Wales Rare Disease

Action Plan – Annual Progress Report

2023-2024

Published 23 January 2025

on behalf of the Wales Rare Diseases Implementation Network

Contents

This progress report details the progress made by the Wales Rare Diseases Implementation Network (RDIN) in the second 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 the next 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

1.4 Underpinning themes within the Framework are:

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

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, while working together through the UK Rare Diseases Framework Board, the national teams

[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

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 two updated plans in 2017[5], and more recently in 2022, the group worked with key stakeholders within NHS Wales and its strategic partners, third sector, 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[6] as a Rare Disease Implementation Network (RDIN) in February 2024. RDIN is hosted by the Child Health Strategic Clinical Network which is led clinically by Dr Claire Thomas and is managed by Alex Hicks, the Child Health Network Manager, and supported by Ms Rhiannon Edwards as RDIN Support Manager. The RDIN Clinical Reference Group is chaired by the Medical Director of the Joint Commissioning Committee (JCC), Professor Iolo Doull, and is supported by a clinical lead for RDIN who sits on both the RDIN and Child Health Leadership Groups, Dr Jamie Duckers (Consultant in Respiratory Medicine). Going forward the clinical lead for RDIN will report to the clinical lead for Child Health, reflecting the relationship in the NHS Wales Executive between Strategic Clinical Networks and Implementation Networks.

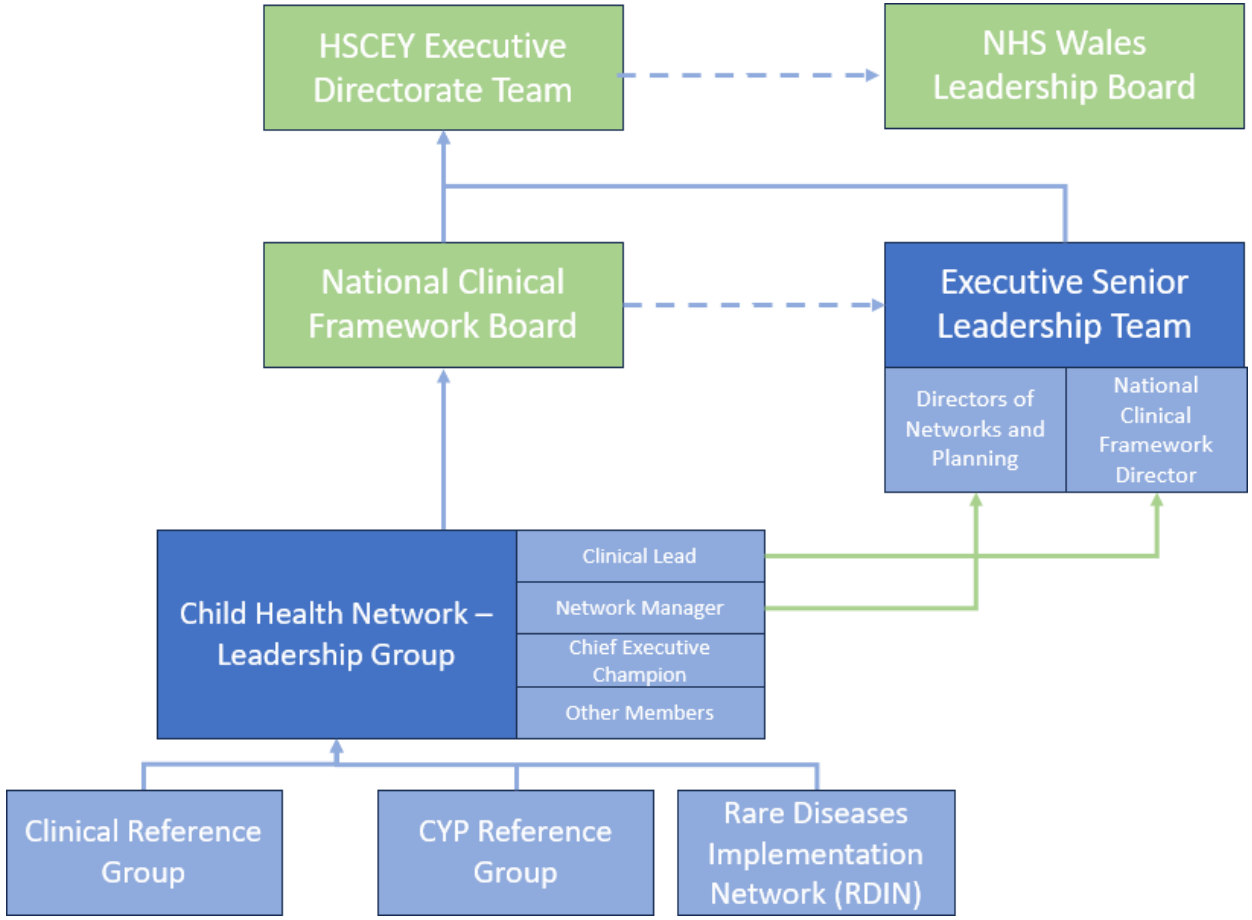
1.8 RDIN meets quarterly, to discuss information regarding national and international priorities, the progress against the implementation of the Wales Rare Diseases Action Plan and to act as an advocate for those affected by rare diseases.

[4] Welsh Implementation Plan for Rare Diseases (europlanproject.eu)
[5] welsh-rare-diseases-implementation-plan-july-2017_0.pdf (gov.wales)
[6] https://executive.nhs.wales/networks/implementation-groups/rare-diseases/

1.9 A leadership group for rare diseases was formed in November 2023, to support strategic prioritisation, and includes genomic, policy, clinical, and third sector support. Terms of reference are to be developed and agreed, although meetings have been held quarterly in between RDIN meetings. This Leadership Group will transition into a Clinical Reference group in 2024/25, reflecting the transition to an implement network.

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

Figure 1 – Strategic Clinical Network Structure, NHS Wales Executive



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 Four Nations UK Rare Diseases Framework.

2.2 Dedicated funding from the Welsh Government enabled an implementation group for rare diseases to be established, which in turn led to the appointment of a clinical lead for rare diseases (the first across the four nations) and a co-ordinator to support the implementation of the All-Wales Rare Disease Action Plan, including the twenty-nine actions, across the four priorities.

2.3 Developing the actions into workstreams enabled progress to be monitored, but also supported nationally agreed focus areas, including education, passports and clinical pilots to support coordination of care. One formal subgroup has been developed, chaired by the Congenital Anomaly Registration and Information Service (CARIS) focusing on Data and surveillance.

2.4 Key strategic alignment with the Genomics Delivery Plan for Wales[7] (launched in December 2022 reflecting the Welsh delivery of UK strategy for genomics and Genome UK: The future of healthcare[5]) focused elements of priority one actions, to support challenges of capacity within our workforce.

2.5 The robust engagement prior to the launch of the Wales Rare Disease Action Plan, enabled the plan to reflect the needs of the rare disease population in Wales and was clinically led.

2.6 In January 2024, the second iteration of the Wales Rare Disease Action plan was published. This removed three completed actions, streamlined the wording for the remainder of the actions and included 12 new actions to focus on recommendations from the patient empowerment groups.

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

3. Progress Summary

3.1 The development of the RDIN Leadership Group focused on producing a clear vision statement of the function of the network. Three priorities have been decided for the next 12 months:

1. Wales Digital Rare Care Centre
2. Data and surveillance
3. Research

3.2 Over the past year the Data and Surveillance Group has completed projects on Angelman Syndrome and Duchenne Muscular Dystrophy, with DiGeorge Syndrome ongoing. These projects were presented to stakeholder groups with the Angelman syndrome project being currently developed into an academic paper, shared with the UK Angelman Patient Group and potentially can be used to observe any overlap in the labs data compared to that of CARIS. The Duchenne's Muscular Dystrophy presentation highlighted gaps in Mental Health and respite care within Wales and the intention is to work with the Palliative and End of Life Board and Mental Health Strategic Programme to understand opportunities for collaboration.

3.3 The Rare Disease Research Network is nearing the end of the stakeholder mapping exercise, with a launch event planned in December 2024.

3.4 The RDIN pharmacogenomics sub-group was initiated to support new guidance from NICE on predictive genetic testing on risk for hearing loss in aminoglycoside prescribing. RDIN introduced Cardiff and Vale University Health Board neonatal teams to Manchester neonatal teams to build a consortium application for National institute for Health and Care Research (NIHR) Invention for Innovation (i4i) and Office for Life Sciences (OLS) Real World Evidence Call: NIHR208090: Pharmacogenetics to Avoid Loss of Hearing (PALOH) trial UK [PALOH-UK]. Following the success of this application, this subgroup has been stood down, until such time when the evaluation of this study can be implemented.

3.5 Partnering with RDIN for an application to Bevan Commission Exemplar Programme (BCEP) last year (2023-2024), Cwm Taf Morgannwg University Health Board developed a Paediatric Complex Rare Disease Multi-Disciplinary Team (MDT) clinic. The evaluation of this project identified learning on the resources needed for such a clinic, which is informing

further opportunities. This clinic also provided the opportunity for a follow-up and successful application to the Bevan Commission Exemplar Programme (2024/25), observing the needs of those with cognitive and learning disabilities when accessing digital support mechanisms, via transition support in the digital Rare Care Centre (see 3.7).

3.6 RDIN has continued to support the development of the evaluation and business case for the all-Wales Syndrome Without A Name (SWAN) clinic. Interim results have identified the positive impact of co-ordination of care, in terms of quality of life, patient report outcome measures (PROMs) and patient report experience measures (PREMS). Digital Health Care Wales (DHCW) have also supported understanding of health care utilisation developing a data dashboard to understand the impact of healthcare utilisation on this patient group, this was following input from the SWAN clinical team and support of some baseline economic health modelling.

3.7 RDIN submitted a successful application to the Bevan Commission Exemplar programme (https://bevancommission.org/programmes/bevan-exemplars/) to use the proposed Digital Rare Care Centre as the focal point to improve transition care for those with rare diseases. This south-east regional project will be a one point of access platform, that will be used by clinical teams to support their patients with pooled and existing generic and rare disease transition resources. This will share learning and resources between teams who have different levels of support for this period of health care provision. Information governance processes are underway across the region following conditional approval in Cardiff and Vale University Health Board.

3.7 The previously published all-Wales guidance document 'Guideline for the Investigation of Moderate / Severe Early Developmental Impairment and Intellectual Disability' which was published in January 2023[8], is due for refresh, and RDIN will be supporting the engagement process to facilitate this process to provide guidelines for paediatricians (acute and community based), who assess children and young people. This will be used to support referral for an assessment of early developmental impairment and/or intellectual disability, with or without consideration of an underlying Rare Disease. This will be implemented in conjunction with the National Strategic Clinical Network for Child Health.

3.8 Linked to the action plan, RDIN has been successful at developing key strategic partnerships in the past year, to develop services for people living with rare diseases in Wales. Once example of this is the work in partnership with the Wales National Pharmacogenomics Group (NPPG), which has focused the opportunities on developing a work-stream

[8] https://gweithrediaeth.gig.cymru/swyddogaethau/rhwydweithiau-a-chynllunio/clefydau-prin/canllawiau-clinigol/

that impacts on point of care genomic testing to enhance patient prescribing safety, focusing initially on Wales inclusion in the consortium application: NIHR Invention for Innovation (i4i) & Office for Life Sciences (OLS) Real World Evidence Call: NIHR208090: Pharmacogenetics to Avoid Loss of Hearing (PALOH) trial UK [PALOH-UK]. This piece of work aligns with the work of the Data and Surveillance sub-group to develop registers and the future impact of facilitating safe prescribing of Advanced Medicinal Treatments and Products (ATMP).

3.9 Through the leadership of RDIN, research prioritisation was actioned as a result of the publication of the UK Rare Diseases Research Landscape Project Report[9] which was published by the Medical Research Council (MRC) and National Institute for Health and Research (NIHR). The Life Sciences Hub Wales developed a Wales focused report on behalf of RDIN, Intelligence Report: Rare diseases research landscape in Wales[10] to enable background information to be understood prior to the development of a Rare Disease Research Network. This work was supported by partners in Welsh Government, and the Wales Innovation Network.

4. Progress by Priority

Analysis of progress by priority illustrates some areas where progress hasn't been made in the past year, despite the overall positive progress described in the summary above. This is due to a couple of primary reasons. Firstly, the past year saw the introduction of a new leadership group which required supporting from within the current establishment. This included facilitating meetings and development sessions to establish a vision for the new group. This has inevitably reduced RDIN's ability to keep abreast of the progress of certain actions in the plan. Secondly, 12 new actions were added (a net gain of nine as three actions were completed last year) to the action plan, without a clear resource commitment to deliver them. As a result, RDIN will review the current list of actions to ensure deliverability in 2024/25 within the resource envelope.

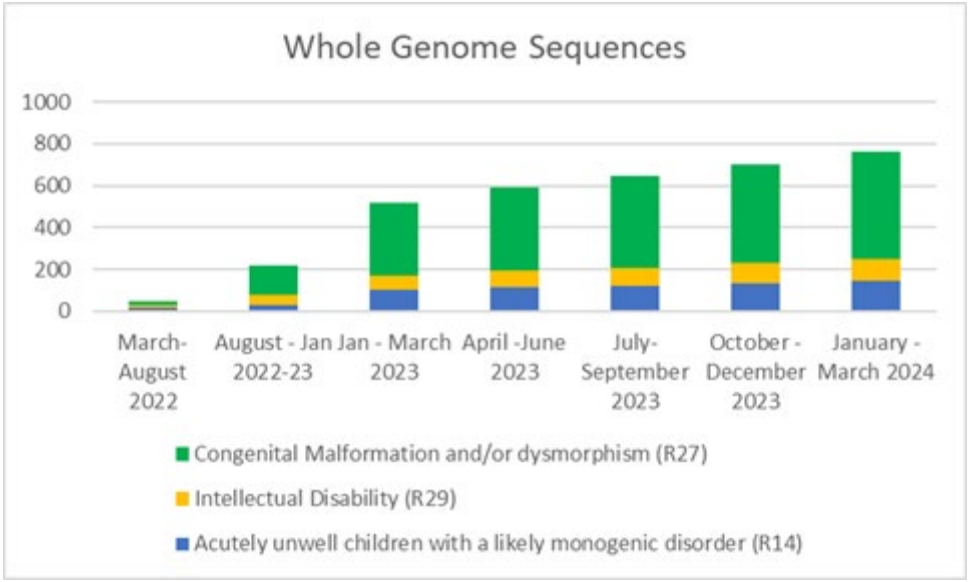
[9] https://openresearch.nihr.ac.uk/documents/3-45

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

4.1 Helping patients get a final diagnosis faster.

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

As can be seen in graph alongside, there has been a reduced trajectory of levels of testing for whole genome sequencing for Rare Disease in Wales, risking impacting on the overall target of 3000 tests being completed by December 2025. This is due to All Wales Medicine Genomic Service (AWMGS) being faced by challenges and barriers including increased vacancy rate for Clinical Scientists (analysis and reporting capacity), implementation of new Laboratory Information Management System and estates move – all adding to pressure against this specific delivery. AWMGS are satisfied with our WGS test activity figures to date considering all the other pressures they are facing.



4.1.2 Ensure a consent strategy is developed that enables researchers to securely and safely access routine genomic data generated by AWMGS for translational research purposes. This work is being taken forward by Genomic Partnership Wales, as part of the genomic strategy.

4.1.3 Engagement with Health and Care Research Wales (HCRW) to ensure access to research studies for rare diseases patients. RDIN links to HCRW via Welsh Government Research lead. Current activity from Welsh Government is to support the Rare Disease Research Network development, which will enhance understanding of the current access to research studies for individuals impacted by Rare Diseases.

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. The development of transcriptomic sequencing for Rare Disease in AWMGS is currently paused due to service pressures.

4.1.5 Co-produce research questions with service users, to bring Rare Disease research closer into policy and practice. Research was prioritised by the RDIN Leadership Group, therefore RDIN have engaged with the Welsh Government Research Manager, Wales Innovation Network and Life Sciences Wales Hub to establish a rare disease network to discuss the recommendations of the Rare Disease Research Landscape Project (Wales' version). This network is in development, with stakeholder lists completed and a launch event planned for December 2024.

4.1.6 Increase research activity in Paediatric Rare Disease research.

This action has not progressed. The development of the Rare Disease Research Network will streamline the governance processes for this action.

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. RDIN are supporting conversations with PHW Newborn screening (NBS) service, to understand the opportunities to pilot the use of remaining blood spots samples for predictive testing to impact on Pharmacogenomic pathways.

4.1.8 Explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices. Please see the update below in 4.1.9.

4.1.9 Non-Invasive Pre-natal Testing (NIPT) will be expanded to other reproductive pathways to improve patient outcomes and optimise resource utilisation. AWMGS was involved in testing within the cfDNA testing in Early Pregnancy Loss study. This study is now complete, and the publication is in draft. Following the evaluation of the results, ongoing expansion will be considered.

4.1.10 Equal access to genomic testing across the UK. Provision within Wales or referral outside of Wales. This action has been risk logged by AWMGS (Risk Assessment - 23-140-GEN) - It is becoming increasingly difficult to find NHS genomic

providers with sufficient capacity to take on external income generating activity within the UK. In addition, for those who will provide genomic testing there are commonly significant reporting delays across multiple services providing this service. Currently due to the Genomic Laboratory Information Management System (LIMS) project, estates move and ongoing vacancies, there is insufficient capacity within the AWMGS laboratory to undertake repatriation of any of these services within 2024. This is recognising the sudden unavailability or poor reporting performance of external suppliers and AWMGS ability to respond to these unplanned challenges at short notice. Therefore, there is a risk that the quality and accessibility of results for patients will be impacted due to lack of suitable alternative testing options.

4.1.11 All nations should develop actions which support diagnosis and care for non-genomic conditions. In Wales, the SWAN clinic supports diagnosis and care for those with non-genetic rare disease conditions, as well as genetic rare diseases. RDIN has continued to support the evaluation and business case for this clinic, which is understood as an example of good practice within the UK. Professor Griffiths, Swansea University, was successful in obtaining funding from MRC and NIHR to research metabolomics as a mechanism of improved diagnosis of rare diseases and the RDIN quarterly meeting as a mechanism of communicating about both examples to the wider Rare Disease stakeholders about progress.

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.

This action has not progressed in the past year.

4.2 Increasing awareness of rare diseases amongst healthcare professionals

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 has published in Respiratory futures Journal ‘Person centred

Care - the grail of the NHS?[11] and has represented NHS Wales at the European workshop on 'Enabling the integration of a federated meta data infrastructure and novel digital diagnostic tools for rare diseases in healthcare institutions'.

Education and Shared Learning

4.2.2 Survey qualified Health Care Professionals (HCP) undergraduates on their understanding and learning needs in rare disease. The Rare Disease Evaluation Study (RISE) survey has been translated into use into a qualified HCP survey, but this action was deprioritised by the RDIN Leadership Group, so no longer forms part of the RDIN activity in 2024. RDIN understand the previous RISE survey deployed to undergraduate medical students, a collaboration between Medics 4 Rare Diseases and Cardiff University is due for submission to the Orphanet Journal of Rare Diseases.

4.2.3 Use results to develop training and development plan from baseline information on HCP understanding of rare diseases. The RISE survey was not deployed into the undergraduate HCP population in Wales due to the issues highlighted in 4.2.2. As a consequence there are no results to develop into a training and development plan.

4.2.4 Incorporate rare diseases module in the undergraduate curriculum for medical students. Genomics Partnership Wales Workforce and Training Implementation Group modules for undergraduate medical students are in their third year now and continue to be extended from Cardiff University to Swansea University medical schools.

4.2.5 Ensure development of specialist consultant roles - interest and confidence. No progress to report in 2023-24.

4.2.6 Clinical Nurse Specialists – build understanding of paediatric and adult CNS workforce. No progress to report in 2023-24.

4.2.7 Continue to develop active partnerships with patients and patient advocacy groups (PAGs). Patient and public involvement and engagement (PPIE) involvement continues to be provided by Genomics Partnership Wales (GPW) sounding board. This enables activity to be built around the needs of service users and brings in lived experience into strategic conversations, such as the development of the refresh of the action plan, our initial RDIG strategic direction, and implementation of the action plan. More recently members of the sounding board have agreed to support a

[11] https://www.respiratoryfutures.org.uk/features/person-centred-care-the-grail-of-the-nhs/

workshop to develop a proposal for an in-person Rare Care Centre, being led by GPW and AWMGS. There has been agreement in principle from the RDIN Leadership Group to include patient representation in the meetings. This decision will need to be reviewed based on the transition to a clinical reference group.

4.2.8 Recognise and celebrate Rare Disease Day in secondary and primary care. Rare Disease Day was celebrated in Wales by announcing that a chain of rare disease gardens would be installed across Wales. The first garden was planted in the new Genomics Health centre in Wales in January 2024 and was covered in the media by TV and online and physical newspaper articles. The second garden has been planned and approved by PPIE engagement in North Wales within a hospice garden. There is interest in Western Australia to roll this initiative beyond Wales to create a global opportunity. RDIN took part in UK Genetic Alliance Rare Disease Day activities in February 2024 online and in-person at the Norwegian Church, Cardiff Bay.

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. A mapping exercise of activity was carried out via the education workstream to include activity from the Wales Gene Park. No current educational activities are taking place within primary care, but there are plans for a medical student awareness event in Swansea University by the Medics 4 Rare Disease ambassadors in the future. RDIN's collaboration with Health Community pathways is scoping opportunities to develop pathways of care from primary care in Wales into support mechanisms across the health and social care and third sector environments. The intention is to start with Cystic Fibrosis and consider a generic Rare Disease, red cell and Duchenne Muscular Dystrophy service pathway's for the future.

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 forthcoming Genomics Workforce Plan being led by HEIW. The mainstreaming and genomics educational resources have also been included within the draft Genomics Workforce Plan due to be published in autumn 2024.

Improving Awareness of Rare Diseases with Data

4.2.11 CARIS team expansion to include adults affected by rare conditions. Rare Disease data was a priority decision made in RDIN Leadership Group. Adult registries have been expanded in the past year in CARIS, with De George Syndrome being validated by staff within the PHW CARIS team. Decisions on prioritisation were based on clinical insight from RDIN from within the RDIN/CARIS sub-group. Further work is needed in the next year, to understand how this 'diagnosis register' can be expanded to include PROMs and PREMS and clinical data which includes support from the Heath Intelligence team in the Data, Digital, Innovation and Value Directorate within the NHS Wales Executive

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 UHBs/Trusts by WRDIG. This is currently being considered by the CARIS team, there are challenges with publishing rare disease data, due to low numbers. A pilot was supported to look at generic rare disease data from Cwm Taf Morgannwg University Health Board to support the Bevan Commission Exemplar Paediatric MDT Rare Disease clinic. Learning from this activity is being considered by CARIS.

4.2.13 Consider collection of rare diseases data at both a national all-Wales level drilled down to lower-level geographies (such as UHB/Trust footprint) where numbers of patients with specific diseases allow. See update from 4.2.12

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 having a named worker and digital care plan linked to a patient passport. This action was de-prioritised by RDIN leadership group for 2024. However, an application to the Bevan Commission Exemplar programme by Cardiff and Vale University Health Board, supported by RDIN will focus transition support via the proposed Digital Rare Care Centre. RDIN will link with the Strategic Clinical Network for Child Health to support work being undertaken by the Network in 2024-2026. This will improve transition and handover in line with the WG review of the implementation by NHS Wales of guidance published in 2022.

4.3.2 Establish Rare Diseases as a "Community of Practice" and develop example/exemplar clinical pathways for rare disease conditions, including MDT involvement. Please see updates in 4.2.9.

SWAN Clinic

4.3.3 Continue to build the establishment and assess/evaluate SWAN clinic. RDIN supports the SWAN clinical effectiveness working group which is leading the development of a commissioning specification and proposal to the JCC. This enables shared learning from this business case development into other RDIN work-streams. This has supported discussions with Value in Health over development of future rare disease patient outcome data sets and DHCW to develop Rare Disease data dashboards, to highlight pathways of care.

4.3.4 Understand the usefulness of PREM and PROM collation to develop enhanced service provision. RDIN has supported the SWAN Clinic working group over the past year leading to the inclusion of PROMs and PREMs in the data dashboard, as reported in 4.3.3 above.

Digital Patient Record

4.3.5 Establish an easily used "app" to enable a "patient passport" for rare disease patients RDIN facilitated a rare disease global stakeholder meeting to understand the opportunities for a global Rare Disease passport. An existing rare disease patient passport developed by CamRare (https://www.camraredisease.org/) was discussed at the stakeholder meeting, enabled this to be piloted in Western Australia. RDIN continues to build collaborative relationships with the NHS Wales app development team, to influence their 'about me' section in the app with the aspiration to include questions which are more aligned to a strategic approach and supports the validation of the tool. There is an interest within NHS Wales app team to signpost to the rare care centre when it is launched. RDIN were invited to the Digital Services for Patients and the Public (DSPP) Prioritisation workshop to discuss these opportunities in September 2024.

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. The UK Rare Disease Forum workshop on children's and young people (CYP) mental health and wellbeing, initiated conversations between RDIN and the Strategic Clinical Network for Mental Health, which is part of the wider national programme for mental health in Wales. These discussions identified the need to map the current mental health pathway support for children and young people diagnosed with rare diseases.

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 Network Support Manager attended the recent Equity, Diversity and Inclusion in Research Association (EDIRA) conference and developed contacts with Sickle Cell Association and Diverse Cymru. The discussions have continued in relation to building trust in under-served communities and collaboration by developing the EDIRA partnership. RDIN, as part of this partnership, has provided input into a skills and engagement matrix to support ongoing opportunities for shared learning and engagement.

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. This is not under the current workstreams within RDIN. The RDIN quarterly reports on the new processes within the All-Wales Therapeutics and Toxicology Centre (AWTTC). ATMP are increasingly being considered as treatment options for those with a rare disease. However, identification of the individuals with rare diseases in Wales has an impact on the access to these drugs. RDIN was represented on the Wales JCC in relation to ATMP outcomes, and shared learning on the patient focused outcome measures that could impact on commissioning treatments.

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. AWTTC is responsible for highlighting new medication approved by NICE to approve for use in Wales. This information is reported to RDIN quarterly to understand any challenges that can be influenced by

our stakeholder group. This process does not directly require input from RDIN but will maintain an overview due to the importance to people diagnosed with rare diseases in Wales.

4.4.3 Monitor uptake of new rare diseases medicines and prescribing. This action has not been progressed by RDIN. However, the development of the CARIS adult registers in the future could support the monitoring of specialist medications. This could be developed as a separate workstream, understanding the prescribing data from secondary care that is currently not available to the CARIS team.

4.4.4 Continue to develop improvements in the monitoring of use of medicines for patients with rare diseases including Blueteq. This action will enhance the above action but has not been prioritised in the past year. However, next year RDIN will endeavour to identify a lead in NHS Wales to map the Blueteq technology with patient focused information from CARIS.

4.4.5 RDIN will build actions which support the use of repurposed and off-label medicines and devices. RDIN has been introduced to a North Wales company called Ambrose, that works within the repurposed and off-label medicines sector. Currently RDIN isn't involved in any NHS focused priorities in this strategic space.

4.4.6 RDIN will observe opportunities to understand how pharmacogenomics can improve the effective management of those with rare diseases in Wales. As reported in the summary of achievements, the RDIN pharmacogenomics subgroup was initiated to support new guidance from NICE on predictive genetic testing on risk for hearing loss in aminoglycoside prescribing. This was also supported by the NPGG. This group will reestablish when the evaluation of this project identifies the opportunities for all Wales pathways to be developed for equal access of this technology into neonatal settings.

Access to specialist Care

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

5. Opportunities and Challenges

5.1 The Rare Diseases Implementation Network has focused on developing its Clinical Reference Group as a mechanism to partner and collaborate with colleagues, whilst providing leadership direction for the implementation of the Rare Diseases Action Plan. Developing the purpose statement collaboratively enabled robust engagement with partners and enabled priorities to be set around the strategic direction of the NHS Wales Executive and specifically RDIN. The strong desire for improving the care of those in Wales impacted by rare diseases has led to invitations in leadership and stakeholder events where RDIN was able to discuss the impact of current service provision upon the health and care needs of the rare disease communities. This also progresses the awareness of rare diseases within wider strategic conversations. The newly formed Digital, Data, Technology, Innovation and Value (DDTIV) Directorate in the NHS Wales Executive, which incorporates the Health Intelligence team, as well as the communications function in the Networks and Planning Directorate are supporting all these priorities and have enabled actions to be progressed.

5.2 Rare disease pathways of care from primary, secondary and community care are challenging due to the extensive number of rare diseases, their complexity and people with lived experience often interacting with a significant number of health and social care services, whilst relying on generalists who might only understand part of the problem, with limited or no specialist knowledge about their rare disease condition. Therefore, RDIN is working towards developing a cohesive approach at a regional, national or even international level in some cases. This is reflected by the lack of actions in the plan relating to individual health boards. It is imperative therefore that RDIN's approach flourishes through the leadership of the NHS Wales Executive to maximise opportunities for innovation and collaboration to improve the quality of services and health outcomes for people living with rare diseases. The working arrangements for the Strategic Clinical Networks and their delivery structures, including Clinical Reference Groups (described as the engine room of the Strategic Clinical Networks) and Implementation Networks like RDIN, focus attention on the health boards being accountable for the active service improvement and adherence to the quality and safety framework for clinical pathways. With an estimated 7,000+ rare disease conditions, an estimated 200,000+ individuals from a population of 3.2m in Wales will be impacted.

Next Steps

RDIN has been incorporated within the Child Health Strategic Clinical Network to strengthen the relationship between

the strategic clinical network and the implementation network for rare diseases and to maximise the opportunities for collaboration with other networks and national programmes. The RDIN Clinical Lead has been co-opted as a member of the Senior Leadership Group of the National Strategic Network for Child Health for this purpose. The RDIN Leadership Group also transitioned to become a Clinical Reference Group in October 2024. The priorities of the proposed Rare Care Centre, data and surveillance and research will be the focus for RDIN in the year ahead, with the support of other strategic clinical networks and programmes within the NHS Wales Executive. Cross cutting themes across the NHS Wales Executive will pool resources and enable a streamlined and strategic approach to impacting on sustainable change; examples of these will be technology, transition and data/insight gathering. Planning and redrawing relationships within newly formed networks will enhance collaboration and awareness of the rare disease priorities, for inclusion in relevant annual plans and delivery.

6. Conclusion

During 2023/24 there has been a period of transition, as RDIN establishes itself as an implementation network within the wider framework for strategic clinical networks in NHS Wales Executive, which has also the establishment of new networks for child health, women's health, musculoskeletal, gastroenterology and others and a new Clinical Framework Implementation Board in September 2024, representing further opportunities for RDIN to explore in the year ahead.

Progress has been made in terms of building the foundations for RDIN and the beginning of work-streams which will produce defined outputs that will improve health outcomes for people living with rare diseases in Wales. Working alongside strategic partners in Wales, the UK and globally to share learning and implement crosscutting opportunities for innovation and service model redesign in NHS services for people living with rare diseases.