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Performance and Improvement

Wales Rare Disease Action Plan Annual Progress Report 2024-2025

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Contents

This progress report details the progress made by the Wales Rare Diseases Implementation Network (RDIN) in the third and final year of the Rare Diseases Action Plan for Wales, published in June 2022[1].

[1] Action Plan 2022 - 2026 - NHS Wales Executive

1. Background / Strategic Context

1.1 On 9 January 2021, the UK Rare Diseases Framework[2] was published and included a joint Ministerial foreword by all four respective UK Health Ministers.

1.2 The Framework was based on the outcomes of the 'National Conversation on Rare Diseases', launched in 2019.[3] The conversation gathered views from across the rare disease community on the major challenges faced by people affected by rare conditions across the UK.

1.3 The Framework outlines the UK's priorities for rare diseases over a five-year period.

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

1.4 Underpinning themes within the Framework are:

- Patient voice
- Collaboration
- Research
- Data and technology
- Wider policy alignment

[2] https://www.gov.uk/government/publications/uk-rare-diseases-framework

[3] https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework#annex-a

1.5 Whilst the Framework remains a UK-wide document, each of the four UK nations operates its own delivery or implementation group responsible for drafting and monitoring nation-specific action plans. Tailored to the needs of individual populations, whilst working together through the UK Rare Diseases Framework Board, the national teams ensure as much alignment across the four nations as possible. The implementation group for Wales is the Wales Rare Diseases Implementation Network (RDIN).

Rare Diseases Networks in Wales

1.6 The Wales Rare Diseases Implementation Group was established in 2014 when the Welsh Government first instituted the Welsh Implementation Plan for Rare Diseases[4] (published in 2015). It affirmed the Welsh Government's commitment to both empowering those with a rare disease and ensuring those affected by any kind of rare disease had timely access to high quality pathways of care. Since the three updated plans in 2017, and more recently in 2022 and 2023[1], the group worked with key stakeholders within NHS Wales and its strategic partners, the third sector, the Welsh Government, academia, and patient representatives to provide support and expertise to those providing care for individuals in Wales with rare diseases.

1.7 The group transitioned into the NHS Wales Executive and then NHS Wales Performance and Improvement (NHSPI)[5] in 2025 and within these new structures the implementation group became the Rare Disease Implementation Network (RDIN). RDIN is hosted by the National Strategic Clinical Network for Child Health (Child Health Network) which is led clinically by Dr. Claire Thomas. RDIN is supported by Rhiannon Edwards as Network Support Manager and Dr Jamie Ducker as the National Rare Disease Clinical lead. Dr Duckers also represents rare diseases in the Child Health Strategic Clinical Network Leadership Group. The Rare Disease Clinical Lead for RDIN reports to the Clinical Lead for Child Health, reflecting the relationship in the NHSPI between strategic clinical networks and implementation networks.

[4] Welsh Implementation Plan for Rare Diseases (europlanproject.eu)
[5] https://performanceandimprovement.nhs.wales/

1.8 The RDIN Clinical Reference Group meets quarterly and is chaired by Professor Iolo Doull, Medical Director of the national NHS Wales Joint Commissioning Committee (NWJCC). Agenda items include information regarding national and international priorities and progress in implementing the Wales Rare Diseases Action Plan, and the group seeks to act as an advocate for those affected by rare diseases.

1.9 The Child Health Network via RDIN is responsible for implementing the action plan, overseeing the delivery of strategic and national pieces of work, and supporting Welsh health boards and trusts to develop, deliver and report on their integrated medium-term plans.

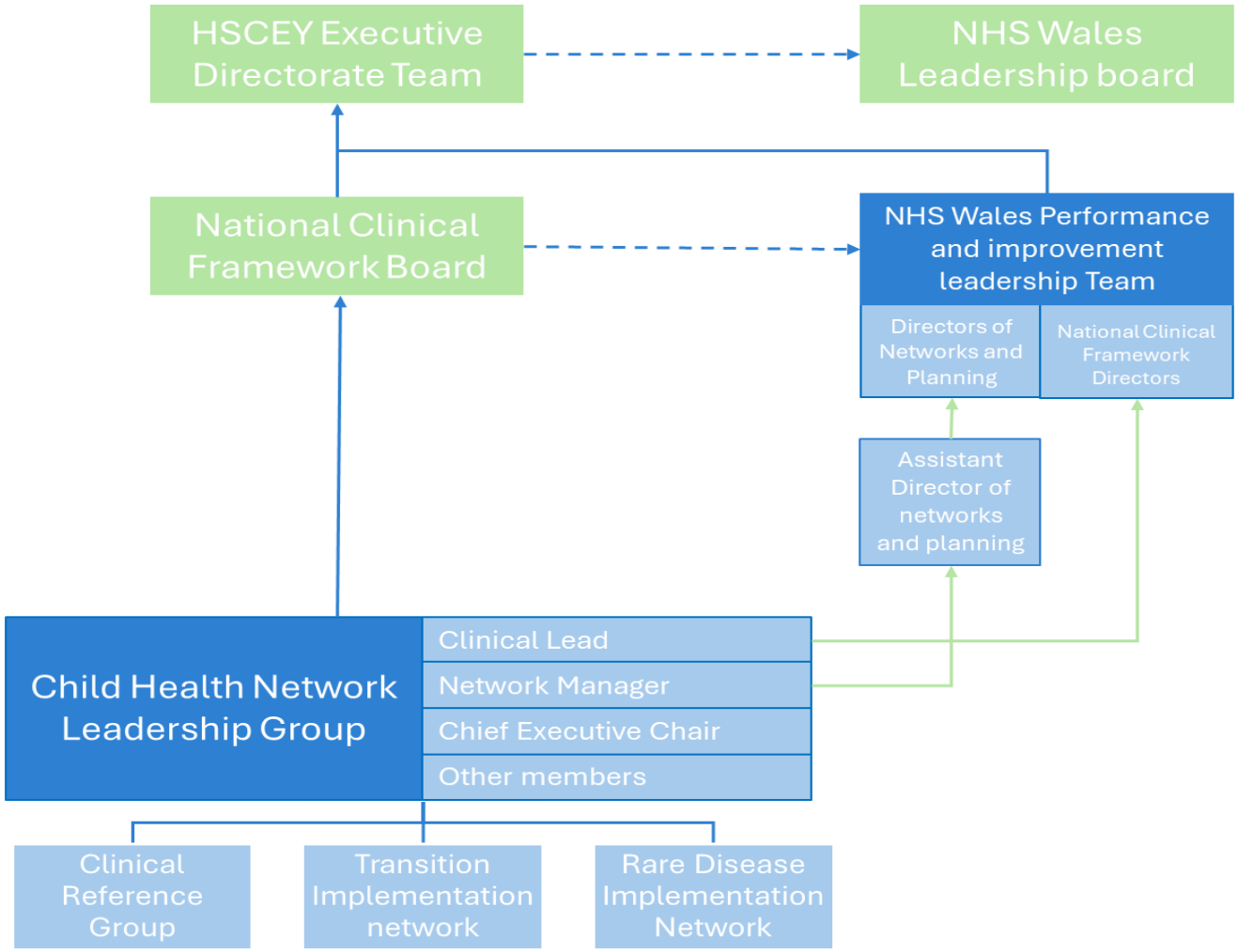

Figure 1 – Strategic Clinical Network Structure, NHS Wales Performance and Improvement

2. Wales' Rare Disease Action Plan

2.1 In June 2022, the Welsh Implementation Plan was replaced by the Wales Rare Disease Action Plan, following the development of the UK Rare Diseases Framework.

2.2 Through dedicated funding from the Welsh Government to establish an implementation group for rare diseases, the appointment of a Clinical Lead for Rare Diseases (the first across the four nations) took place, alongside a Network Support Manager working in partnership with NHSPI to support the implementation of the Wales Rare Disease Action Plan, including twenty-nine actions across the four priorities.

2.3 Data is considered a priority for RDIN, therefore two workstreams focus on rare disease registers, supported by the Congenital Anomaly Registration and Information Service (CARIS) and a rare disease data dashboard, supported by Digital Health and Care Wales (DHCW). NHSPI's current workplan priorities also include the delivery of a mapping report of current rare disease activity in NHSPI and horizon scanning for partnership working across networks and programmes for 2025/26.

2.4 Key strategic alignment with the Genomics Delivery Plan for Wales[6] (launched in December 2022, reflecting the Welsh delivery of UK strategy for genomics and Genome UK: The future of healthcare[7]) focused elements of priority one actions, to support challenges of capacity within our workforce. The Genomics Delivery Plan is currently being refreshed for 2025-2030.

2.5 The robust engagement prior to the launch of the refreshed Wales Rare Disease Action Plan in 2023 enabled the plan to reflect the needs of the rare disease population in Wales and the patient voice continues to be embedded into discussions via RDIN's Clinical Reference Group.

2.6 In January 2025, the annual progress plan was published, with a rollover of the Wales Rare Disease Action Plan agreed with the Welsh Government for 2025/26.

[6] https://www.gov.wales/sites/default/files/publications/2022-11/genomics-delivery-plan-for-wales_0.pdf
[7] https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare

3. Progress Summary

3.1 In 2024, The development of the RDIN Leadership Group focused on producing a clear vision statement for the function of the network. Three priorities were decided (research, the digital rare disease hub, and data) and continued until April 2025, when the new 2025/26 workplan was agreed by the NHSPI. This focused activity across data and insight.

3.2 Over the past year the Data and Surveillance Group has continued supporting projects on DiGeorge Syndrome and developing the adult rare disease register, using RDIN's Clinical Reference Group for the prioritisation of projects associated with this group. This work will be enhanced by a newly appointed research funded data analyst post (https://www.lifearc.org/project/lifearc-translational-centres-for-rare-disease/) within the CARIS team. This post will support the continued work to build an all-age rare disease register for Wales and associated Patient Reported Outcome Measures (PROMs) datasets. In addition, RDIN has led the development of a rare disease data dashboard, in partnership with DHCW, NHSPI's Value Transformation Directorate and CARIS. This will provide a visual representation of healthcare utilisation across the NHS Wales secondary care and ambulance system, for those diagnosed with a rare disease and provide much-needed data about the healthcare journeys experienced by our population. This is the first of its kind in the United Kingdom and will support strategic prioritisation when it is launched in 2026.

3.3 Data surveillance in rare diseases is insufficient due to poor clinical coding and misdiagnosis. Therefore, PROMs can create a significant opportunity to understand the impact of patient pathways in rare disease care and provision. Linking these within the newly established rare disease data dashboard, via the nationally procured Promptly platform, will enable visualisation and support strategic prioritisation and planning. RDIN has facilitated discussions enabling access for Cardiff and Vale University Health Board (CVUHB) rare disease clinical teams to introduce PROMs in both Duchenne Muscular Dystrophy and Primary Ciliary Dyskinesia. Linking outcomes-based data to health utilisation data will help to map rare disease pathways and the impact of current service provision.

3.4 Access to research, for many, is the only opportunity to access therapies and treatments that may improve survival rates and quality of life, with only 5% of rare diseases having a validated treatment. Currently, Wales has low levels of rare disease research funding[8], despite having robust mechanisms to enhance access and development of rare disease focused studies.

[8] https://performanceandimprovement.nhs.wales/functions/networks-and-planning/rare-diseases/guidelines-and-reports-docs/intelligence-report-rare-diseases-research-landscape-in-wales/

The Wales Rare Disease Research Network was launched in January 2025 via an online webinar to bring key stakeholders together, including people with lived experience and industry partners to build a community of interest to address these challenges. RDIN led the coordination of a stakeholder list (circa. 210) and continues to support the network's activities.

Partnering with the Welsh Government, five Welsh universities, the Life Science Hub and Health and Care Research Wales (HCRW), RDIN received funding from the Wales Innovation Network in April 2025 to deliver a Wales Rare Disease Research Network meeting to promote collaboration and networking. The inaugural event[9] took place at Swansea University Bay Campus in September 2025 and was attended by 98 individuals (with 10% people with lived experience). 13 academic and advocate organisation stands were included, as well as poster and oral presentation competitions. The feedback from participants was overwhelmingly positive and community voting is being analysed to determine the future focus of the research network.

3.6 In June 2025, The UK's first formally commissioned Syndrome Without A Name (SWAN) clinic, which had previously been funded as a three-year pilot by the Welsh Government, was given recurrent funding by the NWJCC. This is a tremendous boost to Wales and the SWAN team, and the clinic provides a unique service for the benefit of Welsh patients suspected but not diagnosed with a rare disease.

3.7 The Bevan Commission Exemplar Programme (https://bevancommission.org/programmes/bevan-exemplars/) continues to support the Wales Digital Rare Care Centre as the focal point to improve transition care for those with rare diseases. The leadership of this project has now transferred to CVUHB, and the work has been recognised nationally and internationally, winning in the category of Excellence in digital health and innovation at the Welsh Healthcare awards and being shortlisted by the Health Service Journal for their patient empowerment digital award.

[9] https://healthandcareresearchwales.org/about/events/rare-diseases-research-network

4. Progress by Priority

In 2025/26, progress on the implementation of the Wales Rare Disease Action Plan has been steady.

4.1 Helping patients get a final diagnosis faster.

4.1.1 Increase Whole Genome Sequencing (WGS) testing for rare diseases.

The Pregnancy Related Rapid Sequencing Service (PRRS) was launched in March 2023 using Whole Exome Screening or WGS trio analysis (duo analysis available in exceptional circumstances only). The poster of the service launch was accepted for the Association for Clinical Genomic Science (ACGS) conference 2025, which identified that clinically significant variants were found in 8 of 16 cases sequenced in 2023.

4.1.2 Ensure a consent strategy is developed that enables researchers to securely and safely access routine genomic data generated by All Wales Medicine and genomics Service (AWMGS) for translational research purposes.

The Wales Gene Park (WGP) Genomic Consent Manager (Rhys Vaughan) working with the Genomic Partnership Wales (GPW) Patient & Public Sounding Board, AWMGS colleagues, and other stakeholders have developed a genomic research consent system: the All-Wales Genomics Databank (AWGDB). The NHS Research Ethics Committee has granted approval for AWGDB, however, there is a delay for approval from the Cardiff University/CVUHB Joint Research Office. The next step following this approval will be a pilot project to recruit 100 NHS patients and family members to the AWGDB. Following evaluation of the pilot and further engagement with patients and the public, funding opportunities and regulatory approval will be explored to scale up recruitment. The long-term vision is that all NHS patients in Wales undergoing genomic testing will be offered the opportunity to consent to research access to their data and to be recontacted for future research participation.

4.1.3 Engagement with Health and Care Research Wales (HCRW) to ensure access to research studies for rare diseases patients.

The new Wales Rare Disease Research Network is partnering with HCRW to highlight the benefits of carrying out research in Wales targeted at co-creation of research with people with lived experience, clinical researchers and industry. Considerations of a one point of access umbrella organisation for rare disease research is being considered, with webpages currently under development in HCRW.

4.1.4 Ensure validation of a whole transcriptome service which will enable better understanding of Ribonucleic acid (RNA) sequences to determine if a Deoxyribonucleic acid (DNA) sequence is turned on and whether proteins have changed.

Please see work undertaken by all-Wales Genomics Laboratory (AWGL) response in 4.1.1.

4.1.5 Co-produce research questions with service users, to bring Rare Disease research closer into policy and practice.

The new Wales Rare Disease Research Network will develop collaboration between researchers, clinicians, academics, NHS supported organisations and industry partners. The first in-person network event was held in September 2025 at Swansea Bay University Campus and was financially supported by the Wales Innovation Network. Leadership and infrastructure discussions are now being facilitated to support patient-centred rare disease research in Wales. Themes include: mechanisms of collaboration, awareness, recruitment for research and considering systems and care pathways that would better meet the needs of people with rare diseases. This work will now directly support preparation for a strong proposal to a Great Ormond Street Hospital Children's Charity (GOSH Charity) grant opportunity, which is committed to supporting ambitious research that will lead to a transformative impact on the lives of children with rare and complex health conditions.

RDIN also supported calls for funding to support a James Lind Alliance Priority Setting Partnership (JLA PSP) for priorities in rare disease research, which will be led in CVUHB by the newly established Rare Disease Consultant Nurse post. Funding for the JLA PSP was accessed from HCRW via the Rare Disease VPAG funding stream.

4.1.6 Increase research activity in Paediatric Rare Disease research.

The SWAN clinical team, supported by partners, accessed VPAG funding to develop the Wales Rare Disease Research Hub, which will be hosted by the Child Research Centre in CVUHB. The funding was agreed in May 2025 and will enable the governance structures to be developed to embed greater access to funding to support rare disease research into paediatric settings in Wales.

Prevention and Early detection

4.1.7 Establish a public health and screening system in Wales that uses genomics to strengthen the current biochemical screening, diagnostic and care pathways in those at high risk.

- Planning for additional conditions being recommended for newborn screening programmes which have rare disease genomics consideration as part of the test such as Spinal Muscular Atrophy (SMA) and Severe Combined Immune Deficiency (SCID). These are currently undergoing 'in-service' evaluations in England or are being planned. Public Health Wales screening services are aware of this, as part of the UK National Screening Committee, and Dr Graham Shortland, who is a committee member, provides updates within the RDIN's quarterly Clinical Reference Group meetings.
- Planning for consideration of further in-service evaluations of other potential newborn conditions (which have genomics considerations), as part of the test to build up the evidence base to enable recommendations in Wales to be made based on benefits and potential harms.

4.1.8 Explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices.

Research has been conducted regarding genomic testing, reproductive medicine and patient choice and is awaiting publication. When this is completed, conversations will take place about how to bring this into routine care as a possible pilot.

4.1.9 Non-Invasive Prenatal Testing (NIPT) will be expanded to other reproductive pathways to improve patient outcomes and optimise resource utilisation.

Publication in draft to highlight collaboration between (1) Department of Metabolism, Digestion and Reproduction, Imperial College London, Hammersmith Hospital Campus, Du Cane Road, London (2), All Wales Medical Genomics Service and (3) West Midlands Regional Genetics Laboratory, Birmingham Women's & Children's Hospital, Birmingham on NIPT. Paper summary; Aneuploidy causes 50-60% of miscarriages, but detection relies on cytogenetic testing of miscarriage tissue. Cell-free fetal DNA (cffDNA) could offer an alternative non-invasive method but requires validation in first trimester miscarriage. In this prospective multi-centre study participants with an ultrasound diagnosis of miscarriage <14/40 gestation were recruited from two London hospitals between February 2021 and November 2023. Maternal blood was collected for genome-wide cffDNA sequencing while miscarriage tissue remained in utero, cytogenetic testing of miscarriage tissue (SNP microarray, QF-PCR, karyotyping or BACS-on-beads) was used as the reference standard.

4.1.10 Equal access to genomic testing across the UK. Provision within Wales or referral outside of Wales.

The AWGL is an NHS Wales national genomics service commissioned by the NWJCC against the Specialised Services Specification: CP99. This specification states that referrals for genetic testing will be accepted for patients that meet the testing criteria for the NHS England National Test Directories and local NHS Wales protocols. Three services alone require massive infrastructure investment and rapid WGS tests for critically ill babies and children (R14) (initiated April 2020). Further developments have extended WGS to in-house Congenital Malformation and Dysmorphism Syndromes (R27) and Intellectual Disability (R29) services. In addition, AWGL is delivering Early Onset or Syndromic Epilepsy (R59) and Primary Immunodeficiency or Monogenic Inflammatory Bowel Disease (R15) by whole exome sequencing (WES). AWGL enables this provision through both providing and purchasing specialist testing services for rare diseases. With agreement from the NHSE Genomics Unit, the genomics laboratory at Exeter has recently agreed to provide specialist rare disease services for NHS Wales. This agreement is important as the sourcing of services from external UK providers has led to a significant increase in costs and additional cyber security risks associated with non-NHS providers when seeking to provide an equitable service to the NHSE Test Directory.

4.1.11 All nations should develop actions which support diagnosis and care for non-genomic conditions.

Swansea University is leading the 'Lipidomics and metabolomics' node as part of the Rare Disease Research UK, a UK-wide platform funded by the Medical Research Council (MRC) and the National Institute for Health and Care Research (NIHR). The Swansea team collaborate closely with the SWAN clinic providing access to state-of-the-art research technology for the investigation of non-genomic conditions as well as genetic rare disease. RDIN quarterly meetings provide a mechanism for communication of progress to wider rare disease stakeholders.

Service/Digital/Technical infrastructure

4.1.10 Increase awareness of additional UK genomic tests newly commissioned within the genomic test directory for rare and inherited disease.

Wales Gene Park (WGP) continue to highlight awareness of genomic tests and pathways of care for those with genetic rare diseases, via genomic cafes, university sessions, and conference activities. In addition, Dr Graham Shortland represents Wales at the UK National Screening Committee and alongside public health newborn screening leads, disseminates new genomic testing authorisation and information to RDIN stakeholders and NHSPI's Networks and Planning Directorate, as appropriate.

4.2 Increasing awareness of rare diseases amongst healthcare professionals

In addition to the actions detailed below, in priority two of the UK Rare Diseases Framework, RDIN led a NHSPI survey to understand current rare disease focused work across networks and programmes and raise awareness of rare disease priorities and opportunities in NHS Wales. This will form the foundation to focus additional activity across the organisation in workplans in 2026/27.

In collaboration across the UK, two focused workstreams had Welsh representation.

- The UK Rare Disease Forum Independent Advisory Group for Priority Two was initiated in 2025 because of the need for a joined-up approach across the four nations for raising awareness of rare diseases, and this was facilitated by Medics 4 Rare Diseases. The final report was delivered to the UK Government's Department of Health and Social Care, to disseminate across the four nation's policy teams to recommend actions for the new period of rare disease planning activity.
- The UK Rare Disease Forum Independent Advisory Group for Rare Disease Quality Standards. This task and finish group has now transitioned into a working group to focus activity from scoping and mapping exercises across the UK, into quality statement recommendations for the National Institute for Health and Care Excellence (NICE) for their endorsement and development into Rare Disease Quality Standards. RDIN supported this work through representation from nursing and undiagnosed rare disease communities.

Lead clinician for rare disease

4.2.1 Monitor ongoing role and work programme of Clinical Lead and Clinical Champion for rare diseases to raise profile of rare diseases.

The national Clinical Lead for Rare Diseases continues to engage with colleagues, industry and third sector organisations across Wales as well as internationally to improve awareness of rare diseases and the impact of living with these conditions. Highlighting opportunities for clinicians to support new ways of working and support partnership working across sectors, Dr Duckers has contributed to identifying rare diseases as an emerging priority across the health and social care sectors, promoting the need to understand patient pathways and access to specialist services, and to continue implementation of the Wales Rare Disease Action Plan.

Education and Shared Learning

4.2.2 Survey qualified Health Care Professionals (HCP) undergraduates on their understanding and learning needs in rare disease.

RISE study in undergraduate students is nearly ready for submission, with additional data capture and support from the Medical Schools Council ongoing, as there was limited distribution across the UK initially. The RISE study in HCP (allied health professionals and nurses) has been developed, however, the absence of a Principle Investigator (PI) in

this field to drive forward the introduction of the study has delayed implementation. Agreement in principle for support to disseminate the survey to professional groups by Health Education and Improvement Wales (HEIW) was planned in 2024.

4.2.3 Use results to develop training and development plan from baseline information on HCP understanding of rare diseases.

A RISE related project is looking at the learning needs of rare disease undergraduates (medical) in more detail through bespoke focus groups. Data capture and transcripts are complete, and the analysis phase is in progress. This work will be used as learning within HCP's understanding of rare diseases.

4.2.4 Incorporate rare diseases module in the undergraduate curriculum for medical students.

The Genomics Partnership Wales Workforce & Training Implementation Group's modules for undergraduate medical students are now in their fourth year and fully established at Cardiff University. However, HEIW currently has no mechanism to influence or shape medical student curricula at a local level.

4.2.5 Ensure development of specialist consultant roles - interest and confidence.

The Wales Rare Disease Research Network is building awareness of rare diseases into common disease areas. Scoping workstreams and potential collaborations within NHSPI will lead to partnership working across existing areas of interest, including Neurological, Musculoskeletal (MSK), Diabetes, Mental Health and Palliative end of life care (PEoLC).

4.2.6 Clinical Nurse Specialists – build understanding of paediatric and adult CNS workforce.

RDIN supported discussions to develop a Rare Disease Consultant Nurse post in CVUHB to support ongoing conversations regarding the support and mapping of the nurses working within rare disease clinics in Wales. Current work is ongoing to highlight career pathways, embedding training and capability mapping for nurses alongside HEIW workstreams in genomic medicine.

4.2.7 Continue to develop active partnerships with patients and patient advocacy groups (PAGs).

Building active partnerships within the Wales Rare Disease Research Network between professional groups and patients and advocacy groups will continue to provide the foundation for patient-centred research in Wales. Wales' leadership of the Global James Lind Alliance Rare Disease Priority Setting Partnership will enhance these opportunities to

focus industry and research organisations around the top 10 priorities set by people with lived experience. RDIN has medical and nurse representation on the UK Quality Statement task and finish group to support and develop NICE rare disease quality standards, to highlight NHS Wales quality and safety principles of care for rare diseases patients.

4.2.8 Recognise and celebrate rare disease day in secondary and primary care.

Genetic Alliance UK hosted an evening event in Cardiff Bay which was attended by the NHSPI Clinical Director and was opened by the Cabinet Secretary for Health and Social Care to recognise Rare Disease Day 2025. Agenda and presentations were supported by RDIN, and included a person with lived experience, information regarding the Wales Digital Rare Care Centre and the Wales Rare Disease Research Network.

4.2.9 Improve health professional awareness through joint working between primary/secondary and tertiary care, such as local pilot (Hywel Dda) Webinars for General Practitioners from AWMGS.

Medics for Rare Disease Ambassadors delivered training and education to undergraduate medical students at Swansea University in summer 2025, supported by RDIN with additional information about the UK Rare Diseases Framework. RDIN is also continuing to support CVUHB in developing its one-point-of-access Digital Rare Care Centre for education and training purposes, in partnership with industry (Carecircle[10]). The NHSPI rare disease scoping report has identified opportunities to collaborate with the Primary Care National Programme in NHSPI.

4.2.10 Ongoing programme of WGP education and engagement with HCP and students including Genomic Counselling role (across Welsh Health Boards and HEIs) including precision medicine.

Wider workforce understanding of rare diseases is within scope of the Genomics Workforce Plan being led by HEIW. The mainstreaming and genomics educational resources has also been included within the Genomics Workforce Plan, which was published in autumn 2024. RDIN has been involved in additional workstreams to build a capability framework as part of this plan and focus attention on rare disease training and education opportunities. RDIN has also represented rare diseases in ongoing discussions on pathway mapping for complex/novel therapies into practice (Coalition of the Willing), led by Advanced Therapies Wales (ATW) and the All-Wales Therapeutics and Toxicology Service (AWTTS). This document is awaiting endorsement by strategic leadership in Wales.

10. https://www.carecircle.org/

Improving Awareness of Rare Diseases with Data

4.2.11 CARIS team expansion to include adults affected by rare conditions.

Projects in 2025 included reporting on the validation of the Di George adult rare disease register. The first surveillance report will be presented in collaboration between CARIS and RDIN in January 2026.

4.2.12 Confirm and regularly share the agreed metrics to be used for rare diseases patients, providing data to each UHB/Trust to raise awareness of performance in the UHB's/Trusts by WRDIG.

Welsh health board-specific data on healthcare utilisation is included in the newly developed rare disease data dashboard. This dashboard will include data about patients held within the CARIS register, on unplanned admission data, the Welsh ambulance service, in line with the 'healthy days at home' strategic direction of the Primary Care Strategic Programme. Geographical data will be developed in collaboration with Welsh health boards to increase awareness of resource impacts for regional services and support Integrated Medium-Term Planning (IMTP) processes.

4.2.13 Consider collection of rare disease data at both a national All-Wales level drilled down to lower-level geographies (such as university health board/trust footprint), where numbers of patients with specific diseases allow.

RDIN supports CARIS via the data subgroup to act as a clinical governance route for embedding appropriate projects within CARIS to provide impact and validation for the rare disease register.

4.3 Better coordination of care

Pathways of Care

4.3.1 Ensure implementation of transition guidance with all paediatric patients transitioning to adult services should have a named worker and digital care plan linked to a patient passport.

All seven health boards in Wales were given feedback forms (annex 6 from the Welsh Government Transition and Handover Guidance[11]) to be completed by April 2025 to understand adherence to transition handover guidance, via the All-Wales Nursing Leadership forum. Five health boards submitted a response. Included in the feedback were direct questions regarding named workers and health passports.

Although there were reported pockets of adherence to the guidance, with some use of health passports and other recommended practice such as joint clinics and transition practitioners (often a youth worker or nurse and usually in specialities such as Cystic Fibrosis or Diabetes), there was no general access to a named transition worker for all patients to date (there were some plans underway in Aneurin Bevan University Health Board and Cwm Taf Morgannwg University Health Board). This insight will form part of planning processes within the transition implementation network, Child Health Network.

4.3.2 Establish Rare Diseases as a "Community of Practice" and develop example/exemplar clinical pathways for rare disease conditions, including MDT involvement.

The UK Rare Disease Quality Standard task and finish group (UK Rare disease forum) has transitioned into a working group to facilitate the next steps for NICE endorsed quality standards; these will become a mechanism to support assessments about the quality and safety of rare disease service provision.

RDIN has carried out scoping work to understand the extent of rare disease focus across current NHSPI networks and programmes and is developing into an expert reference group to support network priorities. The scoping report will be submitted to the Welsh Government, via NHSPI reporting processes to inform RDIN's annual workplan for 2026/27.

SWAN Clinic

4.3.3 Continue to build the establishment and assess/evaluate SWAN clinic.

The UK's first formally commissioned Syndrome Without A Name (SWAN) clinic was previously funded as a three-year pilot by the Welsh Government.

[11] https://www.gov.wales/sites/default/files/publications/2022-02/transition-handover-guidance-children-adult-services_2.pdf

In June 2025, the service was given recurrent funding by the NWJCC. This is a tremendous boost to the SWAN team and provides a unique service for the benefit of Welsh patients suspected but not diagnosed with a rare disease.

The SWAN clinic is hosted by CVUHB and has been set up to improve pathways for adult and paediatric patients living with rare, undiagnosed conditions across Wales. As part of the improved pathways, the SWAN Clinic aims to shorten the time that people living with an undiagnosed condition wait for a diagnosis ('diagnostic odyssey'), improve medical knowledge and foster research. The clinic offers hope to children and adults with syndromes so rare they have not had a diagnosis confirmed, or they have a new syndrome that does not have a name. With referrals of over 100 patients (adult and paediatric) seen across Wales, and diagnostic rates varying according to where patients are on the pathway, the clinic has achieved an overall diagnostic rate of 30%. The service has been evaluated using PROMs with excellent results, particularly enhancing patients experience of care co-ordination, and has been published in a peer review journal[11]

4.3.4 Understand the usefulness of PREM and PROM collation to develop enhanced service provision.

PROMs and PREMs collection is being expanded to Duchenne Muscular Dystrophy and Primary Ciliary Dyskinesia in Wales. Ongoing evaluation will consider the population level reporting required to impact on service provision, and how PROMs can be included in the rare disease data dashboard to understand the impact of service provision on outcome measures.

Digital Patient Record

4.3.5 Establish an easily used "app" to enable a "patient passport" for rare disease patients.

CVUHB is piloting a Wales Digital Rare Care Centre[12] as a one-point-of-contact support mechanism for those impacted by rare diseases and their support networks and professionals, including education, workplace, social care and community care.

11 https://www.sciencedirect.com/science/article/pii/S2950008726000013
12 https://bevancommission.org/falling-off-a-cliff-a-digital-carabiner-for-rare-disease-young-people-and-their-families-transitioning-to-adult-care/

Information governance has been approved to develop a rare disease health tracker and patient passport within the Wales Digital Rare Care Centre, which is expected to be evaluated over the next 2 years (October 2027). This will build on the Camrare paper passport[13] and utilise artificial intelligence to support improved communication.

Mental Health Services

4.3.6 Ensure the mental health needs of rare disease patients and carers are considered as part of the overall mental health strategy for Wales and consider whether further guidance is needed such as a good practice guide for rare disease patients.

Understanding current activity in the national Mental Health Strategic Programme regarding rare diseases has been a theme of the rare disease scoping report across NHSPI. This will form the basis of ongoing collaborations across NHSPI and support gap analysis of current access and highlight good practice.

Equity, Diversity and Inclusion

4.3.7 The Wales Rare Diseases Action Plan will consider equity, diversity and inclusion (EDI) throughout the refresh of the development and implementation of future Wales Rare Disease Action Plan.

RDIN continues to be a partner in the Equity, Diversity and Inclusion in Research Association (EDIRA), representing Wales' rare disease population. In addition, RDIN has supported focused activities on rare diseases that impact ethnic minority groups, such as Sickle Cell (community health pathway) and Primary Ciliary Dyskinesia (PROMS).

13 https://www.camraredisease.org/rare-patient-passport/

4.4 Improving access to specialist care, treatment, and medicines

Access to Medicines and Treatment

4.4.1 Ensure continued access to orphan and ultra-orphan medicines in Wales.

RDIN has represented rare disease communities on the national group on pathway mapping for complex/novel therapies into practice (Coalition of the Willing). A national process and governance structure has been presented for endorsement to AWMSG, ATW, NWJCC, CEMT, the Planned Care Programme Board, prior to the National Oversight Panel, and three Operational Planning and Delivery Groups. There will be a disbanding of the national pathway mapping group upon implementation.

4.4.2 Ensure horizon scanning for new medicines for patients in Wales to allow timely awareness of new products and availability of new medicines.

See above new pathway mapping for complex/novel therapies into practice. A risk was highlighted by RDIN and has been noted by the national group, that the lack of a National Rare Disease Leadership Board (operationally similar to the National Cancer Leadership Board) will impact support for horizon scanning and implementation of the pathway.

4.4.3 Monitor uptake of new rare diseases medicines and prescribing.

Blueteq systems is being used by NWJCC for monitoring the use of new rare disease medications. There is scoping work underway to embed this information into the rare disease data dashboard.

4.4.4 Continue to develop improvements in the monitoring of use of medicines for patients with rare diseases including Blueteq.

Combined with 4.4.3.

4.4.5 RDIN will build actions which support the use of repurposed and off-label medicines and devices.

The All-Wales Toxicology and Therapeutics Service (AWTTS) and Health Technology Wales (HTW) horizon scan for new therapies and devices, which impact rare disease patients, as well as other members of the Welsh population. Due to limited capacity, RDIN has been unable to focus work on understanding any gaps that exist and will consider this as an opportunity in 2026/27.

4.4.6 RDIN will observe opportunities to understand how pharmacogenomics can improve the effective management of those with rare diseases in Wales.

RDIN continues to link to the national pharmacogenomics group to act as a clinical expert group, if required.

Access to specialist Care

4.4.5 RDIN will work with the NWJCC and Health Education and Improvement Wales (HEIW) to ensure appropriate consultant specialist services in Wales.

Consultant Specialist Services are not commissioned by HEIW, which commission postgraduate medical training and support individuals themselves to develop interests within their training, some of which may be in rare diseases. Mapping of opportunities to develop interests in rare diseases for clinicians across NHSPI's networks and planning is intended to lead to an increased understanding of pathways to specialist services in rare diseases.

5. Opportunities and Challenges

The extension of the UK Rare Diseases Framework by 12 months (to 2027) gives RDIN and its stakeholders the opportunity to reflect on and review our progress over the last four years as the Rare Disease Implementation Group and Network within NHSPI, to consolidate our learning and build a renewed focus for the future. This includes how we as a network engage and collaborate within our organisation and how we support rare disease focused priorities alongside condition or life course networks and programmes. The current workplan focuses on scoping these pieces of work and builds a greater awareness within the organisation of the impact of rare diseases on the Wales population, and how collectively they are common in terms of perceived impact on health and social care in Wales. The cross-cutting

themes identified in many patient-centred reports from our third sector colleagues highlight the importance of mental health, access to treatments and specialist services, communication and coordination of care, and additional insight from our colleagues recognising a lack of awareness in our clinical environment, including primary care. These aspects of the framework are yet to be addressed significantly, and our improvements are yet to reach the people we hope to support.

5.1 The complex system-wide challenges that are faced by many in our population are not sufficiently visible to health and care strategic leaders due to poor coding and challenges in mapping pathways of care. In Wales, we are proud to have developed the first rare disease data dashboard, and although it does not map primary and community care services, it is hoped it will highlight the common journeys our patients and their support networks experience on a day-to-day basis within planned and unplanned care in secondary care services. This currently only observes those within the CARIS and rare disease registers, but with the support of research grant funding, these registers will build across the UK to visualise the population we aim to support.

5.2 Building on the structures and governance in place within NHSPI, networks and programmes are already impacting on patient care, working alongside quality statements and patient pathways to deliver safe, equitable, effective, timely and patient centred care. The scoping report will enable these workplans across the organisation to consider the rare disease population across these pieces of work, future proofing mechanisms of service delivery through the lens of digital and technology and new models of care. Innovation at a local, regional and national level building capacity and excellence to patient care. By working alongside these strategic clinical networks, the rare disease voice will be amplified in existing and future work to deliver on prudent and collaborative approaches.

5.3 Learning from and with people with lived experience, the future holds the potential of observing how the foundation of what we have achieved in Wales impacts the future wellbeing of our rare disease population. Building from collaboration across the United Kingdom and globally, to improve the timeliness of a diagnosis, awareness, coordination of care and access to specialist care and treatments.

6. Next Steps

Review the NHSPI Rare disease scoping report (2025–2026)

- Collaborate with the Welsh Government and NHSPI Directors Group to review the current action plan, including its progress and align the next year's workplan with the current NHSPI governance structures (noting the organisation is undergoing changes to its structure and governance).
- Consider the requirement of a refresh of the current Wales Rare Disease Action Plan, due to the restrictions of a one-year extension to the UK Rare Diseases Framework.

Strengthen Data, Insight and Surveillance

- Finalise and launch the *rare disease data dashboard* in partnership with DHCW, CARIS, and the Value Transformation Directorate within NHSPI, which is chairing this project.
- Expand rare disease registers and scope the integration of *Patient Reported Outcome Measures (PROMs)* to inform service planning and evaluation.

Enhance Research and Innovation Capacity

- Build on the success of the *Wales Rare Disease Research Network* to establish a sustained research and innovation ecosystem across Wales.
- Support the James Lind Alliance Priority Setting Partnership and further develop the Wales Rare Disease Research Hub hosted by the Child Research Centre in CVUHB.

Improve Coordination and Transition of Care

- Support CVUHB to evaluate the *Wales Digital Rare Care Centre* and digital patient passport, for further dissemination across Wales if successful.
- Collaborate with the Child Health Network to ensure that the Welsh Government's Transition and Handover Guidance is fully implemented across health boards, with named workers for paediatric rare disease patients moving into adult services.

Expand Awareness, Education and Workforce Development

- Use findings from the RISE study to shape a national training framework for healthcare professionals and support integration of rare disease education into Nursing and Allied Health Professionals undergraduate curricula.
- Develop career pathways and capability mapping for rare disease specialist nurses in collaboration with HEIW.

Secure Sustainable Governance and Strategic Alignment

- Identify opportunities for collaboration and influence the inclusion of rare diseases across NHSPI and within Welsh health boards and trusts, including inclusion within the Integrated Medium-Term Planning (IMTP) process.
- Continue cross-UK and international collaboration through new forums to ensure alignment and shared learning.

7. Conclusion

The Wales Rare Disease Implementation Network (RDIN) has made substantial progress in advancing the objectives of the Wales Rare Disease Action Plan during its third and final year of implementation. This progress reflects a maturing system of collaboration across NHS Wales, academia, government, and the third sector—placing Wales at the forefront of UK efforts to deliver equitable, data-driven, and person-centred rare disease care.

Significant achievements include the establishment of the UK's first commissioned Syndrome Without A Name (SWAN) Clinic, the development of a national rare disease data dashboard and adult rare disease register, and the creation of the Wales Rare Disease Research Network. These initiatives have strengthened the evidence base, enhanced diagnostic capacity, and fostered a culture of innovation and patient engagement in rare disease policy and research.

Progress has also been made in embedding genomic testing pathways, advancing patient-reported outcome measures and digital care innovations, and integrating rare diseases into broader workforce and education strategies. However, persistent challenges remain in data quality, equitable access to specialist care, mental health support, and ensuring that service improvements translate into tangible benefits for individuals and families affected by rare diseases.

As the current UK Rare Diseases Framework has been extended to 2027, Wales is well-positioned to build on these foundations. The RDIN's evolving governance within NHSPI, and its alignment with strategic clinical networks and national programmes, offer a sustainable mechanism to embed rare disease considerations across health system planning. The next phase must focus on consolidating data and digital infrastructure, strengthening cross-sector collaboration, and ensuring that lived experience continues to drive priorities.

Ultimately, the progress achieved demonstrates Wales' commitment to delivering prudent, high-quality, and compassionate care for all people affected by rare diseases—transforming national ambition into measurable improvement and setting a strong platform for the next generation of rare disease strategy.