GIG CYMRU NHS WALES
Cydweithrediad Iechyd GIG Cymru
NHS Wales Health Collaborative

Wales Rare Diseases Action Plan

2022 - 2026

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1.0 Foreword

In January 2021 the four nations of the United Kingdom launched the UK Rare Diseases framework which sets out the UK wide priorities.

The Welsh Government remains committed to improving the lives of people living with a rare disease through improvements in diagnosis, awareness, treatment and care.

The Rare Diseases Implementation Group (RDIG) brings together delivery partners to develop and monitor Wales progress. This plan highlights progress already made and sets out a number of specific actions we will take in Wales to deliver against the priorities in the UK framework.

It is important to recognise the context in which this plan was developed during the COVID-19 pandemic and the impact this has had and the ongoing pressures within the NHS.

Much has been achieved since the first plan was launched in 2014 and some of those achievements are highlighted in this plan.

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.

This is an exciting opportunity and through collaboration and joint working, Wales has the ability and opportunity to achieve significant change and improvements.

It is particularly pleasing to see funding for an all-Wales Clinical Lead, co-ordinator support and the two-year pilot funding for the Syndrome Without a Name Clinic now in place.

Whilst continuing joint working with all four UK Nations it is imperative to the success of the plan that this is a unique plan for Wales to draw on long established strengths, such as the excellence in genetic services and strengthen and develop new areas and ideas for improving care.

I would wish to say thank you to all those who have helped by contributing content and suggested ideas for the plan in addition to the members of RDIG, without which this plan would not have been possible.

It has been a privilege to Chair RDIG since 2014 and as I hand over the Chair in May 2022 to Professor Iolo Doull, I wish success to all those contributing to the work and most important of all improved services for patients, carers and their families.

Diolch – Thank you

Dr Graham Shortland OBE, Outgoing Chair and on behalf of the Rare Diseases Implementation Group

May 2022

2.0 Abbreviations/Glossary of Terms

ATMPs	Advanced Therapy Medicinal Products
AWGL	All Wales Genomics Laboratory
AWMGS	All Wales Medical Genomics Service
AWTTC	All Wales Therapeutics and Toxicology Centre
CA	Congenital Anomalies
CARIS	Congenital Anomaly Register and Information Service
CES	Clinical Exome Sequencing
GPW	Genomics Partnership Wales
HSST	Higher Specialist Scientist Training
ID	Intellectual Disability
MDT	Multi-disciplinary Team
M4RD	Medics 4 Rare Diseases
NCF	National Clinical Framework
NIPT	Non-Invasive Prenatal Testing
NSL	New-born Screening Laboratory
PAG	Patient Advocacy Group
PEG	Patient Empowerment Group
PHW	Public Health Wales
PREM	Patient Reported Experience Measures
PROM	Patient Reported Outcome Measures
RDIG	Rare Diseases Implementation Group
RDNN	Rare Diseases Nurses Network
SNP	Single Nucleotide Polymorphism
STP	Scientist Training Programme
SWAN	Syndrome Without a Name
UHB	University Health Board
WSC	Wales Screening Committee
WES	Whole Exome Sequencing
WG	Welsh Government
WGP	Wales Gene Park
WGS	Whole Genome Sequencing
WHSSC	Welsh Health Specialist Services Committee
WINGS	Wales Infants and Children’s Genome Service
WSL	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.

RDIG has sought to bring together stakeholders and with university health board (UHB) and Trust representatives across Wales and Welsh Government to raise the profile of rare diseases since the first Welsh Rare Diseases Plan. It is particularly pleasing that collectively the members of RDIG have successfully secured Welsh Government (WG) funding for co-ordinator support, a new all-Wales Clinical Lead for Rare Diseases, and also setting up a two-year pilot for a Syndrome Without a Name Clinic (SWAN).

The Wales Rare Diseases plan is being launched as part of the new UK Rare Diseases Framework (January 2021). RDIG brings 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. Areas such as mental health and the wider importance of support following a diagnosis by agencies wider than healthcare including social care and the third sector are being properly recognised and will need action.

There is a need to ensure wide representation of views from across the patient community. Previous successful areas of work along with new initiatives need to be taken forward at pace with appropriate resourcing. Sometimes that will require new funding and business cases. It is important also to remember that much can be achieved without additional resource through new innovative practice and collaboration between various stakeholders.

Throughout development of the plan, we have held a number of stakeholder events and joint working with Genetic Alliance UK and Wales Gene Park to try and ensure a wide range of views and ideas about what should be included in the plan are captured. An important principle in those discussions was to consider the available published literature such as the CONCORD study.

Despite the unprecedented pressures from COVID-19 on the NHS and social care services more generally there are positives that have come out of the COVID-19 pandemic which can lead to improved outcomes for rare diseases patients. Not least the use of virtual consultations and improved information technology.

Learning between the four nations will continue through co-ordination and alignment of the plan by the UK Rare Diseases Framework Board and the formal and informal structures of the UK Rare Diseases Forum. 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 have the on-going opportunity to influence Welsh Government through the Senedd Cross Party Group on Rare, Genetic and Undiagnosed Conditions, working with Genetic Alliance UK.

Working collaboratively across Wales we 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 on an annual basis. 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 diseases 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 was expanded to adult diseases during the COVID-19 pandemic.

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 is ongoing work. Work with Cardiff University has enabled the development of rare diseases teaching to undergraduate medical students as part of their curriculum.

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.

Research initiatives locally have included Wales Orphan and Rare Lung Disease which promoted the awareness and understanding of rare lung disease in West Wales.

Engagement with the rare disease community has been very important in developing the new Welsh plan and has been a feature of ongoing engagement over a number of years.

The first Genomics Cafes were held in June 2019. Organised by Wales Gene Park (WGP) on behalf of Genomics Partnership Wales (GPW), Genomics Cafes are free events for people affected by a rare or genetic condition and held in various locations across Wales.

Genomics Cafes are a relaxed and informal opportunity for individuals to meet others, find out more about new advances in genomic medicine in Wales and advise GPW how they can be better supported. Genomics Cafes are a networking opportunity, and guest speakers are also present to highlight new initiatives and give attendees the chance to shape activities in genomics. Since the COVID-19 restrictions were introduced in March 2020, these cafes have moved to an online platform.

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. A Patient and Public Sounding Board was established in 2019 and membership includes individuals from across Wales with varied experience of genetic conditions including rare disease.

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.

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 - 2022

To implement the UK Rare Diseases Framework, Wales has developed their own action plan, outlining commitments to meet the priorities of the Framework. In Wales, the RDIG has oversight for the development of a Welsh 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.

This included over 230 responses from Wales which have 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 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 a number of 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.

Learning from coronavirus (COVID-19)

It is important to recognise the context in which this plan was developed during the COVID-19 pandemic and the impact this has had on NHS services, the ongoing pressures within the NHS and the necessary changes which have taken place such as video consultations.

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 their experiences with COVID-19 and how this can 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, will be beneficial for the rare diseases community in the long-term but we must also learn where we can do better. There will 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 COVID-19 pandemic. Access to appropriate support, information, care, and treatments has become more difficult and levels of social isolation have increased. Services have been disrupted and it is unclear when reintroduction of some services will resume. Access to personal protective equipment has been challenging, and there has been inflexibility in the education system to respond to individual needs and to adapt practices for children with rare conditions.

6.0 Setting out the Plan

The details of the plan are set out for each of the four main priorities. Each section is further supplemented with some background information as to the reasons for the actions in the plan and divided into relevant sub-sections with "colour" coding for easier consideration of the actions. 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 45% of patients, allowing for diagnostic and prognostic information to inform patient management.

A detailed 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 five thousand genomes annually within the next five years. Raising of awareness of rare diseases (priority 2) and the methods of diagnosis to ensure the capacity is used will be required.

Further understanding of the genetic causes of illness will be achieved by validation of 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.

In April 2018, the AWMGS launched Non-Invasive Prenatal Testing (NIPT) as part of the antenatal screening pathway for pregnant women in Wales who are found to have a higher chance of a child with Down's, Edwards and Patau syndromes.

The NIPT is offered as an alternative to an invasive test. Wales was the first UK nation to introduce NIPT. Building upon this, NIPT will be expanded to other reproductive pathways to improve patient outcomes and optimise resource utilisation.

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	Measure/ Outcome
1.1	Increase Whole Genome Sequencing testing for rare diseases.	AWMGS/WG/ WHSSC	2022/23	Increased number of tests performed

Whole Exome Sequencing (WES) for rare diseases

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure
1.2	Return Fetal Whole Exome Sequencing trios testing (FAGP service) to Wales.	AWMGS/WG/ WHSSC	2022/23	Number of tests performed /returned to Wales for testing

Whole Transcriptome Sequencing for rare disease

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
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.	AWMGS	2022/23	Validation of methodology

Research Eco-system

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
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.	WGP	2022	Publication of consent strategy allowing improved access to genomic

				data for research purposes. Increased number of patients entering research studies.
1.5	Engagement with Health and Care Research Wales to ensure access to research studies for rare diseases patients.	RDIG/ Health and Care Research Wales	2022-2026	Increased number of rare diseases patients entering studies.

Prevention and Early Detection

Priority	**Action**	**Delivery partners/ Stakeholders**	**Timeline**	**Measure/ Outcome**
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.	UK National Screening Committee, New-born Genomes Programme, Wales Screening Committee (WSC), GPW, PHW, National Screening Laboratory (NSL), WHSSC	2022-2026	Increased number of rare diseases diagnosed by screening.

1.7	Explore how genomic testing can continue to be best used in reproductive medicine to support parents to make informed choices. 1. NIPT will be expanded to other reproductive pathways to improve 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.	AWMGS, PHW, WSC	2022-2026	Number of tests performed

Service/Digital/Technical Infrastructure

Priority	**Action**	**Delivery partners/ Stakeholders**	**Timeline**	**Measure-Outcome**
1.8	Ensure horizon scanning for commissioning requirements to inform the current National Genomic Test Directory for rare and inherited disease.	AWMGS/ WHSSC/	2022-2026	Improved test availability

Priority 2 Increasing awareness of rare diseases amongst healthcare professionals

Background

An important development to improve clinical engagement (funded by Welsh Government) is the appointment, in April 2022, of a senior clinician as Clinical Lead and Clinical Champion for rare diseases working with RDIG, health boards, trusts and all stakeholders to raise the profile of rare diseases and initiate appropriate workstreams in discussion with partners.

To understand and improve current levels of 'healthcare professional awareness', a number of 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 affected, 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 diseases community.

Lead Clinician for Rare Diseases

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
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 2022	Review of achievements of the role by RDIG, NHS Wales Health Collaborative and WG after two years in post

Education and Shared Learning

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
2.2	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.	HEIW M4RD (undergraduate project in planning stage Universities Rare Diseases Nurses Network (RDNN) RDNN	2023/24	**Within two years:** Improved awareness of rare diseases amongst healthcare professionals
2.3	Incorporate rare diseases module in the undergraduate curriculum for medical students.	RDIG, HEIW, Universities	2022 - 2026	Improved awareness of rare diseases amongst medical students.

2.4	Continue to develop active partnerships with patients and patient advocacy groups (PAGs)	HEIW, Welsh training institutions, Genetic Alliance, RDIG, WGP	2022-2026	Increased number of people with a rare disease involved in course delivery.
2.5	Recognise and celebrate rare disease day in secondary and primary care.	RDIG, Genetic Alliance	Annually	Reporting to RDIG on health board/trust programmes by their representatives
2.6	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	2022	Improved awareness of rare diseases in primary care.
2.7	Ongoing programme of WGP education and engagement with HCP and students including Genomic Counselling role (across Welsh Health Boards and HEIs) including precision medicine.	WGP GPW AWMGS Rare Disease Community, Wider genomics community	2022/23	Metrics (attendance) and evaluation of activities including number of workforce engaged.

Improving Awareness of Rare Diseases with Data

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
2.8	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	2022/23	Increased number of new conditions incorporated into the CARIS programme.
2.9	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	2022/23	Maintain and improve compliance.
2.10	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	2022-2026	Improved access to specific condition-based data on a geographical basis.

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 diseases 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 co-ordinated 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 flags' 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.

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 addition, joint work between RDIG, Betsi Cadwaladr UHB and industry is being enabled by funding from the Welsh Health Hack (Life Sciences Hub Wales) to develop a 'patient passport' for rare diseases patients to improve patient experience.

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)".

Pathways of Care

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
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	2022-2026	Improved transitional care for rare disease patients.
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	2022 (create first pathway) 2023-2026 (continue pathway development)	Improved patient experience and improved pathways of care.

SWAN Clinic

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
3.3	Continue to build the establishment and assess/evaluate SWAN clinic.	WG, WHSSC, Cardiff and Vale UHB	2021 - 2023	Improved patient outcomes/ diagnosis.
3.4	Develop suitable PREM, PROMs for use in evaluation in the SWAN clinic with potential use across all rare disease patients.	WHSSC, Cardiff and Vale UHB	2022	Improved patient reported outcomes/ Experiences

Digital Patient Record

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
3.5	Establish an easily used "app" to enable a "patient passport" for rare disease patients	RDIG, Betsi Cadwaladr UHB, Life Sciences Hub Wales, Industry partners.	2022	All rare disease patients have access to a 'patient passport'.

Mental Health Services

Priority	Action	Delivery partners/ Stakeholders	Timeline	Measure/ Outcome
3.6	Ensure the mental health needs of rare disease patients and carers are considered as part of the overall mental health	RDIG, health boards, WG	2022 - 2023	Improved mental well-being for rare disease patients.

	strategy for Wales and consider whether further guidance is needed such as a good practice guide for rare disease patients.			

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	Measure/ Outcome
4.1	Ensure continued access to orphan and ultra-orphan medicines in Wales.	AWTTC, RDIG and WHSSC	2022-2025	Improved access to orphan and ultra-orphan medicines.
4.2	Ensure horizon scanning for new medicines for patients in Wales to allow timely awareness of new products and availability of new medicines.	RDIG (health board representatives), AWTTC	2022/23	Improved access to new medicines and appropriate uptake.
4.3	Monitor uptake of new rare diseases medicines and prescribing.	RDIG, AWTTC	2022	Improved access to new medicines for rare disease patients.
4.4	Continue to develop improvements in the	WHSSC, AWTTC, RDIG	2022-2026	Improved access and

	monitoring of use of medicines for patients with rare diseases including Blueteq			effective use of medicines.

Access to Specialist Care

Priority	Action	Delivery Partners/ stakeholders	Timeline	Measure/ Outcome
4.5	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).	RDIG, HEIW, WHSSC	2022-2026	Rare disease patients have access to appropriate specialist opinions.

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.

That 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 four 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

- Department of Health and Social Care. The UK Rare Diseases Framework. 2021. Available at: https://www.gov.uk/government/publications/uk-rare-diseases-framework/the-uk-rare-diseases-framework .
- 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
- Medscape. Rare Disease Education: Insights on what clinicians know, want and need. 2020. Unpublished data available from Lucy McKay on request 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

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 Collaborative

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

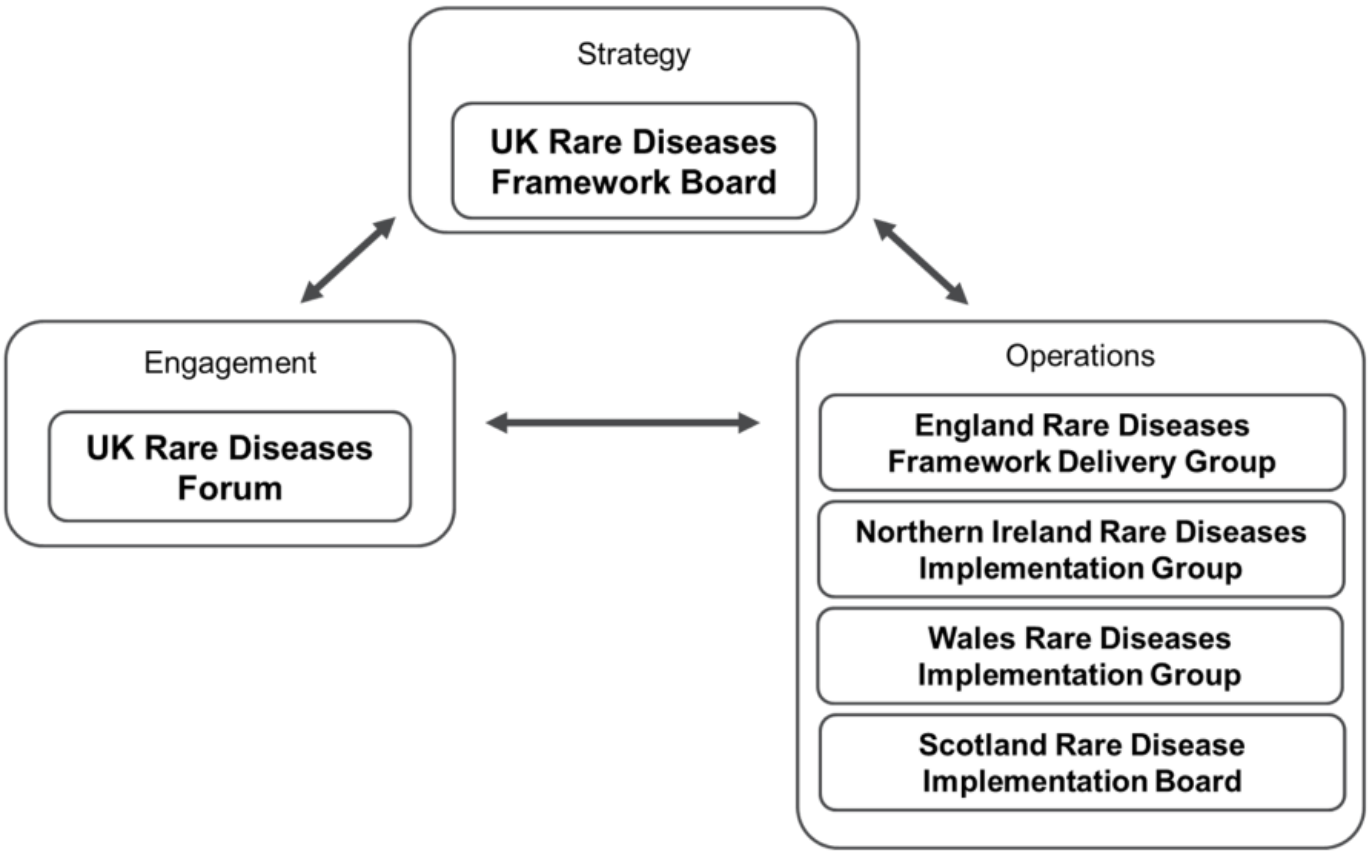
Wales Infants and Children's Genome Service

Wales Screening Committee

Genetic Alliance UK

Wales Gene Park

10. Governance



As shown above, 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.