

Improving Lives, Optimising Resources: How a strategy for rare diseases will benefit the people of Wales



RARE DISEASE | UK

The National Alliance for people with rare diseases & all who support them

In Wales, 175,000 people are likely to be affected by a rare disease at some point in their lives (1 in 17 people). Collectively, rare diseases are not rare and they represent a significant health burden to health and social services in Wales.

Rare Disease UK's (RDUK) report *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy* describes how currently there are a number of obstacles preventing patients and families being able to access the high-quality services, treatment, care and support that would ensure they benefit from optimum health outcomes. The report sets out recommendations developed in collaboration with a broad range of stakeholders in Wales and across the UK to address these issues. These recommendations should form the basis of an effective strategy for rare diseases.

A strategy for rare diseases would ensure that patients receive the highest quality healthcare whilst ensuring the most efficient and effective use of NHS Wales resources.

How RDUK's recommendations for a strategy for rare diseases would enable the Welsh Assembly Government to improve healthcare for people with rare diseases in Wales:

Patient-centred, holistic care based on the needs of the individual

RDUK's recommendations encourage this by, for example, the production of individual and regularly updated care plans, the designation of Care Coordinators, the development of specialist clinics for rare diseases and the production of personalised information prescriptions for patients. Holistic care is promoted by the inclusion of the delivery of appropriate psychological, social and financial support within a patient's care plan, and easier access to information on patients' and families' non-medical needs.

Top quality care and services available to all in Wales regardless of their location or condition

RDUK's recommendations facilitate equitable, timely access to effective diagnostic tests, drugs and therapies for all patients that need them, regardless of their location or the rarity of their condition. Recommendations to aid timely diagnosis would avoid unnecessary or harmful interventions and ensure early access to effective interventions and better management of a patient's condition.

We also make recommendations for planning structures that enable equitable access to health services and treatments across Wales and to ensure that unnecessary barriers are removed to allow patients to access the best care and support for their condition.

Collaborative approach to healthcare service provision and integrated care

Adopting RDUK's recommendations would encourage effective collaboration and communication between everyone involved in a patient's care; it would improve continuity of care at transition periods; it would help to ensure clear communication between patients and professionals, including information on their condition, management and treatments. We promote the development of centres of excellence, care pathways and networks of experts to aid communication, and the use of multidisciplinary teams and, where relevant, multidisciplinary clinics to encourage collaboration and holistic care.

Making the most effective use of available resources in Wales to improve health outcomes for patients with rare conditions

Implementing RDUK's recommendations will ensure the most effective use of resources whilst promoting the best possible health outcomes and experiences of the NHS for patients with rare diseases in Wales. Patients with rare diseases already make heavy demands on the resources of both health and social services, but these resources are often used inefficiently due to delays in diagnosis, misdiagnosis, fragmented care, a lack of information, few guidelines on the effective management of rare conditions and limited effective treatment options.

In June 2009 the UK Government adopted the Council of the European Union's *Recommendation on an action in the field of rare diseases*. This calls on member states to develop plans or strategies for rare diseases. The Welsh Assembly Government (WAG) has committed to working collaboratively with the other home nations to develop a strategy for rare diseases. RDUK calls on the WAG to take on board the recommendations outlined in *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy* and implement an effective strategy for rare diseases in Wales.

RDUK looks forward to working in partnership with WAG and other key stakeholders to develop an effective strategy to ensure that people with rare diseases are able to access the highest quality care, treatment and support.

Some excellent examples of good practice already exist in Wales. It is vital that these are developed or used as models on which to base new services. Where expertise already exists, it should be shared and utilised to benefit others.

Research and Education in Wales: an example of good practice

The **Wales Gene Park** brings together all those with genetics expertise in Wales, facilitates knowledge transfer to the NHS and commercial sectors and promotes professional education and public involvement in the field of genetics. It also helps to support genetics researchers throughout Wales.

The key activities of the Wales Gene Park are:

- Putting Wales at the forefront of genetics research
- Identifying the genetic cause for diseases
- Translating research into medical practice
- Education of professionals
- Stimulating public debate of genetic issues

As well as carrying out strategic research in various areas of genetics, the Wales Gene Park organises courses, seminars and workshops aimed at enhancing the understanding of genetics throughout Wales for students, teachers and healthcare professionals.

This is an excellent example of how to raise awareness of genetics and genetic conditions, and could be expanded on or used as a model for a similar initiative for rare diseases.

Outreach and education: an example of good practice

The **multidisciplinary Rett syndrome clinic** held at St David's Hospital in Cardiff is an example of good practice in how outreach and staff exchanges can facilitate exchange of information and expertise whilst providing high-quality care. The whole team has gone to other hospitals outside of Wales including in Worcestershire and Devon, to hold clinics. Individual team members have also visited Scotland to assist in education of staff there. Staff from other hospitals have been able to visit the clinics held in Cardiff and this has resulted in St. Mary's Hospital in Manchester establishing its own service which will commence in March 2011.

Patient centred care: an example of good practice

Immunodeficiency patients often require lifelong immunoglobulin replacement therapy to remain healthy. This treatment is provided either intravenously every 2 to 4 weeks or subcutaneously weekly. Previously this has meant patients frequently having to attend hospital appointments for their treatment; but the **University Hospital of Wales (UHW)** is now able to provide most patients with the training and equipment to administer their own treatment at home.

Patients eligible for home therapy first receive training in how to self-deliver the immunoglobulin therapy, and prior to their first home infusion they must pass a competency assessment. Once they have done so, the immunoglobulins and ancillary equipment, such as sterile needles, pumps etc required for home therapy are delivered as part of a home care package and they are then able to administer their own treatment. Outreach is also an important part of this service and patients receive home visits from nurses, and monitoring is also possible from home with blood samples being sent to the central immunology laboratory at UHW to monitor immunoglobulin levels.

This home therapy service enables patients to have much more flexibility around when they receive their treatment, and so reduces disruption to daily life and enables them to maintain regular attendance at work or school. It also reduces exposure to hospital-acquired infections, and reduces the inconvenience and cost of having to travel to frequent hospital appointments. Home therapy also has benefits to the NHS, such as increasing the number of available hospital day beds and a reduction in the time specialist nurses have to spend administering immunoglobulin therapy.

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