carecircle
the world's health service tm

“Falling off a cliff: A digital carabiner for rare disease young people and their families transitioning to adult care”

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On behalf of the Wales Digital Rare Care Centre team

22nd January 2026

GIG CYMRU NHS WALES
Bwrdd Iechyd Prifysgol Caerdydd a’r Fro
Cardiff and Vale University Health Board

Rare Diseases

1 in 17 people are affected, 180-194,000 in Wales (in comparison 200,000 are affected by Diabetes)

7000-8000 different Rare Diseases

Limited or non-existent clinical service provision in Wales

Only 5% of Rare Diseases have validated treatments available

Approx 75% high-cost drugs are for rare diseases.

Challenges for families

- Difficult for families to access relevant, accurate and up-to-date information
- HCPs lack knowledge and awareness of rare diseases
- Burden for families to source information themselves
- Lack of integration across health, education and social care
- Inequitable access to resources depending on condition and/or clinical team's knowledge
- You don't know what you don't know
- The rarity of rare diseases makes it difficult to connect with support networks or those with lived experience

Challenges for Clinicians

- Wellbeing Support
- Losing engagement through transition
- Patient Empowerment
- Our patients live 2+ hours away
- Lack of psychological support
- Need to significantly pivot our service model
- Pts unaware of all the information
- (not) Creating dependency on service
- Limited capacity
- Support outside of scheduled clinic appointments.

Wales Digital Rare Care centre (WDRCC)

Co-ordinated care

Advanced Therapies

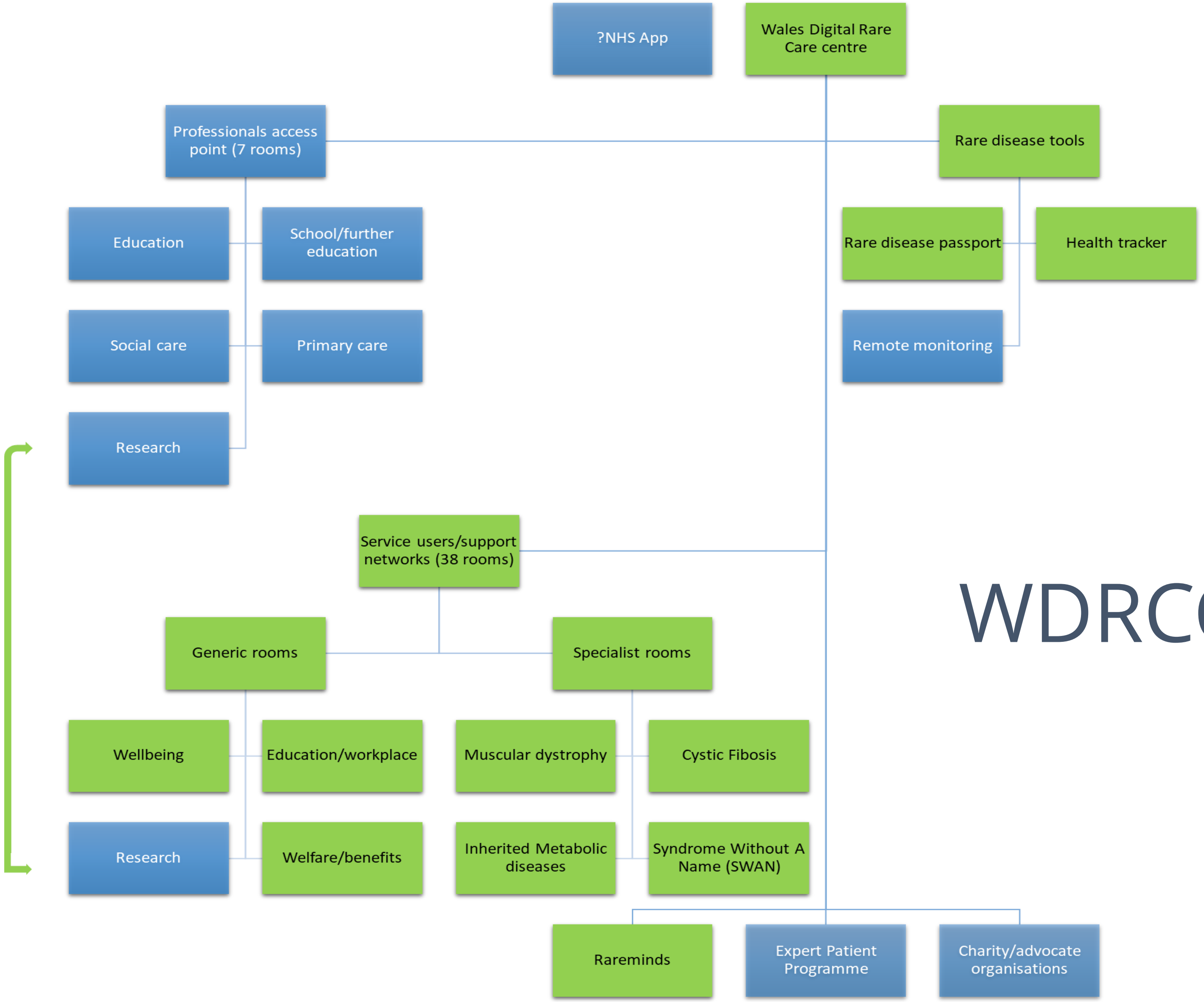
Research

Self empowerment

Multi-system care

Support

WDRCC Room set up


?NHS App

Wales Digital Rare Care centre

Professionals access point (7 rooms)

Education

School/further education

Social care

Primary care

Research

Rare disease tools

Rare disease passport

Health tracker

Remote monitoring

Service users/support networks (38 rooms)

Generic rooms

Specialist rooms

Wellbeing

Education/workplace

Research

Welfare/benefits

Muscular dystrophy

Cystic Fibosis

Inherited Metabolic diseases

Syndrome Without A Name (SWAN)

Rareminds

Expert Patient Programme

Charity/advocate organisations

🟩 Current

🟦 Future

Benefits

- Connections with all sources of support
- Access to directive and simplified information
- Expansion of information across all populations
- Track health using clinical tools
- Store and share health data including genomic data and images
- Ability for support networks to connect with others with lived experience
- Building community and reducing feelings of isolation
- Patient voice will be represented at unprecedented scale.
- Research tools which support real time interventions
- Communicate and receive information in their native language.
- Ownership of own data.
- Tools to help prepare for appointments.

A new model of support for rare diseases communities across Wales

Develop a Wales Digital Rare Care Centre (WDRCC)

- 3 SE Wales health boards
- 5-year contract between health board and industry partners
- Engage four rare disease clinical teams.
- Rare Disease Patient passport
- Health tracking

Pilot peer support & horizon scan for technology.

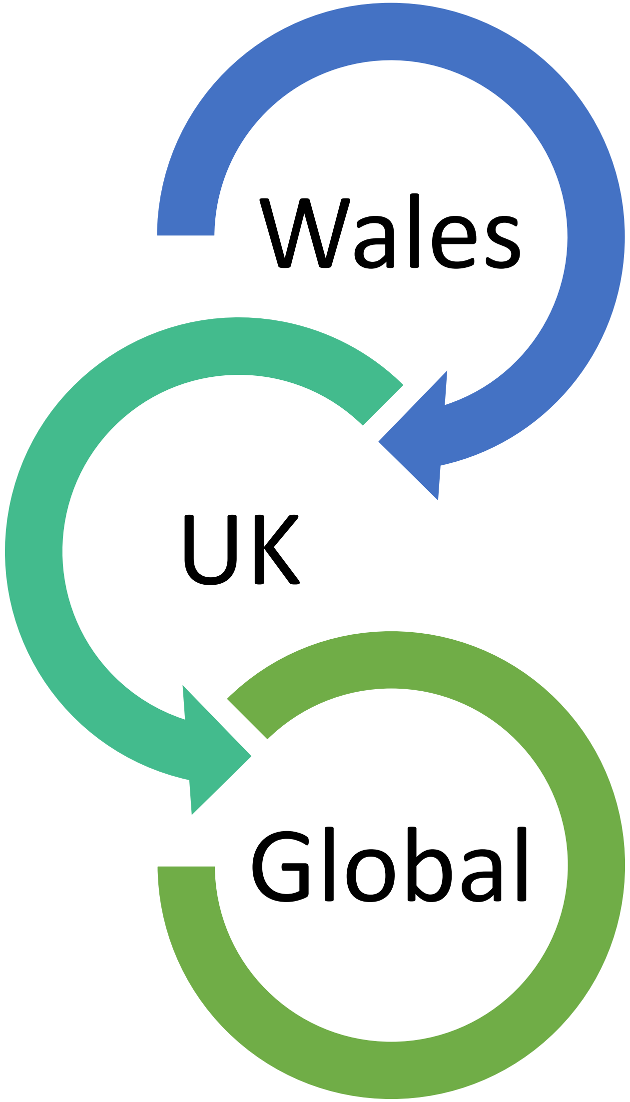
- Develop peer support groups and access to resources
- To understand the current gaps in support and potential solutions.
- Work with NHS and non -NHS services.

Impact of online service provision to NHS Wales

- Continue stakeholder engagement about the impact of the WDRCC on the rare disease population.
- Work alongside Welsh Government digital and data strategy for health and social care in Wales.

Opportunities across rare disease clinical

- Health and social care working together
- Bring together all resources that can support all people impacted is RD.
- Educate and support non specialist clinicians



Wales

UK

Global

High Level Successes

- ✓ 5-year industry/health board partnership. IG successfully obtained to focus ongoing development of rare disease pt passport.
- ✓ Collaborative Rare Disease clinical support group (64 members)
- ✓ Shared resources, open access to all with rare diseases (186, translatable into 5 languages)
- ✓ Charity support and piloting new ways of working
- ✓ Global awareness – DAVOS and HIMMS conferences
- ✓ Clinical leadership post recruited too – Rare Disease Consultant Nurse
- ✓ Award winning Digital Innovation that will transform Rare Disease care globally