSC system in NI. By working more effectively and efficiently through regional standardisation and best practice, the long-term ambition is to create better experiences for those receiving, using, and delivering services — including the rare disease population in NI.

Work will continue to maintain and build on the progress made, with the Department represented on the UK Rare Diseases Framework Board and the UK Rare Diseases Forum, hosted by the Department of Health and Social Care (DHSC), which collectively oversee the development and delivery of Framework priorities across the UK.

NI will work with counterparts in England, Scotland, and Wales to progress rare disease policy on a UK wide basis. Importantly, the voices of people with lived experience — and those who care for them — will remain central to these plans as we consider the next iteration of the Framework.

Continued collaboration with Rare Diseases Ireland and other all-island partners will remain a key focus, with joint initiatives such as the All-Ireland Children and Young Adults Research Advisory Group (CRAG) helping to strengthen cross-border learning and service development.

Conclusion

A great deal of progress continues to be made against the actions in the Northern Ireland Rare Diseases Action Plan, and the achievements described in this report would not have been possible - just as in previous years - without the valuable input and engagement of our stakeholders. The voice of the rare disease community, through the efforts of the Northern Ireland Rare Disease Partnership, underpins the Action Plan, and continued collaboration and support are deeply appreciated.

Sincere thanks also goes to our NIRDIG and Working Group members, whose hard work, drive and commitment to the rare disease agenda has been invaluable in progressing and maintaining the impetus of the Action Plan. Appreciation also goes to rare disease policy officials and counterparts across the four nations and Ireland for sharing their collaboration, ideas, and experiences.

The Department and NIRDIG remain committed to implementing the Action Plan in full and to building on community and stakeholder engagement during 2025–26, ensuring that the Action Plan continues to deliver on the issues that matter most to the rare disease community in Northern Ireland.

Links/Further Information

The NI Rare Disease Action Plan

Progress Report 2022-23

Progress Report 2023-24

The UK Rare Diseases Framework

NI Rare Disease Partnership

All-Party Group on Rare Disease

RD Education Toolkit