Wales Rare Disease

Action Plan Progress Report

2022-2023

Published January 2024

on behalf of the Wales Rare Disease Implementation Group

Contents

This report details the progress made by the Wales Rare Diseases Implementation Group (WRDIG) in the first year of the Rare Diseases Action Plan for Wales published in June 2022.

1. BACKGROUND / STRATEGIC CONTEXT

1.1 On 9 January 2021, the UK Rare Diseases Framework[1] 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[2] 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 & drugs

1.4 Underpinning themes within the Framework are:

- Patient Voice
- Collaboration

1. https://www.gov.uk/government/publications/uk-rare-diseases-framework
2. https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework#annex-a

- Research
- Data & 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 ensure as much alignment across the four nations as possible. The implementation group for Wales is the Wales Rare Diseases Implementation Group (WRDIG).

Wales Rare Diseases Implementation Group (WRDIG)

1.6 WRDIG was established in 2014 when the Welsh Government first instituted the Welsh Implementation Plan for Rare Diseases (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, and more recently in 2022, WRDIG, has worked with key stakeholders, within NHS Wales and its supporting organisations, 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 sits within NHS Wales Executive[3] and is chaired by the Medical Director of the Welsh Health Specialised Services Committee (WHSSC), Professor Iolo Doull and is supported by a clinical lead, Dr Jamie Duckers (Consultant in Respiratory Medicine) and group coordinator, Ms Rhiannon Edwards. They meet 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.

3. https://executive.nhs.wales/networks/implementation-groups/rare-diseases/

1.8 WRDIG is responsible for overseeing the action plan, delivering national pieces of work, and supporting health boards and trusts to develop, deliver and report on their local medium-term plans.

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 Through dedicated funding from the Welsh Government to establish WRDIG, a clinical lead (the first in the four nations) and coordinator were appointed 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 informal workstreams enabled progress to be monitored, but also supported nationally agreed focus areas, including education, data linkage and clinical pilots to support coordination of care.

2.4 Key strategic alignment with the Genomics Delivery Plan for Wales[4] (launched in December 2022 reflecting the Welsh delivery of UK strategy for genomics, Genome UK: The future of healthcare*ADD REF) 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 supported a clinically led leadership.

4. https://www.gov.wales/sites/default/files/publications/2022-11/genomics-delivery-plan-for-wales_0.pdf

3. Progress Summary

3.1 The decision to deliver this plan over five years, with a refresh each year, has enabled much of the foundations of the action plan to be developed. Although, not defining workstreams based on the priorities, collaboration has been achieved by addressing the needs of our clinical colleagues and the patient population of Wales.

3.2 The opportunity for a clinically lead WRDIG, through the understanding and engagement with clinicians across Wales has enabled enthusiasm to build regarding the opportunities that can be developed through a bottom-up approach, that aligns with policy to impact on the delivery of service provision in Wales. This has been highlighted across the Four Nations and internationally to enhance our ability to collaborate.

3.3 Key sub-groups (Data, Paediatric RD MDT clinic) have been established during the year, with the coordination of the groups determined by the expertise, and membership built by interested parties. Like other nations, funding and resources have been limited, so the passion and dedication of our members has been the driving force in our progress and impact over 2022-23.

3.4 Despite the challenges of engaging all Health Boards in Wales, the WRDIG reporting template was used as a mechanism in three HB regions to highlight to their Chief Executives the need to provide representation in WRDIG to drive change. This has resulted in a localised launch of the plan; a pilot paediatric Rare Disease clinic and primary care education sessions being delivered. In addition, and with the support of Genetic Alliance Cymru, Rare Disease Day was celebrated across Wales.

3.5 The Syndrome Without A Name (SWAN) clinic, funded by the Welsh Government and highlighted in priority 3 of all-Wales Rare Disease action plan continues to be an exemplar for national and international colleagues, with the funding extended now to March 2024. The 2-year pilot seeks to evaluate the clinical effectiveness of this project, aiming to shorten the diagnostic odyssey for patients with rare diseases. In the first 16 months it has successfully assessed 105 referrals from across Wales.

3.6 Coordination of care for those with Rare Diseases has also been highlighted as an important focus across Wales, with three coordinators (two Clinical Nurse Specialists within the SWAN Clinic and one further Paediatric Matron in the Paediatric pilot) supporting patients and carers through their experience of living with Rare Diseases.

3.7 Collaboration between the Congenital Anomaly Register and Information Service (CARIS) team hosted by Public Health Wales and WRDIG, is drawing on the expertise of clinicians and data analysts to understand how rare disease registers can be improved and drive the improvement of pathways of care. In addition, this is also improving relationships between our clinical specialists and WRDIG, whilst developing Rare Disease champions across Wales.

3.8 Supporting our patient population in Wales is challenging, due to our geography and accessibility to service provision. Therefore, the development of a patient passport, a Rare Disease virtual health hub and the delivery of patient leadership/peer support, have been seen as an essential aspect of delivering on our Action Plan. We are pleased to report that the planning and delivery is underway for all three of these elements.

3.9 Developing and delivering education and training sessions for our clinical colleagues has been a priority for WRDIG. Collaboratively, Medics4Rare Diseases, with support from Cardiff University and WRDIG, have launched an undergraduate medical student survey on Rare Diseases (Rare DISease Evaluation (RISE) UK 2022). This has resulted in the participation of over 180 medical students across over 30 medical schools. WRDIG are additionally supporting Medics4RD to develop a similar version for Nurses and Allied Health Professionals (AHP). This will be circulated and supported by Health Education and Improvement Wales (HEIW) and will assist in understanding the needs of our clinicians of the future.

3.10 The All-Wales guidance document 'Guideline for the Investigation of Moderate / Severe Early Developmental Impairment & Intellectual Disability' was published in January 2023[5]. This provides guidelines for paediatricians (acute and community based), who assess children and young people referred for an assessment of their early developmental impairment and/or intellectual disability, with or without consideration of an underlying Rare Disease.

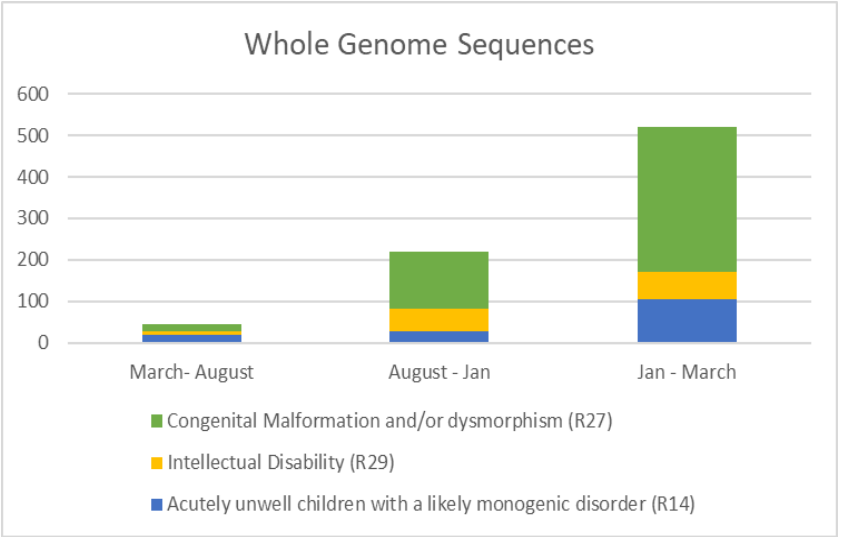
5. https://executive.nhs.wales/networks/implementation-groups/rare-diseases/clinical-guidelines/

4. Progress by Priority

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 by diagram 1, there has been a significant rise in WGS in year ending March 2023.



agram 1

4.1.2 **Return (repatriate) Fetal Whole Exome Sequencing trios testing (FAGP service) to Wales**. All whole exome sequencing now being undertaken in Cardiff.

4.1.3 **Ensure validation of a whole transcriptome service which will enable better understanding of RNA sequences to determine if a DNA sequence is turned on and whether proteins have changed**. Wales continues to be involved in a UK Wide Research study.

Research Eco-system

4.1.4 **Ensure a consent strategy is developed that enables researchers to securely and safely access routine genomic data generated by AWMGS for translational research purposes.** RDIG believe all patients receiving genomic sequencing of any kind in Wales should be offered a consent for engaging in research. Genomics Partnership Wales (GPW) and Wales Gene Park (WGP) have undertaken extensive public and patient engagement on this topic. However, resources to

develop and implement this strategy is challenging, therefore this is an ongoing action within the Wales Rare Disease Action plan.

4.1.5 **Engagement with Health and Care Research Wales to ensure access to research studies for rare diseases patients**. This is a 5-year strategy; however, we are starting to see an increase in participants entering studies with a 1.15% increase in studies (99) supporting Rare Diseases and 1.10% increase in participants (1172). In addition, the UK Cystic Fibrosis (CF) international conference was hosted by Cardiff (March 2023). A Rare Disease Clinical Fellow has increased the capacity for research and the All-Wales CF Service has been selected for early phase Trials centre.

Prevention and Early detection

4.1.6 **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**. AWMGS partnered with Ilumina in Cambridge[6], has recently published the results of research study concluding that genomic variants can be detected via Whole Genomic Sequencing (WGS), from standard dried blood spot (DBS) new-borns screen technique. Genomics England is continuing to launch their whole genome newborn screening pilot (England only research project). Therefore, in Wales we will monitor this evaluation over the lifetime of the pilot and observe opportunities to develop services based on the evidence.

4.1.7 **Explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices**. Co-working with the early pregnancy unit in UCL research, testing products of conception to run Non-Invasive Prenatal testing (NIPT) studies (in recurrent miscarriage). This is being reported on shortly, and work will be continued on translation of this research into clinical practice.

4.1.8 **NIPT will be expanded to other reproductive pathways to improve patient outcomes and optimise resource utilisation**. The Translation of the research discussed in 4.1.7 will be continued following the reporting of the study outcomes.

4.1.9 **Implement a next generation sequencing service to detect genomic alterations when fetal structural abnormalities have been identified on ultrasound scan**. This has been completed.

6. https://www.mdpi.com/2409-515X/9/3/52

Service/Digital/Technical infrastructure

4.1.10 Ensure horizon scanning for commissioning requirements to inform the current National Genomic Test Directory for rare and inherited disease. This action will be modified to consider the increase of awareness of new tests rather than increase in accessibility. This is due to commissioning being dependent on the UK genomic test directory.

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. Specific tasks completed: Leadership activities and horizon scanning for engagement opportunities. Focus groups, Undergraduate lectures, lectures to genetic MDT, primary care events with M4RD, Undergraduate specialised study module in rare disease.

Education and Shared Learning

4.2.2 Survey qualified HCPs, undergraduates on their understanding and learning needs in rare disease. The RISE survey created by Medics4RD has been supported by WRDIG and Cardiff University and successfully implemented into undergraduate medical trainees (>30 med schools in UK, >180 respondents) This is being modified to address Nurse and AHP learning needs and will be distributed via HEIW.

4.2.3 Use results to develop training and development plan from baseline information on HCP understanding of rare diseases. Genomics Partnership Wales Workforce & Training Implementation Group are developing a big 'mainstreaming' initiative, which will involve reaching out to a lot of non-genetics clinicians to raise awareness and upskill individuals, this includes Genomics and Pharmaceutical clinical leads in HIEW. All Wales Medical Genomics Service (AWMGS) have

helped HIEW to recommission a range of master's level modules on genomics that will be freely available to all HCPs. The RISE survey has an education module linked to the survey, which will be reviewed, dependent on the needs of AHP's. Conversations are ongoing with HEIW to develop requirements to include Rare Diseases in the commissioning contracts for Nurses and AHP in higher education institutions.

4.2.4 Incorporate rare diseases module in the undergraduate curriculum for medical students. Genomics Partnership Wales Workforce & Training Implementation Group modules for undergraduate medical students in 3rd year now, extending from Cardiff University to Swansea University.

4.2.5 Continue to develop active partnerships with patients and patient advocacy groups (PAGs). Patient involvement is included in undergraduate training in Cardiff University. Wales Gene Park delivers training regularly to clinicians, with integrated patient stories. Patient representation is achieved in all WRDIG strategic discussions, by means of patient advocacy groups, patient representatives and the use of the third sector and the Genomic Partnership Wales sounding board.

4.2.6 Recognise and celebrate rare disease day in secondary and primary care. All Health Boards were asked to report on activities linked to Rare Diseases Day. These included: inclusion of Rare Disease in Health Board social media posts and newsletters, grand round presentations and to the support of hospital foyer stands by Genetic Alliance.

4.2.7 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 genomics workshop for Primary care was delivered in Hywel Dda UHB, included Rare Diseases. This was well attended. A partnership between HEIW, Medics4RD and WRDIG delivered a Rare Disease 101 online course for primary care, this is now hosted on the All-Wales HEIW education platform.

4.2.8 Ongoing programme of WGP education and engagement with HCP and students including Genomic Counselling role (across Welsh Health Boards and HEIs) including precision medicine. HEIW in conjunction with AWMGS have created e-learning modules on genomics that will be freely available online. Lectures given on precision medicines in Cystic Fibrosis – the CFTR variant dependent modulators).

Improving Awareness of Rare Diseases with Data

4.2.9 CARIS team expansion to include adults affected by rare conditions. The CARIS team started an adults rare diseases registry during the pandemic. Work has progressed with current priorities determined collaboratively with a sub-group of WRDIG. Current CARIS/WRDIG working group (including the funded rare diseases clinical fellow) are focusing on respiratory and Neurological Conditions. We have observed some of the challenges in acquiring and verifying adult rare diseases data, with current clinical information not matching the CARIS registry. We have been lucky to have secured a clinical fellow to work on this data and are learning lessons about coding that may support enhanced data collection. The CARIS team also joined a 4 Nations data research node bid. Although the bid was unsuccessful, the ability to harness collaborative support within Wales, including clinicians, data experts, academic and surveillance/coding expertise was noted. The CARIS team has also been scoping how a co-production approach can be applied to the register. This work was progressed by a trainee in Public Health. To date, opportunities to collaborate with patient organisations to pilot potential improvements, such as patients self-registering, have begun.

4.2.10 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. CARIS data is reported on yearly via the public health Wales web pages. Ongoing work in a data workstream is developing understanding on how regional data can be provided within specialty areas to understand the patient pathways that exist. Metrics are yet to be designed, due to the complexity of rare disease diagnosis, and there remains a gap in identification and registering of non-genetic rare diseases.

4.2.11 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. This has been scoped with the development of the first report to a health board being completed. Preliminary results and potential methods for drilling down the data have also been presented to the WRDIG. Local analysis has been undertaken to demonstrate the impact of providing data to each Local health Board to raise awareness of the numbers of patients within their region, to facilitate an understanding of patient need. Resource capacity within the CARIS team and a lack of clinical champions currently impair the ability to develop this on an all-Wales level.

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.** Extrapolating work from the transition of people with CF and using in other conditions. An online transition pack to care circle, based on the newly published Transition and handover guidance[7] published by the Welsh Government in February 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.** Working with the CARIS team, WRDIG are understanding the data within the registers. Focusing workstreams on condition specific data to engage with clinicians to understand patient pathways, with initial work within respiratory conditions and neurology. This will increase awareness of Rare Diseases and focus on collaboration across networks within the new NHS Wales Executive.

We are also adding wellbeing support to Care Circle[8], learning the lessons from the all-Wales Adult Cystic Fibrosis (CF) Centre, developing bespoke support rooms for Service users, carers, and significant others. Initial draft pathways have been developed for CF, and scoping work continuing mapping standards and specifications of health care touch points for those impacted by Rare Disease.

SWAN

4.3.3 **Continue to build the establishment and assess/evaluate SWAN clinic.** Progress with improving care-coordination has been made during the first 12 month of the pilot. The specialist adult and paediatric nurses assist patients and their significant others/carers before, during and following their appointments. Supporting the collection of PREMS/PROMS and contacting families after clinic appointment to address any outstanding queries or concerns.

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

Without this service there will be absence of:

- Support for children when they transition to primary school.
- Co-ordinated support within a school to help produce an individualised health plan.
- Support for a child with visual impairment
- Grants for equipment for patients such as wheelchairs and computing equipment.
- Individualised health plan for a patient with long standing health concerns.

De-brief appointments with the genetic counsellor are being planned. Regular meetings with WRDIG and SWAN UK CYMRU and other charities take place to establish needs and identify best mechanisms of support PROMS/ PREMS/ health outcome assessments.

4.3.4 **Develop suitable PREM, PROMs for use in evaluation in the SWAN clinic with potential use across all rare disease patients.** A 'Task and Finish' group was facilitated by WRDIG, to develop PROMS and PREMS for the SWAN clinic. This brought together patient groups to co-produce and co-design PREMS, and ensure PROMS were selected on the nearest validated measure of those impacted by a SWAN diagnosis. These have subsequently been used across other sectors to commence Value-Based care evaluation in Rare Diseases. These developments were supported by Centre for Healthcare Evaluation, Device assessment and Research (CEDAR), we are also scoping opportunities to approach ICHOM to identify a Rare Disease Validated PROM and PREM dataset. This has the potential to impact on validation of evaluation measures across the world.

Digital Patient Record

4.3.5 **Establish an easily used "app" to enable a "patient passport" for rare disease patients** A patient passport has been launched in Wales by Care and respond[9] (Scienap) and supported by WRDIG, linking patients with emergency services.

8. https://www.carecircle.org/

This webapp enables patients and carers to collate their own important information to be shared with healthcare professions when required (cost incurred by user or healthcare organisation). There are also opportunities within Care Circle[8] to enable similar services to be accessible linking data across healthcare (subject to current LifeArc grant application), this will be free to the user.

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.** Mental wellbeing work to be extrapolated from CF to digital hub for use in rare diseases. This will provide generic information from psychologists, youth workers and welfare officers to understand where support can be accessed.

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.** There are 2 process to get access to medicines prior to market authorisation - either clinical trials or the Early Access to Medicines Schemes (EAMS). EAMS is useful when evidence is not strong for the MHRA (relevant in many rare diseases). If there is positive Final Appraisal Document (FAD), England must provide the medicines and fund within 90 days. In Wales, it is 60 days.
If AWMSG endorse a medication through their 'One Wales' authorisation process it is available within 10 days. In Wales, medicines that either do not have a health technology appraisal or may not have marketing authorisation can be commissioned for a review by WHSSC.

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.** Since January 2022, WHSSC has commissioned 10 new medicines for rare disease through

the NICE Process, 11 through EAMS, three through the One Wales process and one medicine was brought through the WHSSC prioritization process.

4.4.3 **Monitor uptake of new rare diseases medicines and prescribing.** Blueteq has now fully deployed through Wales for all medicines commissioned by WHSSC, and therefore monitoring for the uptake of new rare disease medications is in place.

4.4.4 **Continue to develop improvements in the monitoring of use of medicines for patients with rare diseases including Blueteq.** There are concerns regarding the access of drugs through clinical trials or the Early Access to Medicines Schemes (EAMS) decreasing in Wales compared to England, because of funding of research and development infrastructure.

Access to specialist Care

4.4.5 **WRDIG to continue to work with WHSSC and HEIW to ensure appropriate consultant specialist services in Wales.** WRDIG has supported the SWAN clinic to understand the opportunities to develop their clinic service and support the Clinical Nurse Specialists to impact on the wider agenda of Rare Diseases. WRDIG is also a key stakeholder in a newly developed pilot paediatric Rare Disease MDT Clinic in South Wales. This will enable learning and evaluation of localised clinical support within Paediatric generic clinics.

5. Challenges.

5.1 The progress of implementing the all-Wales Rare Disease Action plan, has been impacted by the lack of capacity of the WRDIG clinical lead and coordinator. This has led to a reduced ability to drive forward opportunities in research and engagement activities, to collaborate fully with the genomics strategy, and the development of Rare Disease education frameworks and robust workstreams/leadership plans. However, opportunities in wider UK and European research studies have been recently supported by the recruitment of a Head of Life Sciences Research and Industry Partnerships, Welsh Government.

9. https://www.careandrespond.com/

5.2 The lack of dedicated funding for WRDIG, with the exception of funding the Clinical lead, Coordinator and project support has led to constraints in achieving levels of success on aspects of the plan, including robust clinical engagement across Wales. This has resulted in high levels of responsibility being driven by the core team rather than the team having the ability to delegate tasks to others to lead. The newly established Wales Rare Disease Implementation Network (WRDIN) may give more opportunities for resources to be more focused, to enable developments in these aspects.

5.3 The re-organisation of NHS Wales governance into the NHS Wales Executive, in addition to the implementation of the national clinical framework, has impacted on the prioritisation of Rare Diseases within the Health Board intermediate term plans. There is the need for increased awareness across Wales at Health Board level to align the Rare Disease Action plan with long term condition Quality statements. This will acknowledge the responsibility of NHS organisations to meet their obligations with respect to the actions required.

6. Next Steps

6.1 There is much to be celebrated within this progress report, and we are sincerely grateful to our WRDIG members and wider colleagues for the continued enthusiasm, time, effort and drive to the support the implementation of the Wales Rare Diseases Action Plan. However, we acknowledge that there is still so much to be done, and the refresh of the action plan acknowledging our completed actions as well as our ambitions for the future is the start of the next phase in impacting on the pathways of care for those with Rare Diseases.

6.2 With enhanced resources available to us and a robust foundation to work from, the next stage of our refresh of the action plan will show that we will aim to continue to deliver on focused actions in 2023/24. We are mindful of the challenges that exist within NHS Wales, with the financial challenges at the forefront of our minds. We also acknowledge the upheaval we will face, as we transition from an implementation Group to a Wales Rare Diseases Implementation Network, as part of the NHS Wales Executive. However, we will continue to use a collaborative and partnership approach

to ensure we keep the patients at the centre of our ambition, and ensure we use the principles of 'A healthier Wales: long term plan for health and social care'[10]

6.3 Our drive to develop a digital hub for those impacted by rare diseases continues to a key focus in the future, learning from the positive outcomes of the All-Wales Adult CF Centre support. Meeting all four priorities, as an accessible access to clinical support and research opportunities. It is anticipated that this will be a mechanism of reducing the burden of living with a rare disease to improve participation in research. Considering patient reported outcome and experience measures to enhance validation studies and clinical care whilst providing access to early-stage research and support.

6.4 Wales Rare Disease Implementation Network (WRDIN) will continue to work alongside Scotland, Northern Ireland, and England to improve the lives of those impacted by Rare Diseases. We look forward to joining Independent Advisory groups to share and learn from each other and identify new ways to develop solutions to common challenges in Rare Disease, as addressed in the Action Plans.

7. Conclusion

7.1 2022-23, has been an exciting time for Wales, with the start of a new team to drive WRDIG from delivery of the Wales Rare Disease Action Plan to its implementation. We would like to express our thanks to all those in Wales that have supported us previously and continue to do so, to build the foundations needed to for us to fulfil our ambitions. We have had continued support from patient representatives and third sector organisations, as well as our NHS partners, to ensure we remain focused on our objectives and are constantly challenged on our approach.

7.2 We remain focused on continuing to advocate for those in Wales who can't speak for themselves. Learning from others and collaborating nationally and internationally, we look to a time when all of those that are impacted with Rare Diseases can look forward to care closer to home, equity of access to treatments and services and support to be empowered to take an active part in their care.

10. https://www.gov.wales/healthier-wales-long-term-plan-health-and-social-care

Appendix A. Proposed Refresh Actions (minus measures)

Priority one: Helping patients get a final diagnosis faster.

Framework Priority section	Action	Refresh details
	Increase Whole Genome Sequencing testing for rare diseases.	Modify Action to detail target of 3000 WGS tests to be carried out by December 2025
	Whole Exome Sequencing (WES) Return Fetal Whole Exome Sequencing trios testing (FAGP service) to Wales.	Complete. Remove from actions in the forthcoming reporting period
Research Eco-system	Ensure a consent strategy is developed that enables researchers to securely and safely access routine genomic data generated by AWMGS for translational research purposes. Engagement with Health and Care Research Wales to ensure access to research studies for rare diseases patients.	Develop more robust collaborative approach between RDIG and Genomics Partnership Wales to ensure Rare Disease strategic aims are represented on Wales and UK consent conversations. Continue to look at opportunities to engage our Rare Disease population in research opportunities. Collaborate with the UK Rare Disease Forum and Framework boards to ensure challenges and actions are delivered systematically to reduce the inequalities of accessibility to research for the population in Wales.
Whole Transcriptome Sequencing	Ensure validation of a whole transcriptome service which will enable better understanding of RNA sequences to determine if a DNA	

	sequence is turned on and whether proteins have changed.	
New Action	Coproduce research questions with service users, to bring Rare Disease research closer into policy and practice.	Develop a working group to focus on Research and policy alignment. Bringing together Health and Care research Wales, Research and development department in Welsh Government and innovation leads across Wales. Ensure a Nurse representative is included alongside the research lead to ensure greater patient advocate voice within meetings. Understand a mechanism to bring together service users and researchers to understand the population needs. Engage with the current Rare Disease Research teams to bring together a community of practice to understand the needs of both the scientific, industry and academic environments.
New Action	Increase research activity in Paediatric Rare Disease research.	Engage with the Health and Care Research Wales to understand actions to improve opportunities to develop a strategy for paediatric research in Rare Disease. Observe research developments in the All-Wales Cystic Fibrosis service, to learn from opportunities for engagement.
	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. Explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices. 1. Non-Invasive Prenatal Testing (NIPT) will be expanded to other reproductive pathways to improve	Engage with Public Health Wales to understand the current process for developments in Newborn Screening for Rare Diseases. New Measures Ensure support and pathways are considered when new screening opportunities for families are developed. Combine NIPT into previous action.

	patient outcomes and optimise resource utilisation.	
	2. Implement a next generation sequencing service to detect genomic alterations when fetal structural abnormalities have been identified on ultrasound scan.	Complete. Remove from actions in the forthcoming reporting period
Service/Digital/Technical infrastructure	Ensure horizon scanning for commissioning requirements to inform the current National Genomic Test Directory for rare and inherited disease.	Modify measure to demonstrate increase in awareness of ability to test based on UK commissioning for additional UK genomic tests within the directory.
New Action	Equal access to genomic testing across the UK. Provision within Wales or referral outside of Wales.	WRDIG will Monitor the aligning of the respective test directories and information hubs so that they offer the same test options.
New Action	All nations should develop actions which support diagnosis and care for non-genomic conditions	WRDIG will Observe and report on opportunities to diagnose individuals with non-genomic conditions. Including supporting the MRC/NIHR research grant funding in Swansea University. WRDIG will develop a strategy on ongoing actions in the Wales Rare Disease Action plan that will build on the actions from the four nations workshop on non-genomics.

Priority two: Increasing awareness of rare diseases amongst healthcare professionals

Framework Priority section	Action	Refresh details
	Lead clinician for rare disease Monitor ongoing role and work programme of Clinical Lead and	

	Clinical Champion for rare diseases to raise profile of rare diseases.	
New Action	Clinical Champion Network development. Develop Implementation network with robust clinical insight	Develop a WRDIG 'Task and finish' group to understand how we engage with clinical colleagues across Wales, including how do we access knowledgebase in the wider workforce of qualified clinicians. Engage with strategic partners in Health Boards to enable discussion on implementing increased understanding of the Rare Disease patient pathways in regional. Develop and support the opportunities to bring together Nurses and Allied Health professional via the Rare Disease Nursing Network or the Global Nursing Network, supported by M4RD. Engage with unions and medical councils to promote published materials (RCN, GMC)
Education and Shared Learning	Education and Shared Learning •Survey qualified HCPs, undergraduates on their understanding and learning needs in rare disease. •Use results to develop training and development plan from baseline information on HCP understanding of rare diseases. •Incorporate rare diseases module in the undergraduate curriculum for medical students. •Continue to develop active partnerships with patients and patient advocacy groups (PAGs) •Recognise and celebrate rare disease day in secondary and primary care	New Measures Develop an WRDIG education working group to understand a framework to understand current education opportunities for Rare Disease awareness. Use the framework to understand the gaps in the provision of education to clinical and non-clinical individuals (including patient leadership) across Wales. Share learning and develop a communications strategy in collaboration with HIEW to disseminate a modified RISE survey into AHP and nursing undergraduate students. Consider the need for a post graduate qualification in Rare Diseases, based on the Valencia model. Consider the opportunity to influence Nursing and Allied Health Professionals training and delivery, based on evidence and access, via undergraduate commissioned contracts.

	•Improve health professional awareness through joint working between primary/secondary and tertiary care such as local pilot (Hywel Dda) Webinars for General Practitioners with AWMGS •Ongoing programme of WGP education and engagement with HCP and students including Genomic Counselling role (across Welsh Health Boards and HEIs) including precision medicine.	Remove HIEW from actions 2.3, 2.4 and 4.8, where they are not in the position to influence the outcomes
Improving Awareness of Rare Diseases with Data	Expand CARIS expansion to include adults affected by rare conditions. CARIS to collaborate with a small number of patient organisations to pilot research projects and generate patient data for a new adult register and allowing patients to self-report. 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 RDIG. 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.	WRDIG formally develop and support a 'Data Integration' working group to understand the opportunities to pilot collaboration between clinical colleagues and data analysts. To include Cardiff University DTII department and the digital and Innovation Office, Welsh government. Objectives: - Continue to develop new adult Rare Disease registries. - Scope the opportunities to use a digital health hub to support self-registration and use of PROMS and PREMS to support clinical care. - Consider the opportunity to use the new National Data Resource of Wales to understand the data linkage through NHS and non-NHS data sources to understand patient pathways. - Optimal use of data and data linkage for furthering rare disease care and research.

New Action	WRDIG will collaborate in the development of their registries.	WRDIG will collaborate with other nations to enable data scientists to link registries across the network to enhance accessibility to research capability.

Priority three: Better Coordination of Care

Priority Action section	Action	Refresh details
	Pathways of Care 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. Establish Rare Diseases as a "Community of Practice" and develop example/exemplar clinical pathways for rare disease conditions, including MDT involvement. Continued clinical pathway	Develop a process map for a selected rare condition to understand the components of service provision that can impact on the coordination of care, including: - the implementation of the transition guidance - Sibling support. - The use of 'Burdett' Nurses, Scotland. - Bridges the gap between healthcare and other services, such as education and benefits. Use the evaluation of a care navigator (case worker) within the SWAN clinic and the Paediatric Rare Disease MDT clinic in Cwm Taf Morgannwg UHB to understand the role and opportunities to share learning. - Delivering well-organised logistical support - Coordinates complex medical care - Integrates support from rare condition charities and support groups.
SWAN	Continue to build the establishment and assess/evaluate SWAN clinic.	
	Develop suitable PREM, PROMs for use in evaluation in the SWAN clinic with potential use across all rare disease patients.	Complete. Remove from actions in the forthcoming reporting period

New Action	Understand the usefulness of PREM PROM collation to develop enhanced service provision.	
Digital Patient Record	Establish an easily used "app" to enable a "patient passport" for rare disease patients	
Mental Health Services	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.	Develop collaboration alongside the Mental Health Strategic Clinical Network to develop a good practice guide for those impacted by Rare Diseases? New Measure WRDIG will scope collaboration with the Mental Health Strategic Clinical Network for Wales and charities, to develop opportunities to highlight and develop a strategy for mental health support for those impacted with Rare Diseases in Wales.
New Action Equity, Diversity and inclusion	The Wales Rare Diseases Action Plan will consider equity, diversity and inclusion throughout the refresh of the development and implementation of the action plans	The consultation on the refresh of the Wales Rare Disease Action plan will be considered by the Genomics Partnership Wales Sounding board. WRDIG will look for opportunities to work with RareQOL to define the needs of the Rare Disease communities in Wales. Including invitation to join RDIG (via Sondra Butterworth).

Priority Four: Access to medicines and treatments

Priority Action sections	Actions	Refresh details
	Ensure continued access to orphan and ultra-orphan medicines in Wales.	

	Ensure horizon scanning for new medicines for patients in Wales to allow timely awareness of new products and availability of new medicines. Monitor uptake of new rare diseases medicines and prescribing. Continue to develop improvements in the monitoring of use of medicines for patients with rare diseases including Blueteq	
New Actions	WRDID will build actions which support the use of repurposed and off-label medicines and devices	Measures to be determined.
New Action	WRDIG will observe opportunities to understand how pharmacogenomics can improve the effective management of those with rare diseases in Wales.	Work in collaboration with clinicians, genetics, biochemistry and pharmacy to develop guidance for Point of Care Testing in those with Rare Diseases, with pathogenic genetic alterations that will impact on specific treatment and quality of life.
?New Action	WRDIG will collaborate with strategic clinical networks in the NHS Executive, to develop an expert reference group to support and identify specialists and improve access to these for people with rare conditions.	Measures to be determined.
New Action	WRDIG will enhance the opportunity for research	WRDIG will horizon scan for new research study opportunities, to highlight to Rare Disease Clinical champions in Wales.

	participation for patients in Wales to further advances in treatments	WRDIG will scope the opportunity to develop a trials tracker to enable increased awareness of research studies.
Access to specialist Care	RDIG to continue to work with WHSSC and HEIW to ensure appropriate consultant specialist services in Wales. (Note some services will need to be provided outside Wales for specific conditions to ensure appropriate expertise and critical mass of patients).	New Measure WRDIG will develop an understanding of the specialist support offered to service users in Wales with Rare Diseases.