Response to Rare Diseases Action Plan Powys Teaching Health Board

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

1.1 Powys Teaching Health Board (PTHB) participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

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

1.2 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

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

1.3 This is an action for the All Wales Medical Genomics Service.

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

1.4 This is an action for the Wales Gene Park.

				data for research purposes. Increased number of patients entering research studies.	1.5 This is an action for the Rare Diseases Group and Care and Research Wales.
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.	1.6 This is an action for all-Wales bodies. PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

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	1.7 This is an action for all Wales bodies.

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	1.8 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

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

2.1 PTHB does not directly provide District General Hospital services, specialised services, or clinical laboratory services. It is a provider of primary and community services. This means it does not have the types of secondary care clinicians or clinical directorates which are in place in other health boards. It commissions secondary services from neighbouring DGHs and NHS Trusts. It has a very small number of community paediatricians in-county. PTHB receives the papers of the Rare Diseases Implementation Group but does not have the capacity to attend each meeting. PTHB's key focus is participation in the Welsh Health Specialised Services Committee and improving local care co-ordination for people with complex needs.

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 (undergraduat e 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.

2.2, 2.3, 2.4, 2.5, 2.6, 2.7
There is no University in Powys.
These are actions for all Wales or other bodies.

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.

2.8 This action is for all Wales or other bodies.

2.9 Rare Disease Implementation Group papers are received. PTHB implements data standards issued through Digital Health and Care Wales.

2.10 This is an action to be led by the Rare Diseases Implementation Group and CARIS.

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.

3.1 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.
PTHB is working to implement and ensure compliance with Welsh Government requirements in relation to transition.

3.2 This action is for all Wales or other bodies.

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

3.3, 3.4 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

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'.

3.5 This action is for all Wales or other bodies.

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.			

3.6 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan. WHSSC is carrying out a review of the psychological support services for people receiving specialised services.

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 monitoring of use of medicines for patients with rare diseases including Blueteq	WHSSC, AWTTC, RDIG	2022-2026	Improved access and effective use of medicines.

4.1 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

4.2, 4.3 and 4.4 These are actions led by other bodies. PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.

4.4 The use of Blueteq is now mandated. PTHB has worked with WHSSC and Welsh Government on this matter.

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.

4.5 PTHB participates in the Welsh Health Specialised Services Committee and the Integrated Commissioning Plan for specialised services is reflected in the PTHB Integrated Plan.