“Falling Off a Cliff: A Digital Carabiner for Rare Disease Young People and Their Families Transitioning to Adult Care”

Jamie Duckers, Zoe Morrison, Darcie Williams, Rhiannon Edwards on behalf of the Wales Rare Care Centre Leadership group & Laura MacDonald (Carecircle)

Contact:
Jamie.Duckers@wales.nhs.uk

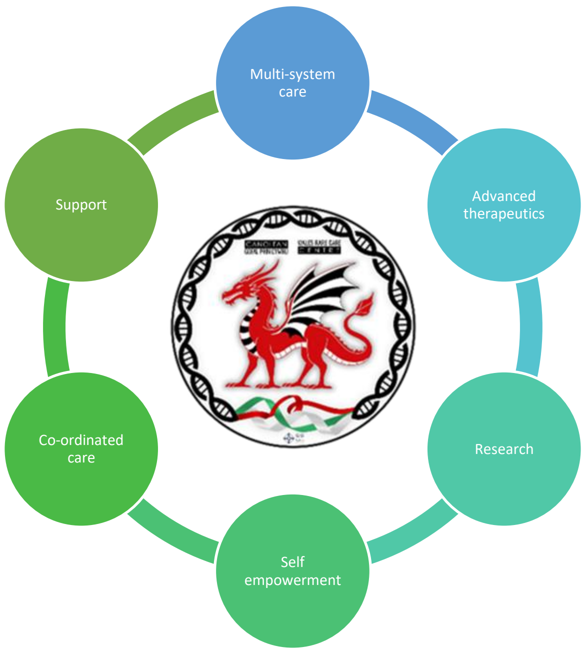
Project Background:

Rare diseases affect 1 in 17 people globally, including around 180,000 in Wales, many facing life-limiting and complex conditions with limited treatment options or care pathways.

Individuals and families often experience isolation, delayed diagnoses, and fragmented support. The “diagnostic odyssey” can take years, with patients repeatedly explaining their history to different professionals.

Even after diagnosis, they face ongoing challenges coordinating health, education, and welfare needs with little support.

This lack of integration fuels health inequalities, limits access to research and NHS services, and contributes to declining mental, physical, and social wellbeing, increasing reliance on care resources



Project approach:

1. Industry partner developed the rare disease platform.
2. Clinicians and lived experience groups defined priorities.
3. Governance, cybersecurity, and partnerships with NHS and charities followed.
4. Stakeholder engagement, PPIE groups, and shared learning shaped progress.
5. Combined resources improved access and equality.
6. Ongoing development invites community input into the Wales Digital Rare Care Centre vision.

Results:

The Wales Digital Rare Care Centre is already highlighted as an example of good practice. The NICE Rare Disease Quality Standards and other UK rare disease framework guidance documents are currently being developed, focusing on public requirements for a one point of access digital platform to improve awareness of rare diseases and to share resources and peer support.

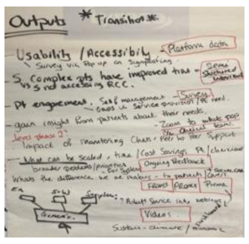
Project aims:

Develop a Wales digital rare care centre (WDRCC)	Pilot peer support content and horizon scan for technology	Understand the impact of online service provision to NHS Wales	Peer clinical support and share opportunities across rare disease clinical services
•In three SE Wales health boards, complete IG guidance to enable access for all. •Complete a 5-year contract between health board and industry partners for continued development. •Engage four rare disease clinical teams to support the pilot; developing resources and onboarding families onto the platform for evaluation and co-creation.	•Develop peer support groups and access to resources developed to facilitate a smooth transition from paediatrics to adult services. •Work with clinicians to understand the current gaps in support and potential solutions. •Work with Expert Patient Programmes to improve access to leadership and empowerment skills support.	•Enrol in the climate emergency dragons heart academy. •Continue stakeholder engagement about the impact of the WDRCC on the Welsh Government digital and data strategy for health and social care in Wales.	•Bringing Medical and social care professionals working with rare conditions together •Signposting each other to relevant resources to help increase access to support materials for professionals and families alike. •Regular peer support sessions and shared learning opportunities.

Project impact:

The Wales digital Rare Care centre is the first of its kind globally

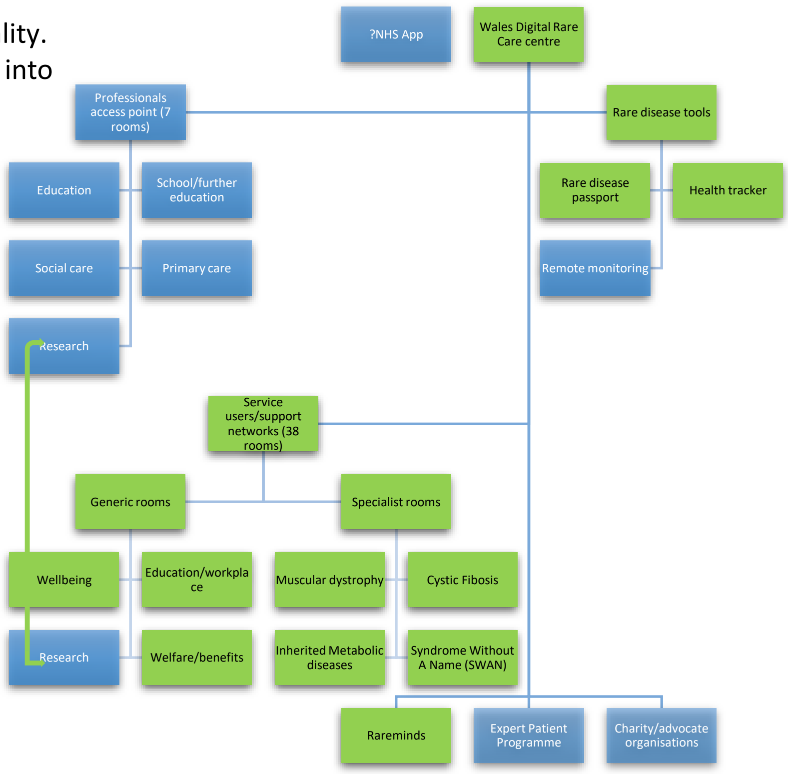
It has been highlighted in Global and national conferences as a mechanism for global health connection.


Outputs *Transition*
Usability / Accessibility
Platform data

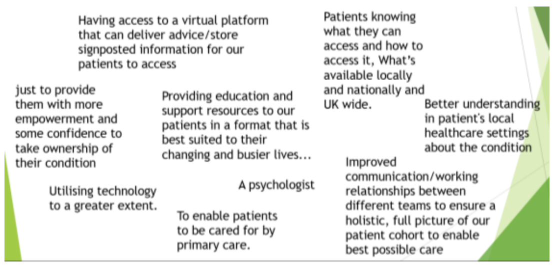
The Rare Disease community of interest group have developed into a peer support group, enhancing collaboration.

Artificial intelligence is being developed within a safe environment and controlled by clinical expertise,

The development of a rare disease MDT peer support group will have benefits further than the boundaries of this project and help the clinical team members involved in rare disease care to share learning and gain valuable support from each other.



Pre Project stakeholder hopes


Having access to a virtual platform that can deliver advice/store signposted information for our patients to access

Patients knowing what they can access and how to access it, What’s available locally and nationally and UK wide.

just to provide them with more empowerment and some confidence to take ownership of their condition

Providing education and support resources to our patients in a format that is best suited to their changing and busier lives…

Better understanding in patient’s local healthcare settings about the condition

Utilising technology to a greater extent.

A psychologist

To enable patients to be cared for by primary care.

Improved communication/working relationships between different teams to ensure a holistic, full picture of our patient cohort to enable best possible care

One access point, currently links to three separate, but interlinked hubs. 48 spaces (rooms) have been requested and developed by stakeholders and accessible in 5 languages (including Welsh). 186 documents have been uploaded, including videos. 64 clinicians and non-clinicians are using the site

#BevanExemplar #EsiamplwyrBevan

Achieving Change Together | **Trawsnewid Iechyd Gyda'n Gilydd** |www.bevancommission.org