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Y Weithrediaeth  
Executive

# Wales Rare Disease Action plan 2022-2026

Refreshed January 2024

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## 1.0 Foreword

Wales continues to strive to improve the diagnosis and care of people with rare diseases, and in many domains is in the vanguard of delivery in the UK. The UK Rare Diseases Framework was launched in 2021, and the Wales Rare Diseases plan is an integral component.

Wales aims to deliver on the four main priorities of the plan - the Syndrome Without A Name (SWAN) clinic has helped patients get a final diagnosis faster; through collaborative working with Medics 4 Rare Diseases (M4RD) and Health Education and Improvement Wales, RDIG is increasing the awareness of rare diseases amongst healthcare professionals; the SWAN clinic and the Bevan Exemplar projects are offering better coordination of care for people with rare diseases and their families; and working in collaboration with the All Wales Medicines Strategy Group (AWMSG) RDIG aims to improve access to specialist care, treatment, and medicines. The emphasis now must be on taking the plan forward to a new level and improving the services and outcomes for patients with rare diseases, carers, and their families.

However, there are stormy waters ahead, and the NHS in Wales faces unprecedented clinical, societal and financial pressures. It will be challenging to maintain the progress made to date while also improving on the four priorities. RDIG are grateful for Welsh Government support - increasing the provision of rare disease coordinator time is particularly welcome, and this will be particularly important as RDIG transitions into the Wales Rare Diseases Implementation Network within the framework of the new NHS Executive in Wales.

I am very conscious that the plan as set out is based on the excellent work undertaken by my predecessor Dr Graham Shortland. He has been pivotal in improving the care of children with rare diseases in Wales, but with the success of the SWAN clinic he is now helping improve the care of adults with rare diseases in Wales. Finally, I would like to express my thanks to the members of RDIG and the support team without which no progress would have been made, and those many others who have contributed to the evolution of this plan.

Diolch yn fawr – Thank you  
Professor Iolo Doull RDIG Chair

## 2.0 Abbreviations/Glossary of Terms

<b>ATMPs</b>	Advanced Therapy Medicinal Products
<b>AWGL</b>	All Wales Genomics Laboratory
<b>AWMGS</b>	All Wales Medical Genomics Service
<b>AWTTC</b>	All Wales Therapeutics and Toxicology Centre
<b>CA</b>	Congenital Anomalies
<b>CARIS</b>	Congenital Anomaly Register and Information Service
<b>CES</b>	Clinical Exome Sequencing
<b>CU</b>	Cardiff University
<b>GPW</b>	Genomics Partnership Wales
<b>GRDNN</b>	Global Rare Disease Nurses Network
<b>HSST</b>	Higher Specialist Scientist Training
<b>ID</b>	Intellectual Disability
<b>MDT</b>	Multi-disciplinary Team
<b>M4RD</b>	Medics 4 Rare Diseases
<b>NCF</b>	National Clinical Framework
<b>NIPT</b>	Non-Invasive Prenatal Testing
<b>NSL</b>	New-born Screening Laboratory
<b>PAG</b>	Patient Advocacy Group
<b>PEG</b>	Patient Empowerment Group
<b>PHW</b>	Public Health Wales
<b>PREM</b>	Patient Reported Experience Measures
<b>PROM</b>	Patient Reported Outcome Measures
<b>RDIG</b>	Rare Diseases Implementation Group

<b>WRDIN</b>	Wales Rare Disease Implementation Network (from 2024)
<b>RDNN</b>	Rare Diseases Nurses Network
<b>SNP</b>	Single Nucleotide Polymorphism
<b>STP</b>	Scientist Training Programme
<b>SWAN</b>	Syndrome Without a Name
<b>UHB</b>	University Health Board
<b>WSC</b>	Wales Screening Committee
<b>WES</b>	Whole Exome Sequencing
<b>WG</b>	Welsh Government
<b>WGP</b>	Wales Gene Park
<b>WGS</b>	Whole Genome Sequencing
<b>WHSSC</b>	Welsh Health Specialist Services Committee
<b>WINGS</b>	Wales Infants and Children's Genome Service
<b>WSL</b>	Welsh Screening Laboratory

### 3.0 Executive Summary

Much has been achieved over the timescale since the first Welsh Rare Diseases Implementation Plan in 2014. The work of the Congenital Anomaly Register and Information Service (CARIS) in Wales continues. This was expanded to adult diseases during the COVID-19 pandemic and needs to continue to be resourced. The Wales Gene Park (WGP) working with patients and the public to involve them in rare and genetic research with Genetic Alliance UK is important on-going work.

The Genomics Precision Medicine Strategy for Wales was published in July 2017. This was followed by significant investment which allowed expansion of testing by All Wales Medical Genomics Service (AWMGS) and the on-going success of the **Wales Infant and Childrens Genome Service (WINGS)** project.

The Welsh Government's £80 million New Treatment Fund, introduced in 2017, has significantly sped up access to innovative new medicines in Wales, with rare diseases treated by medicines in the Fund including cystic fibrosis, Fabry disease, Gaucher disease and Batten disease.

In December 2022, The Genomics Delivery Plan for Wales was published. Published to coincide with the Wales Rare Disease action plan (2022-25). The plan details how Genomics Partnership Wales will continue to work in partnership with Welsh Government and other stakeholders, to harness advances in the understanding and application of genomics to transform public health strategy and delivery of care. Significant actions within this delivery plan, were developed in collaboration with the rare disease community, to enhance the progress of both plans to achieve greater benefit to those impacted by genetic Rare Disease conditions.

RDIG has continued in the last year to bring together stakeholders and with university health boards (UHB) and Trust representatives across Wales and Welsh Government to raise the profile of rare diseases since the first Welsh Rare Diseases Plan. Following the funding for an all-Wales clinical lead for Rare Disease and coordinator, we have seen progress noted within all the priorities, and have received international interest on our two-year pilot for a Syndrome Without a Name Clinic. Demonstrations of this progress has been published in our recent Wales Rare Disease Action plan report 2022/3.

The Wales Rare Diseases Action plan continues to be a rolling action plan, with refresh actions and measures being developed with collaboration to ensure it remains fit for purpose and meets the needs of the Rare Disease community in Wales.

In 2023, the NHS Wales Executive has been launched and within this new organisation RDIG will develop into a Wales Rare Disease Implementation Network (WRDIN). Our aim remains the same, to bring together delivery partners to develop and monitor Wales version of the new action plan. The four main priorities have been identified in the UK Plan:

**Priority 1 Helping patients get a final diagnosis faster.**

**Priority 2 Increasing awareness of rare diseases amongst healthcare professionals**

**Priority 3 Better coordination of care**

**Priority 4 Improving access to specialist care, treatment, and medicines**

This plan sets out the actions needed to improve our outcomes in these priority areas.

Much more still needs to be done. We have taken on the recommendations documented in the recent Genetic alliance reports on the four nations action plan reports. These have been included into our refreshed action plan and highlight continued challenges for those impacted by non-genetic Rare Diseases and with mental health support. The wider importance of support following a diagnosis by agencies wider than healthcare including social care and the third sector continue to be properly recognised and will need action.

There is a need to ensure wide representation of views from across the patient community, especially from those from those in hard-to-reach communities, but we have been fortunate to utilise the Genomic Partnership Wales sounding board, who continue to represent those with rare diseases with a genomic underlying diagnosis. We continue to look to our partners to support enhanced participation in our aims and vision, including the newly launched Llais Cymru. Previous successful areas of work along with new initiatives need to be taken forward at pace with appropriate resourcing. We have learnt a lot through the all-Wales Cystic Fibrosis service and have been utilising funding channels to enable opportunities to develop similar service provision for all those in Wales with Rare Diseases. It is important also to remember that much can be achieved without additional resource through new innovative practice and collaboration between various stakeholders.

In refreshing the plan, we have engaged with key stakeholder groups, these included Health Improvement and Education Wales, GPW and clinical groups. An important principle in those discussions was to consider the recent available published literature.

The NHS in Wales, is being impacted by financial restrictions, leading to recruitment freezes and challenges to maintaining current service provision for all those receiving health and social care support. COVID-19 still has left a lasting impact on accessibility of services, not just on those with Rare Diseases, However, improved accessibility for online support throughout this period and developments peer support and training for those not familiar with information technology, can be used as positives to improve outcomes for rare disease patients whilst avoiding inequalities of access to support.

The four nations Rare Disease Forum and framework boards and the development of the independent advisory groups, continue to highlight areas of good practice and opportunities to collaborate. Wales have been grateful to be highlighted as leaders in clinically led innovations, including the SWAN pilot, patient passport and the scoping of the digital health hub for Rare Diseases, whilst we are also learning from the other nations, in regards to education and training, research and registry conversations. This helps achieve the best outcomes across the UK whilst also realising that each nations plan will be different and unique.

Importantly in Wales we are fortunate to have robust mechanisms to influence policy direction, through the development of the NHS Wales Executive, and via our Rare Disease policy lead and research manager in the Welsh Government. Building our vision for patient centred care in Wales, RDIN will aim to a support priority setting partnership within James Lind to ensure we prioritise the needs of those we aim to support.

RDIN hope that the refreshed Wales Rare Disease Action plan continues to set out to produce a plan that is both realistic in the change we can achieve but aims “high” to produce significant change and meaningful outcomes which are relevant and can be measured. The plan will be constantly monitored, and changes considered periodically. There will be consideration of actions and changes on a rolling basis to ensure delivery of the plan and assessment of the plan’s achievements.

## 4.0 Introduction

### Background

A rare disease is defined as a condition which affects fewer than one in 2,000 people. It is currently estimated that there are more than 7,000 rare diseases, with new conditions continually being identified as research advances.

Whilst 80% of rare diseases have an identified genetic origin, they can also be caused by disordered immunity, infections, allergies, deterioration of body tissues and organs or disruption to development while in the womb.

Although rare diseases are individually rare, they are collectively common, with one in 17 people being affected by a rare disease at some point in their lifetime. In the UK this amounts to over 3.5 million people. This equates to about 170,000 people in Wales. It is therefore important that the NHS and other services provide this large and diverse patient population with the best possible care.

Rare diseases can be both life-limiting and life-threatening, and disproportionately affect children. Seventy-five per cent of rare diseases affect children and more than 30% of children with a rare disease die before their fifth birthday.

Rare disease patients and their families can face a lifetime of complex care and living with a rare disease can also have a huge impact on someone's education, financial stability, mobility, and mental health. It is vitally important that the voice of rare disease patients is included when developing wider health and social care policy.

### UK Rare Diseases Framework

Demonstrating the ongoing commitment to the rare disease community, and to build on the achievements of the previous strategy, the governments of all four UK nations have worked together with the rare disease community to design a new UK Rare Diseases Framework, which was published in January 2021.

This framework identifies the key priorities for rare diseases going forward and creates a vision for the future which is shared by all four UK nations to address health inequalities, improve the quality and availability of care, and improve the lives of people living with rare diseases.

The four key priorities are:

**Priority 1 Helping patients get a final diagnosis faster.**

The vision is for rare disease patients across the UK to get a final diagnosis faster and for research into previously unrecognised conditions to identify new rare diseases and provide new diagnoses.

**Priority 2 Increasing awareness of rare diseases amongst healthcare professionals**

The vision is for healthcare professionals to have an increased awareness of rare diseases and use of genomic testing and digital tools to support quicker diagnosis and better patient care.

**Priority 3 Better coordination of care**

The vision is for rare disease patients to experience better coordination of care throughout the patient journey.

**Priority 4 Improving access to specialist care, treatment, and medicines**

The vision is for rare disease patients to have improved access to specialist care, treatments, and drugs.

## 5.0 Wales Action Plan

### Previous Achievements

Since the first Wales Rare Diseases Plan was launched in February 2014, there have been a number of important developments.

The work of the Congenital Anomaly Register and Information Service (CARIS) in Wales continues, collecting data and reviewing the epidemiology of rare diseases in Wales with comparisons to wider population data being made.

Importantly CARIS works with the other four UK nations in developing registers further. The work of CARIS is continuing to expand into adult diseases during the COVID-19 pandemic, and support from RDIG is enabling further conversations within our clinical and academic communities, to enhance the utilisation of the data to highlight patient needs, enabling further bench marking of service provision.

RDIG has increased awareness of rare diseases by bringing together representatives from the health boards and trusts and professional groups and patient representatives. This enables shared learning and advocacy for different patient groups and services in Wales.

Support has been provided to enable the development of all-Wales initiatives such as guidelines for investigation of the child with developmental delay, which was launched in 2023. Work with Cardiff University has enabled the development of rare diseases teaching to undergraduate medical students and with HIEW and M4RD as new partners in RDIG, we look forward to expanding this within all clinical training and the development of a new RDIG education framework working group.

Representatives of the health boards and trusts have been looking at ways to raise the awareness of rare diseases in their hospital, primary care and community services throughout Wales including the use of patient stories. Clinical engagement remains challenging across Wales; however, we are hopeful that the use of clinical champions may highlight the opportunities to collaborate across Wales, using CARIS data.

Our strengthened Research links within the Welsh government, supported by the recently published rare disease research landscape report, will enable further conversations about how we can promote rare disease research participation and research studies within Wales.

Engagement with the rare disease community has been very important in developing the refreshed plan and has been a feature of ongoing engagement over several years. Following the initial support from the Genetic Alliance, Wales Gene Park and Genomics Partnership Wales (GPW), to develop the Rare Disease action plan in 2022, we continue to look to different ways to engage with our communities. The enhanced capacity of the RDIG coordinator, we hope will develop a strategy to streamline these opportunities.

GPW and its key partners, including NHS Wales, are committed to working in an open and transparent manner with patients and the public with personal or family experience of rare disease. Therefore, the Patient and Public Sounding Board remain a key opportunity to sense check the progress of RDIG. The future vision is for a more coproduction and codesign of our priority setting processes can be supported with our partners, thorough additional support by Llais Cymru and collaboration with Rae QOL.

The Genomics Precision Medicine Strategy for Wales was published in July 2017. This was followed by significant investment which allowed expansion of testing provision by AWMGS and the success of the WINGS project. The Wales Rare Diseases Action Plan will reflect planned expansion of this scheme, our progress report from 2023 demonstrates additional aspects we can be proud of in Wales.

The UK has also made important strides in the treatments made available for rare disease patients. The Early Access to Medicines Scheme (EAMS) was launched in 2014 to give people across the UK early access to new medicines that do not yet have a marketing authorisation, when there is a clear unmet clinical need. Since its launch, rare diseases patients living with, for example, Duchenne muscular dystrophy and haemophilia have benefited from the scheme with earlier access to life-changing treatments.

The Welsh Government's £80 million New Treatment Fund, introduced in 2017, has significantly speeded up access to innovative new medicines in Wales, with rare diseases treated by medicines in the Fund including cystic fibrosis, Fabry disease, Gaucher disease and Batten disease.

### Current Plan - 2023

To implement the UK Rare Diseases Framework, Wales has developed our own action plan, outlining commitments to meet the priorities of the Framework. In Wales, the RDIG has oversight for the development of this refresh of the Welsh Rare Disease Action Plan (see section 10 for description of governance arrangements).

Development of the UK Rare Diseases Framework was based on the [outcomes of the 'National conversation on rare diseases'](#), which took place in 2019. The conversation gathered views across the rare disease community on the major challenges faced by people affected by rare conditions across the UK.

The initial Wales Rare Disease plan was developed based on the 230 responses from Wales from this national conversation, which was been used to inform the development of this plan. The top issues highlighted by rare disease patients and their families were getting the right diagnosis, access to specialist medical care, awareness amongst health professionals and getting the right support.

In addition, in November 2021, two consultation workshops were organised and coordinated by Genetic Alliance UK and the previous RDIG Chair, Dr Graham Shortland. The aim of the workshops was to engage people affected by rare and genetic conditions in Wales and involve them in the initial phase of the development of a Welsh Action Plan, to implement the Framework.

Both sessions were held virtually via Zoom, engaging more than fifty people affected by or representing those affected by rare, genetic, and undiagnosed conditions across Wales. This report details a series of recommendations in relation to the four priority areas of the UK Rare Diseases Framework, based on data collected from the workshops. The underpinning

themes of the Framework must be incorporated across each priority area of the Welsh Action Plan and are also addressed separately.

In addition to the workshops, throughout 2019-2021, the Welsh Cross-Party Group on Rare, Genetic and Undiagnosed Conditions held meetings to discuss priorities for people affected by rare conditions in Wales.

In February 2021, the Cross-Party Group published a report with several recommendations that should be considered in the development of a Welsh plan:

1. The Welsh Action Plan must include commitments to improve mental health planning and service provision for those affected by rare conditions.
2. Transition services must be more flexible when defining the age of transition and supporting individuals holistically with all elements of their care - lack of a diagnosis should not be a barrier to accessing services.
3. Access to orphan and ultra-orphan medicines.
4. Impact of Covid-19 - data should be collected by Welsh Government that will enable assessment of the impact in terms of morbidity and mortality on people living with rare conditions.

During 2021, the Patient Empowerment Group (PEG), a group of patient advocates supporting people affected by rare conditions across the UK coordinated by Genetic Alliance UK produced a report in response to the UK Framework. The report details recommendations to inform the development of action plans across the four priority areas and underpinning themes.

This refresh Wales Rare Disease Action plan, has continued to build on insight from our Patient and Public engagement mechanisms, listening and learning from reports and feedback from our stakeholders. Considering the newly launched coordination of care report from the Genetic Alliance and the Rare Disease UK patient Empowerment Group Recommendations for the Rare Disease Action Plans (both published in 2023).

## Learning from coronavirus (COVID-19)

It is important to recognise the context in which this plan has evolved initially developed during the COVID-19 pandemic, and now in the context of the NHS recovery plans.

There is an ongoing need to provide services differently both in response to COVID-19 and to tackle the harms caused by COVID-19 such increases in waiting lists, delayed diagnoses, and treatment.

Many rare disease patients were identified, or were able to self-register for support, through the government shielding programme and it will be important to work with the rare disease community to understand how these individuals can be supported to share their experiences and shape the commitments developed under each of the priorities in this framework. There are many links between the issues raised by the rare disease community during the national conversation survey, the priorities in this framework, and the challenges faced during COVID-19. Access to coordinated specialist care became virtual.

Many of the measures brought in due to COVID-19, such as the increased use of technology and virtual appointments, remain beneficial for the rare disease community in the long-term but we must also learn where we can do better. There will continue to be opportunities to learn from COVID-19 and ensure that the experiences of the rare disease community feed into the implementation of the priorities in this new framework and wider national responses to infectious disease outbreaks.

People living with rare conditions have been placed under immense pressure by the prioritisation of care following COVID-19 pandemic. The new NHS Wales Executive will have to support increased collaboration between the strategic clinical networks to enable the prudent principles of care are supported across the healthcare settings. This gives us the opportunity to promote and facilitate access to appropriate support, information, care, and treatments for those who continue to have limited support for their complex needs within Wales and continue to suffer increased levels of social isolation.

## 6.0 Setting out the plan

The details of the plan continue to be set out in the same way as previously, with each of the four main priorities supplemented with some background information as to the reasons for the actions in the plan and divided into relevant sub-sections. Each action is structured in a similar way:

1. Action
2. Delivery partners / Stakeholders
3. Timeline
4. Measure / Outcome

### Priority 1 - Helping patients get a final diagnosis faster

#### Background

In taking forward the Welsh UK Rare Diseases Action Plan it is essential that priorities for building upon recent advances in genomic diagnostic technologies to help patients receive a final diagnosis faster and reduce the 'diagnostic odyssey' are included in the plan.

In April 2019, the AWMGS introduced rapid whole genome sequencing in new-born and paediatric intensive care units as the Wales Infants and Children's Genome Service (WINGS). To date a genetic diagnosis has been found in about 41% of patients, allowing for diagnostic and prognostic information to inform patient management.

The three-year genomics delivery plan (2022 - 2025) for Wales has been developed to significantly enhance the ability of genetic techniques to improve the ability of patients to get a final diagnosis faster. A roll-out of whole genome and exome sequencing to patients with a suspected rare disease is planned. The ambition is to sequence three thousand genomes before end of 2025. Raising of awareness of rare diseases (priority 2) and the methods of diagnosis to ensure the capacity is used will be required.

The continued participation in the research study on the genetic causes of illness by the AWMGS, will be achieved aiming to validate a whole transcriptome service. This will enable better understanding of RNA sequences to determine if a DNA sequence is turned on and whether proteins have changed.

The AWMGS, co-working with the early pregnancy unit in UCL research, have been 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, by observing how Non-Invasive Prenatal Testing (NIPT) can become part of the antenatal screening pathway for pregnant women in Wales.

An infrastructure providing suitable service, digital and technical infrastructure will be needed and resourced with a suitable financial structure.

### Whole Genome Sequencing (WGS) for rare diseases

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
1.1	Increase Whole Genome Sequencing testing for rare diseases.	AWMGS/WG/ WHSSC	All	Prioritisation of resources limits opportunities to support this action.

### Research Eco-system

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
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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.	GPW	2024	Prioritisation of resources limits opportunities to support this action.
1.3	Engagement with Health and Care Research Wales to ensure access to research studies for rare diseases patients.	RDIG/ Health and Care Research Wales	2024- 2026	Prioritisation of resources limits opportunities to support this action.
1.4	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.	AWMGS	2024	Prioritisation of resources limits opportunities to support this action.
1.5	Co-produce research questions with service users, to bring Rare Disease research closer into policy and practice.	WG/LSH/HCRW /GA	2023-25	Engage with MDT clinicians, GPW, research, service users and policy members to facilitate the creation of a working group, to discuss the recommendations of the rare disease research landscape report. In doing so, develop a draft

				strategy to ensure meaningful research questions are supported by the end of the September 2024.
1.6	Increase research activity in Paediatric Rare Disease research.		2024-2026	Prioritisation of resources limits opportunities to support this action.

### Prevention and Early Detection

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
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.	UK National Screening Committee, New-born Genomes Programme, Wales Screening Committee (WSC), GPW, PHW, National Screening	2024-2026	Prioritisation of resources limits opportunities to support this action.

		Laboratory (NSL), WHSSC		
1.8	Explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices, by expanding NIPT into other reproductive pathways to improve patient outcomes and optimise resource utilisation.	AWMGS, PHW, WSC	2024-2026	Prioritisation of resources limits opportunities to support this action.
1.9	Equal access to genomic testing across the UK. Provision within Wales or referral outside of Wales.	AWGMS/GPW	2024-26	Prioritisation of resources limits opportunities to support this action.
1.10	All nations should develop actions which support diagnosis and care for non-genomic conditions	GA/CARIS/CU	2024-26	Prioritisation of resources limits opportunities to support this action.

**Service/Digital/Technical Infrastructure**

<b>Priority</b>	<b>Action</b>	<b>Delivery partners/ Stakeholders</b>	<b>Timeline</b>	<b>SMART Objective</b>
1.11	Increase awareness of additional UK genomic tests newly commissioned within the genomic test directory for rare and inherited disease.	AWMGS/ WHSSC/	2024- 2026	Prioritisation of resources limits opportunities to support this action.

**Priority 2 Increasing awareness of rare diseases amongst healthcare professionals****Background**

An important development to improve clinical engagement (funded by Welsh Government) was the appointment, in April 2022, of a senior clinician as Clinical Lead and Champion for rare diseases working within RDIG. This enables the profile of Rare Diseases to be raised within health boards, trusts and stakeholders, and highlights opportunities to collaborate across sectors to the benefit of patients.

To understand and improve current levels of 'healthcare professional awareness', several actions are required:

- Improve the healthcare workforce's current basic knowledge of rare disease including what a rare disease is, national statistics, common challenges in rare disease, where to go for information.

- Improve patient-reported experiences of interacting with Health Care Professionals (HCPs)
- Build the systems required to support healthcare professionals to understand and play their role in their rare disease patient's journey.

Since 2017, WGP has been a strategic partner of GPW and member of its Workforce and Training Implementation Group. WGP has a well-established genetics and genomics-related programme of Education, Engagement and Involvement initiatives, which includes raising health professional awareness of rare, genetic, and undiagnosed conditions.

Integral to this programme is the involvement of those impacted by Rare Diseases, to ensure the lived experience informs the education of health professionals, as well as empowering those taking part. Going forward, this will also be an essential aspect of the new Genomics Delivery Plan for Wales and the Rare Diseases Action Plan.

Also, it is important to raise awareness about rare diseases by improving the collection of data and making available data for healthcare planning. This includes the expansion and continued support for CARIS to develop and record adult registry data.

RDIG will continue work going forward with the other UK nations to develop an information hub to share across the rare disease community.

## Lead Clinician for Rare Diseases

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
2.1	Monitor ongoing role and work programme of Clinical Lead and Clinical Champion for rare diseases to raise profile of rare diseases.	RDIG, health boards, trusts and all stakeholders	April 2025	Prioritisation of resources limits opportunities to support this action.
2.2	Clinical Champion Network development. Develop Implementation network with robust clinical insight	RGIG/HB/Unions and medical councils.	2024-2026	Prioritisation of resources limits opportunities to support this action.

## Education and Shared Learning

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
2.3	Survey qualified HCPs, undergraduates on their understanding and learning needs in rare disease.	HEIW M4RD (undergraduate project in	2023-24	Convert the RISE survey into an Allied Health Professional and Nursing format, to disseminate to undergraduate students. Results will be analysed by May 2024, to support the development of an

	Use results to develop training and development plan from baseline information on HCP understanding of rare diseases.	planning stage) Universities, Global and UK Rare Diseases Nurses Network (GRDNN/RDN N) ,CU		education framework by September 2024.  Consider the opportunity to influence Nursing (midwifery and health visitor) and Allied Health Professionals training and delivery, based on evidence and access, via undergraduate commissioned contracts.
2.4	Incorporate rare diseases module in the undergraduate curriculum for medical students.	RDIG, Universities	2024 - 2026	Prioritisation of resources limits opportunities to support this action.
2.5	Ensure development of specialist consultant - interest and confidence	RDIG, HIEW	2024-26	Prioritisation of resources limits opportunities to support this action.
2.6	Clinical Nurse Specialists – build understanding of paediatric and adult CNS workforce	HIEW, with support from RDIG	2023-24	Incorporate future need for paediatric and adult Rare Disease CNS into nursing workforce plan.  Build profiles in Careersville for existing roles within Rare Diseases
2.7	Continue to develop active partnerships with patients	Welsh training institutions,	2023- 2026	Enhance patients and carers leadership and self-management skills by increasing numbers of

	and patient advocacy groups (PAGs)	Genetic Alliance, RDIG, WGP		participants affected by Rare Diseases on the expert patient programme by 10% in 6 months. Utilising existing resources within Improvement Cymru and Collaborating with patient advocate organisations and clinicians by June 2024
2.8	Recognise and celebrate rare disease day in secondary and primary care.	RDIG, Genetic Alliance	Annually	Collaborate with the Welsh government, Service users and Health boards to develop a plan to celebrate the Rare Disease Day in 2024, by January 2024. This will enable a wider understanding of Rare Diseases in Wales.
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 with AWMGS	AWMGS, RDIG, Hywel Dda UHB	2024	Prioritisation of resources limits opportunities to support this action.
2.10	Ongoing programme of WGP education and engagement with HCP and students	WGP GPW AWMGS	2023-26	Prioritisation of resources limits opportunities to support this action.

	including Genomic Counselling role (across Welsh Health Boards and HEIs) including precision medicine.	Rare Disease Community, Wider genomics community		
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### Improving Awareness of Rare Diseases with Data

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
2.11	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.	CARIS, RDIG, WG, Genetic Alliance, Cardiff University	2023-2024	Facilitate the Rare Disease registry working group (CARIS/Cardiff University and RDIN), to complete one adult rare disease registry in CARIS. Working with clinical specialities and the NHS Wales Executive health intelligence to generate new data and insight regarding patient numbers and demographics By September 2024.

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 RDIG	RDIG and relevant stakeholder groups with health board/trust representatives	2024-26	Prioritisation of resources limits opportunities to support this action.
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.	RDIG (health board/trust members) and CARIS	2024-2026	Prioritisation of resources limits opportunities to support this action.
2.14	WRDIG will collaborate in the development of Rare Disease registries in the UK	NIRDIG/NCARD S/SRDIN	2024-26	Prioritisation of resources limits opportunities to support this action.

### Priority 3 Better coordination of care

#### Background

Care coordination in transition services needs to be improved, particularly between paediatric to adult care and between diagnosis to treatment of rare conditions in line with Welsh Government's ['Transition and handover from children's to adult health services'](#) guidance published in February 2022.

Commissioning agreements should include the provision of care co-ordinators for transition between paediatrics and adults and specialist services generally. All paediatric patients should receive a named worker and care plan to support them through this process. A digital care plan that can be shared with professionals across health, social care and education and controlled by the patient or family would be of greatest benefit. This should link with any technology utilised to support Priority 1 and the patient passport mentioned later in this section.

Sharing of data and communication between healthcare professionals and patients in respect of cross border healthcare and treatment must be addressed.

Commissioning of specialised services must include funding for dedicated care coordination support for patients and their families and to act as a liaison between the patient and centres/ professionals in Wales and the specialist centre.

A major opportunity for the rare disease community is through the implementation of National Clinical Framework published in March 2021. It is a vital part of a much broader effort that was described in 'A Healthier Wales'. It sets out a vision for how clinical services in our NHS fit into that wider picture and how we can begin to realise ambitions through the development of a learning health and care system. It seeks to unleash the revolution and recognises that greater central direction is needed to make that behaviour and philosophy a reality.

The Framework sets out a health system that is coordinated nationally and delivered locally or through regional collaborations such as potentially a rare diseases Community of Practice.

This includes producing the principles of pathways of care for rare diseases patients and some example/exemplar pathways. Pathway development should consider the inclusion of 'red flag' for clinicians and will be a means by which there is improving co-operation between primary, secondary, and tertiary care.

There should be recognition in these pathways of the provision of multi-disciplinary care (the need for co-ordination of psychological services is recognised) and time for multi-disciplinary team (MDT) meetings.

In the past year, RDIG have collaborated with a not-for-profit company Care Circle, to understand how a digital platform could offer support for those impacted by rare disease. This innovation, modelled on the all-Wales Cystic Fibrosis service is scoping a one point of access portal for peer support, AHP guidance, leadership and empowerment coaching, but also can support the development of a patient passport, alongside the existing products being developed in Wales and Cambridge.

The SWAN clinic initiative is an all-Wales initiative to assess (with a two-year pilot) the opportunity for formally establishing this service. This assists a more rapid diagnosis, but also aims to improve co-ordination of care and support for patients for whom there is no diagnosis. In the past year, this pilot has gained international recognition and has led to the development of Rare Disease PROMS and PREMS to support its evaluation. In addition, one of our clinical champions is piloting a paediatric Rare Disease MDT clinic, both these models are observing the effectiveness of coordination of care and the utilisation of a care navigator.

Addressing the mental health needs of rare disease patients is fundamental part of improving their well-being and care. Consideration must be given as to how best to address these needs which could include establishing good practice guidelines to ensure the mental health needs of rare diseases patients are recognised and incorporated into "routine" care for patients. Advice and recommendations are available already as part of the Rare Disease UK publication "Living with a rare condition: the effect on mental health (2018)".

A recent four nation workshops discussing mental health in children and young people with rare diseases, is due to be reported shortly and its guidance will support ongoing actions to develop a strategy to enable support mechanisms to be put in place.

### Pathways of Care

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objectives
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.	RDIG, WHSSC, WG	2024-2026	Prioritisation of resources limits opportunities to support this action.
3.2	Establish Rare Diseases as a “Community of Practice” and develop example/exemplar clinical pathways for rare disease conditions, including MDT involvement.	RDIG, Rare Diseases Clinical Lead WG, Clinical Programme Director for the NCF, UK Rare Disease	2023-2026	Support the development of UK Rare Disease Quality Standards as part of the UK Rare Disease Independent Advisory Group (IAG). By monitoring uptake of service user insight gathering from Wales, to enhance a four nations vision for quality service provision. To raise awareness of Rare Diseases patient needs in Wales and prioritisation of service provision in Wales. Meeting the timelines

		Forum IAG, James Lind Alliance		anticipated by the IAG, and the subsequent James Lind Alliance partnership report on activity by September 2024.
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### SWAN Clinic

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
3.3	Continue to build the establishment and assess/evaluate SWAN clinic.	WG, WHSSC, Cardiff and Vale UHB	2023-2024	Prioritisation of resources limits opportunities to support this action.
3.4	Understand the usefulness of PREM and PROM collation to develop enhanced service provision.	WHSSC, Cardiff and Vale UHB	2024	Prioritisation of resources limits opportunities to support this action.

### Digital Patient Record

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
3.5	Establish an easily used "app"	RDIG, Life	2023-	Collaborate internationally to develop a 'once for

	to enable a “patient passport” for rare disease patients	Sciences Hub Wales, Industry partners, CamRare, Rare Care Centre Western Australia	2026	all’ rare disease passport by facilitating ‘Task and finish’ meetings with the four nations, industry and Australia, to pilot a paper passport to the Welsh Rare Disease population. Using the draft CamRare Patient passport, modifying based on initial evaluation. RDIG will disseminate the draft paper passport by April 2024, and support analysis of initial findings. To support the production of a final version of a paper pt passport by September 2024.
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### Mental Health Services

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objective
3.6	Ensure the mental health needs of rare disease patients, carers and significant others including siblings are considered as part of the overall mental health strategy for Wales and consider whether further	RDIG, health boards, WG	2024-26	Prioritisation of resources limits opportunities to support this action.

	guidance is needed such as a good practice guide for rare disease patients.			
3.7	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.	NHS Executive, mental health strategy, CU, charities	2024-26	Prioritisation of resources limits opportunities to support this action.

### Equity, Diversity and Inclusion

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objectives
3.8	The Wales Rare Diseases Action Plan will consider equity, diversity and inclusion (EDI) throughout the refresh of the development and implementation of future	GPW/ RareQol/ Public Health Wales/Llais Cymru/Genetic Alliance	2024-26	RDIG will engage with key stakeholders in the EDI community to understand how needs differ in underrepresented communities. By meeting with and working with RareQOL, Llais Cymru, Public Health Wales and the Rare disease community, we will ensure we have EDI representation on RDIG. We will use existing resources to highlight

	Wales Rare Disease action plans			opportunities to embed EDI within 2023/4 priorities and in the 2024 refresh of the Wales Rare Disease action plan, by September 2024.
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#### **Priority 4 Improving access to specialist care, treatment, and medicines.**

##### Background

Access to orphan and ultra-orphan medicines are important. The UK should aim to make rare disease medicines available in the NHS as close to receipt of market authorisation as possible where there is a clear unmet clinical need, and the medicine provides (added) value to the NHS in association with an appropriate commercial agreement.

Consideration of real-world NHS Wales evidence collated during a potential period of managed access could be used to inform a pre agreed Health Technology Assessment (HTA).

Horizon scanning for new drugs for patients in Wales is important to ensure timely awareness of new products and availability of new medicines. The team at All Wales Therapeutics and Toxicology Centre (AWTTC) gathers information about new medicines, indications and formulations that are in development and are expected to be licensed and made available in the UK in the next financial year.

They also collect information about Advanced Therapy Medicinal Products (ATMPs) in development that may become available in the next three to five years. This supports the planning, introduction and faster adoption of new medicines in NHS Wales, particularly those that may have significant cost or service planning implications.

The horizon scanning team use several sources to collect information about new medicines being developed including the UK-wide horizon scanning database [UK PharmaScan](#). This is the horizon scanning team's primary source of information about new medicines, indications and formulations in development. There is still a place for good clinical engagement and involvement by clinicians. RDIG needs to ensure proper awareness of the horizon scanning functions and support for clinicians.

Recognition of the availability and use of new medicines, including consideration of medicines for rare cancers, needs to be made freely available and this will be helped by the introduction of new technologies including Blueteq. This is a high-cost drug management system which requires a form to be completed by a doctor for any patient who is prescribed a high-cost drug. This has many benefits including improved speed of access to drugs for patients.

There must also be recognition of the need for a sustainable workforce including Consultant Specialists in Wales requiring workforce planning by HEIW and WHSSC to ensure access to specialist care.

### Access to Medicines and Treatment

Priority	Action	Delivery partners/ Stakeholders	Timeline	SMART Objectives
4.1	Ensure continued access to orphan and ultra-orphan medicines in Wales.	AWTTC, RDIG and WHSSC	2024-2025	Prioritisation of resources limits opportunities to support this action.
4.2	Ensure horizon scanning for new medicines for patients in Wales to allow timely	RDIG (health board)	2024-26	Prioritisation of resources limits opportunities to support this action.

	awareness of new products and availability of new medicines.	representatives), AWTTC		
4.3	Monitor uptake of new rare diseases medicines and prescribing.	RDIG, AWTTC	2024-26	Prioritisation of resources limits opportunities to support this action.
4.4	Continue to develop improvements in the monitoring of use of medicines for patients with rare diseases including Blueteq	WHSSC, AWTTC, RDIG	2024-26	Prioritisation of resources limits opportunities to support this action.
4.5	WRDID will build actions which support the use of repurposed and off-label medicines and devices	WHSSC, AWTTC, RDIG, LSH	2024- 2026	Prioritisation of resources limits opportunities to support this action.
4.6	WRDIG will observe opportunities to understand how pharmacogenomics can improve the effective management of those with rare diseases in Wales.	Biochemistry (C&VUHB), Audiology, neonatology, AWMGS, RDIG	2023- 2025	RDIG will facilitate discussions on how a Point of Care (POC) genomic test can be piloted for within Rare Diseases patients by developing a task and finish working group, including key stakeholders from genomics, biochemistry, audiology and neonatology. To observe how Wales can support NICE guidance on providing preventative

				genomic testing to those at risk of hearing loss in treatment of aminoglycosides in those with a deletion in the m1555A>G. By September 2024
4.7	WRDIG will enhance the opportunity for research participation for patients in Wales to further advances in treatments	WG, LSH, RDIG	2024-2026	Prioritisation of resources limits opportunities to support this action.

### Access to Specialist Care

Priority	Action	Delivery Partners/ stakeholders	Timeline	SMART Objective
4.8	RDIG to continue to work with WHSSC to ensure appropriate consultant specialist services in Wales. (Note some services will need to be provided outside Wales for specific conditions to	RDIG, HEIW, WHSSC	2024-2026	Prioritisation of resources limits opportunities to support this action.

	ensure appropriate expertise and critical mass of patients).			
4.9	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.	NHS Exec, CARIS, RDIG	2024-26	Prioritisation of resources limits opportunities to support this action.

## 7.0 Monitoring and Evaluation

RDIG will continue to meet regularly to report on progress. This will include a process of constant review and any changes to the plan will be considered on an annual basis.

This will include those actions completed during the year and timescales for those not completed and barriers to completion. A number of developments will be subject to agreement of available resources and ongoing/successful business cases.

By developing the plans in a clear way with:

1. Actions
2. Delivery partners / Stakeholders
3. Timeline
4. Measure / Outcome

It will provide a platform for objective measurement of the success of the plan.

Measuring outcomes for patients, carers and families is always going to be difficult. As part of a piece of work flowing from the SWAN pilot, we are looking to develop PREMs, PROMs and patient experience surveys for patients with rare diseases.

This should be a piece of on-going work to roll out across our services and is included in the plan. Patient stories are a powerful way to continue to work with patient groups to tell both positive and negative experiences. The partnership and collaborative working with patients are essential to learn whether we are making real differences to the lives of patients, carers, and families in all four priority areas.

RDIG and partners will work with the other UK nations and their equivalent groups to ensure that best practice is considered for implementation in Wales. Likewise, Wales and its rare diseases community will actively take part in joint working with the other UK four nations to share the work from Wales.

## 8.0 References and Important Publications/Policies

- [UK Rare Diseases Framework \(2021\)](#)- which sets out the priorities for rare diseases over the next five years, including faster diagnosis, increased awareness amongst health professionals, improved care coordination, and better access to treatments
- [SWAN UK CYMRU \(syndromes without a name\)](#) Families in Wales affected by undiagnosed and rare conditions to be better supported
- [Genome UK: 2021 to 2022 implementation plan](#) - setting out the priorities for this period in driving progress for Rare Disease
- [WHSCC Genomics](#) - Specialised Services Policy Position PP184. The Welsh Health Specialised Services Committee approve funding against the National Genomic Test Directory for rare and inherited disease. Details and provision of these tests are available through AWMGS.
- [Life Sciences Vision](#) - outlining the priority areas to build on the advances made in the sector through the pandemic to improve health outcomes for other diseases and health conditions
- [UK NHS Long Term Plan](#) - which aims for 500,000 whole genomes to be sequenced by 2023/24 and places a key focus on supporting the development of innovative technologies and improving patient access to improve health outcomes
- [NHS Genomic Medicine Service Alliances](#) - with the aim of supporting the use of genomics in routine patient care across the country
- [NICE guidance Overview - Risdiplam for treating spinal muscular atrophy \(SMA\)](#) - the NHS have completed a new commercial deal for the drug Risdiplam, which can help improve mobility in children and adults suffering from the rare genetic condition

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- Genetic Alliance UK. Rare Experience 2020: the lived experiences of people affected by genetic, rare, and undiagnosed conditions. Available at: <https://rareexperience2020.geneticalliance.org.uk/wp-content/uploads/2021/05/KW-Changes-050221-Rare-Experience-2020-Report-.pdf>
- Sickle Cell Society. No One’s Listening: An inquiry into the avoidable deaths and failures of care for sickle cell patients in secondary care. Available at: <https://www.sicklecellsociety.org/wp-content/uploads/2021/11/No-Ones-Listening-PDF-Final.pdf>
- Royal College of General Practitioners. Unpublished poll data from event with M4RD. 2020. Data available from Lucy McKay on request [lucy@m4rd.org](mailto:lucy@m4rd.org)
- Medscape. Rare Disease Education: Insights on what clinicians know, want and need. 2020. Unpublished data available from Lucy McKay on request [lucy@m4rd.org](mailto:lucy@m4rd.org)
- Evans, W.R.H., Tranter, J., Rafi, I. et al. How genomic information is accessed in clinical practice: an electronic survey of UK general practitioners. J Community Genet 11, 377–386 (2020). <https://doi.org/10.1007/s12687-020-00457-5>
- Medics4RareDiseases. Priority Two: The key to unlocking the UK Rare Diseases Framework. 2021. Available at: <https://www.m4rd.org/2021/01/31/the-framework/>
- Rare Disease UK publication “Living with a rare condition: the effect on mental health (2018)”.
- [Transition and handover from children's to adult health services | GOV.WALES](#)
- [National clinical framework: a learning health and care system | GOV.WALES](#)
- Co-ordinated care for people affected by rare diseases: the CONCORD mixed-methods study. Health and Social Care Delivery Research, No. 10.5. Morris S, Hudson E, Bloom L, et al. Southampton (UK): [NIHR Journals Library](#); 2022 Mar.
- [A healthier Wales: long term plan for health and social care: Welsh Government](#)

- [Rare Disease Research landscape report](#) Authored by the Rare Disease Research Landscape Steering Group on behalf of MRC/NIHR 2023
- [Rare Disease Patient Empowerment Group recommendations for the rare disease action plans 2023](#) Genetic Alliance
- [Coordination of care, learning from the experiences of people living with rare conditions](#) Genetic Alliance

## 9.0 Partner Organisations

This list sets out those involved in formulating this plan and is by no means exhaustive as individually there have been many contributions for which we are very grateful and have been considered in agreeing this plan.

All Wales Medical Genomics Service  
All Wales Genomics Laboratory  
All Wales Therapeutics and Toxicology Centre  
Congenital Anomaly Register and Information Service  
Patient Advocacy Group, Wales Gene Park  
Genomics Partnership Wales  
Health Education and Improvement Wales  
Welsh Health Specialist Services Committee  
NHS Wales Executive

Public Health Wales

Welsh Government

Rare Diseases Nurses Network

Medics 4 Rare Diseases

New-born Screening Laboratory, Cardiff and Vale UHB

University Health Boards in Wales

Wales Infants and Children's Genome Service

Wales Screening Committee

Genetic Alliance UK

Wales Gene Park

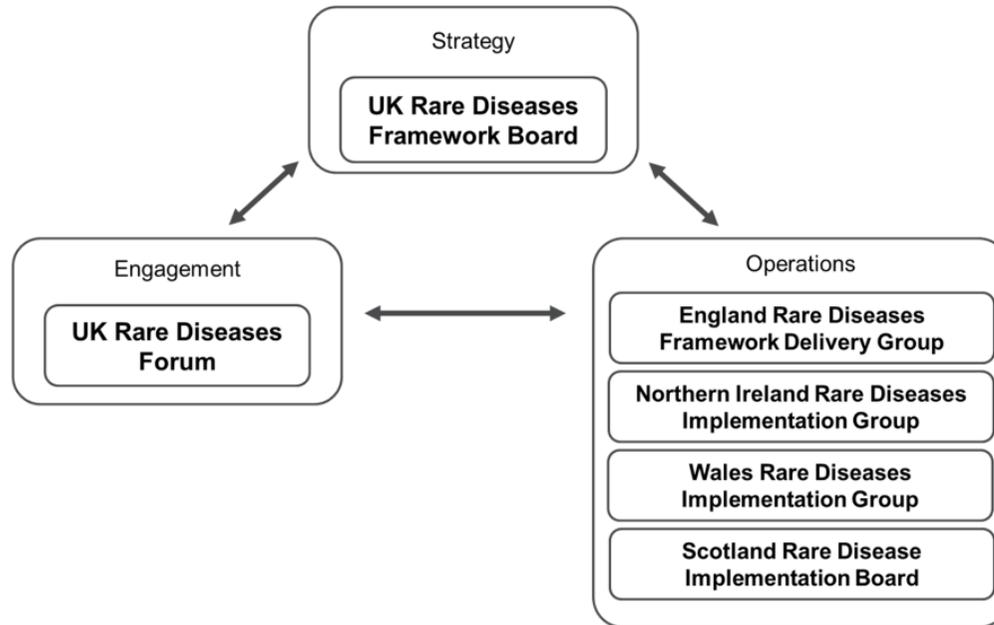
Cardiff University

Life Science Hub Wales

Rare Care Centre Australia

Commonwealth Rare Disease Network

## 10. Governance



As shown, the UK-wide UK Rare Diseases Framework Board provides strategic oversight and facilitates alignment of policy across the four UK nations.

The UK Rare Diseases Forum, also UK-wide, provides a means of engagement with the community. Through the online platform it provides an opportunity to engage continuously with a broad range of people from the rare disease’s community, providing an opportunity for discussion and feedback, as well as a source of updates on progress and related initiatives.

The Wales Rare Disease Implementation Group (RDIG) works with health boards and partner organisations acting as the mechanism for the development and oversight of the action plan for Wales.

A UK wide newsletter is also produced which provides policy updates on implementation and progress as well as relevant news on rare disease developments in each country.