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Llywodraeth Cymru
Welsh Government

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Consultation Document

The Welsh Implementation Plan for Rare Diseases

Date of issue: **28 February 2014**
Action required: Responses by **23 May 2014**

Overview

The Welsh Government is wishing to consult about its response to the commitments set out in United Kingdom Strategy for Rare Diseases.

The Strategy, agreed by the governments of the four countries of the United Kingdom, set out 51 commitments to improve information, services and research in relation to people with rare diseases.

We would be grateful for your views to help us finalise the Plan.

We believe that the Plan needs to respond clearly to all the commitments but remain flexible and adapt to changing needs and possibilities.

The following questions may help in assessing the proposals in the Plan, but are not meant to limit your comments.

Q1 Do you agree that the actions set out relating to empowering those with rare diseases will mean that Wales will meet its commitments?

Q2 Do you agree that the actions set out relating to identifying and preventing rare diseases will mean that Wales will meet its commitments?

Q3 Do you agree that the actions set out relating to diagnosis and early intervention will mean that Wales will meet its commitments?

Q4 Do you agree that the actions set out relating to the co-ordination of care will mean that Wales will meet its commitments?

Q5 Do you agree that the actions set out relating to the role of research will mean that Wales will meet its commitments?

Q6 Do you have any other comments on the draft Plan?

How to respond

If you wish to email us send your response to OCMOMailbox@wales.gsi.gov.uk indicating that it is a response to this consultation. If you wish to send a hard copy please address it to:

Rare Diseases Consultation
OCMO
Department for Health and Social Services
Welsh Government
Cathays Park
Cardiff CF10 3NQ

Further information and related documents

Large print, Braille and alternate language versions of this document are available on request.

Data protection

How the views and information you give us will be used

Any response you send us will be seen in full by Welsh Government staff dealing with the issues which this consultation is about. It may also be seen by other Welsh Government staff to help them plan future consultations.

The Welsh Government intends to publish a summary of the responses to this document. We may also publish responses in full. Normally, the name and address (or part of the address) of the person or organisation who sent the response are published with the response. This helps to show that the consultation was carried out properly. If you do not want your name or address published, please tell us this in writing when you send your response. We will then blank them out.

Names or addresses we blank out might still get published later, though we do not think this would happen very often. The Freedom of Information Act 2000 and the Environmental Information Regulations 2004 allow the public to ask to see information held by many public bodies, including the Welsh Government. This includes information which has not been published. However, the law also allows us to withhold information in some circumstances. If anyone asks to see information we have withheld, we will have to decide whether to release it or not. If someone has asked for their name and address not to be published, that is an important fact we would take into account. However, there might sometimes be important reasons why we would have to reveal someone's name and address, even though they have asked for them not to be published. We would get in touch with the person and ask their views before we finally decided to reveal the information.

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Mark Drakeford AM, Minister for Health and Social Services

Ministerial Foreword

I am pleased to be able to issue this draft Implementation Plan as the initial Welsh response to the Strategy for Rare Diseases which I launched with my counterparts in England, Scotland and Northern Ireland in October last year. We indicated then our strong commitment to act together to improve services to those who live with rare diseases and to issue an Implementation Plan for each of the four countries.

By their very nature rare diseases offer specific challenges to the NHS. There are many such diseases and in any given area there may be very few people affected by any one disease. To achieve the best results we need to support the families concerned by engaging and co-ordinating the contribution of primary care, available everywhere, and various specialised services, which may operate only at regional, national or even international level. Success in supporting those affected will depend on the NHS but also on others including wider local services, the third sector, industry and the research community. In terms of governmental responsibilities, some rest with the Welsh Government, others remain at UK level and there is also European Union-wide interest in ensuring services are available. We need in our response to recognise that complex reality.

Accordingly, this draft Implementation Plan has been prepared by a Task Group which has brought together the NHS in Wales, the Welsh Government, the third sector and the research community and close links have been maintained with government officials and other interests across the UK.

Because of the complexity of the issues surrounding rare diseases and the fast changing science that will help determine how cases are managed, the four Ministers have agreed to the establishment of a UK Rare Diseases Advisory Group which will maintain a continuing overview of the wider issues relating to the management of this agenda across the four countries. In addition a UK-wide Stakeholder Forum will monitor and report on progress in the four countries. Within Wales I am establishing an Implementation Group to oversee the implementation process and keep the Plan fresh and relevant and liaise with the NHS and other interests, taking advantage of developments in Wales and elsewhere in the UK.

To ensure that we have successfully drawn on the range of views in Wales, I have agreed to issue the Plan for a 12 weeks consultation. I welcome your views on this Plan.

Foreword from David Sissling, Chief Executive of NHS Wales

It is vital that Local Health Boards and Trusts deliver safe, sustainable, high quality care to those who have rare diseases. This will be challenging particularly as there are so many and such varied conditions that fall under this heading. Success will depend on co-ordinating effective action in many areas. We will focus on supporting patients and their families and carers. We will ensure efficient detection and rapid diagnosis and we will mobilise high quality treatment supported by excellent patient information. There will be an emphasis on partnership working – across public

services and the third sector and with individuals – to promote successful management of these conditions and support self care. This Implementation Plan will direct our work. It clarifies the main areas of work and signals our intention to start monitoring key outcomes. The Welsh Government will work closely with the Welsh NHS, providing support as necessary. The expectation now is to see rapid, sustained improvement.

1. Introduction

A rare disease is a life-threatening or chronically debilitating disease that affects five people or fewer in 10,000. There are between 5,000 and 8,000 rare diseases and, while each one affects relatively few people, together they affect the lives of three million people across the UK, which would imply some 150,000 people affected in Wales.

This Rare Diseases Implementation Plan provides a framework for action by Local Health Boards (LHBs), NHS Trusts and their partners in local government, the third sector, the education and research communities and industry. It sets out the Welsh Government's expectations of the NHS in Wales to tackle rare diseases for people of all ages, wherever they live in Wales and whatever their circumstances. The Plan is designed to enable the NHS to deliver on its responsibility to meet the needs of people at risk of, or affected by, such conditions.

If we are to sustain, and further develop, high quality healthcare for the people of Wales, there needs to be true partnership, with the NHS supporting people to manage their own condition.

2. Strategic Context

The Welsh Government's Programme for Government and its five year NHS Plan, *Together for Health,* introduced an ambitious programme for health in Wales so that:

- health will be better for everyone
- access to care and patient experience will be better
- better service, safety and quality will improve health outcomes.

Achieving Excellence: The Quality Delivery Plan for the NHS in Wales for 2012-16 describes how we aim to ensure consistent excellence in services. It outlines actions for quality assurance and improvement. We commit to a quality-driven NHS that provides services that are safe, effective, accessible and affordable, and that offer an excellent user experience.

These ambitions should apply no less to rare diseases.

3. Our Vision

The Welsh Government supports the Vision in the UK Strategy for Rare Diseases. All four countries committed themselves to:

- promote equity of access – allowing everyone with a rare disease to follow a clear, well defined care pathway, providing high quality services for every individual through integrated personal care plans
- offer a patient centred, coordinated approach to treatment services, specialist healthcare and social care support which takes into account the needs of patients, their families and others who provide essential support
- deliver evidence-based diagnosis and treatment of rare diseases, developed through the best use of regional and national resources that are easily accessible by patients and professionals

- support specialised clinical centres to provide expert, high quality clinical care and expertise to patients their families and carers and the patient's, multi-professional healthcare team
- promote excellence in research and develop our understanding of and treatments for rare diseases
- deliver rapid and effective translations of advances in the understanding of rare diseases into clinical care by creating appropriate infrastructure, care pathways and clinical competences
- deliver effective interventions and support to patients and families quickly, equitably and sustainably
- promote collaborative working between the NHS, research communities, academia and industry wherever possible to facilitate better understanding about rare diseases and how they can be best treated
- support education and training programmes that enable health and social care professionals to better identify rare diseases to help deliver faster diagnosis and access to treatment pathways for patients
- promote the UK as a first choice location for research into rare diseases as a leader, partner and collaborator

4. Why is This a Priority?

There are good reasons for improving services in relation to rare diseases. While any one disease may affect relatively few people, collectively the diseases affect many people and their families. Discussion on agreeing the UK Strategy for Rare Diseases identified many ways in which services might be improved to give both families and professionals greater support and clarity in managing these problems.

5. Our Journey So Far

The Strategy agreed by the four countries aims to ensure that all those affected by a rare disease will have the best quality of evidence-based care and treatment that our health and social care systems can provide. This will be achieved by the health departments working together with patients and families, patient organisations, the NHS, researchers, industry and all other stakeholders. The Strategy includes specific commitments that each country has undertaken to fulfil.

In responding to the Strategy, each of the four countries will need to take a slightly different approach, reflecting local resources, priorities and practices. Accordingly each is publishing its own Implementation Plan setting out its own way forward. These will build on current services, drawing on best practice and aiming to achieve the most effective use of resources, with an aim to improve access to information, services and research and to bring real, positive change to the lives of people with rare diseases and their families.

6. Aim of the Implementation Plan

This Plan has high level actions that need to be delivered by all LHBs and by other agencies. These actions have been developed in relation to the Commitments set out in the UK Strategy.

Other delivery plans issued by the Welsh Government have specified the population and service outcome measures to drive forward improvements in care for the people of Wales. Such measures are not available at this stage for this Plan. Initially progress will be judged in relation to the specific actions included in the Plan. Over time, working with the other countries of the UK and linking into work being undertaken at a European level, we will aim

to agree a range of specific indicators to judge success, including outcomes. We will aim to have the first set in place by March 2015.

7. What Do We Want to Achieve?

Action will be taken in five areas identified in the Strategy against the 51 Commitments it contains. The annex to this Plan identifies where in this document to find the Welsh responses to the 51 commitments. The five areas are:

Theme 1 Empowering those affected by rare diseases

Theme 2 Identifying and preventing rare diseases

Theme 3 Diagnosis and early intervention

Theme 4 Co-ordination of care

Theme 5 The role of research

8. Working Together

This Welsh Plan identifies the tasks that will fall to the Welsh Government, the LHBs and other bodies. All of us have a key part to play in our efforts to tackle rare diseases. Together the actions specified here will establish a toolkit that will enable all concerned to improve services.

The **Welsh Government** is responsible for strategic leadership through setting the health outcomes it expects for the people of Wales and holding the NHS to account. The lines of accountability are via the Chairs of the LHBs and NHS Trusts to the Minister for Health and Social Services, with the Chief Executives of the LHBs and NHS Trusts reporting to the Chief Executive of NHS Wales, who is also the Director General of the Welsh Government's Department of Health Social Services.

The Welsh Government will review progress against the Plan.

NHS Wales is made up of seven LHBs and 3 NHS Trusts. LHBs are responsible for planning, securing and delivering local services to help prevent rare diseases where possible and to diagnose, treat and care for people affected by rare diseases. Together through the Welsh Health Specialised Services Committee (WHSSC) they plan and fund many of the additional specialist services which may be required to support people living with or at risk of a rare disease.

To plan services effectively for their populations LHBs must build and lead coalitions with NHS Trusts, GPs, pharmacists, opticians, dentists, local government, the third sector voluntary bodies and the public. These services need to be part of an integrated rare diseases service.

Among the important actors in Wales are the All Wales Medical Genetics Service (AWMGS); Public Health Wales which provides LHBs with information and advice to inform service planning and is responsible for screening services; the National Institute for Social Care and Health Research (NISCHR) which leads on promoting research on behalf of the Welsh Government; and the universities which undertake research. Wales is represented on the UK Rare Diseases Advisory Group which addresses among other issues the commissioning of highly specialised services. Wales shares representation with England on the European Union Committee for Rare Diseases.

Local government also has a vital role to play. To promote a co-ordinated approach, they need to work with LHBs through Local Service Boards. This work includes the analysis of the evidence base and development of Single Integrated Plans showing how they can contribute to improving health outcomes and supporting people with rare diseases.

The **Welsh Implementation Group for Rare Diseases** will oversee the implementation process and keep the Plan fresh and relevant. It will liaise with the NHS and other interests, taking advantage of developments in Wales and elsewhere in the UK.

The **third sector** has an important role to play, both in providing services and acting as the voice of individuals.

People with rare diseases can help manage their own conditions and help improve services. A range of opportunities for doing so are mentioned in the Plan.

Because of the complexity of the issues surrounding rare diseases and the fast changing science that will help determine how cases are managed, the four Ministers have agreed to the establishment of a **UK Rare Diseases Advisory Group** which will maintain a continuing overview of how best to deal with the wider issues relating to the management of this agenda across the four countries.

In addition a **UK-wide Stakeholder Forum** will monitor and report on progress in the four countries in taking forward actions in the Implementation Plans and meeting the commitments in the Strategy.

9. Measuring Success

The Quality Delivery Plan sets out how we will monitor performance and progress in improving health and healthcare in Wales. It places requirements on NHS organisations to monitor a set of nationally specified performance measures and report them to the public, the Welsh Government, and their Boards at regular intervals. This Implementation Plan now places a requirement on LHBs to report on this area, as described below, and signals our intention to develop service indicators that can be used in future.

10. Local Plans – Local Action

The main planning task for LHBs will be to review their current arrangements for providing information and services to people with rare diseases and use the findings as the basis for their first annual report on rare diseases, to be issued by 31/03/15 and annually thereafter. This should also address LHB links with the third sector and the research community (see actions on commitments 1-3 and 47).

Theme 1 Empowering those affected by Rare Diseases

The UK Rare Disease Strategy aims to ensure improvements across the whole ‘patient journey’, from the patients’ first contact with the GP through to diagnosis, support and management of a rare condition.

The commitments in the Strategy emphasise the fundamental role that the patient, supported by their family/carer and/or patient organisation will play during this journey. Patients and their families are often those with most knowledge about their condition, its management and the services they require. They are often supported by a wide range of groups; these can be global networks and patient organisations working at a national level as well as those based locally in Wales.

The third sector and particularly patient organisations can help provide the knowledge and guidance patients may need to secure access to health and social care services to manage and improve their condition. Many organisations provide information and support through websites, leaflets and helplines as well as being able to act as the patient's advocate when needed. Patient organisations can bridge the gap between patients, their families and the health service.

While mechanisms for patient involvement in service provision and research have been developed and piloted in some areas, these need to be strengthened in relation to rare diseases, and formally extended to include consideration of their non-medical needs. Currently there are no formal mechanism for patients and families to feed into the planning of such services but tools exist for evaluation and monitoring via patient experience surveys.

Existing guidance specifies that for older people with complex needs, local authorities and LHBs work together to ensure an assessment is co-ordinated by a named lead professional. This will determine the need for a care and support plan. Where a plan is required, it should make clear who the care and support co-ordinator is, who has specific responsibility for completing and monitoring the plan. These arrangements should apply equally in the case of older people with a rare disease, and good practice suggests that they should apply more broadly in all cases where those with rare disease face complex needs.

The involvement of patients, families and patient groups in the planning and delivery of service provision will ensure that patients have a voice during the early stages of service development and on through to the implementation, delivery and continued monitoring of services.

The Welsh Government will continue to work closely with patient organisations, including Genetic Alliance UK and Rare Disease UK to ensure that patients receive adequate information and support.

Issues relating to patients' engagement with training are addressed under theme 3 and with research under theme 5.

Commitments in the UK Strategy on empowering those affected by Rare Diseases	**Actions to be taken forward for Wales**
1. Strengthen the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research, recognising patient groups as key partners.	• WHSSC will establish a clinical evidence group for rare diseases to review evidence for these conditions by 01/03/14, its membership to include patient organisation representatives. • LHBs will review their current arrangements for working with the third sector organisations supporting people with rare diseases and for putting people in contact with them by 31/03/15. • As part of these they will ensure that the NHS clinical team leading on the care of each rare disease involves patients and their carers in the provision of the service.

2. Improve awareness amongst service providers and others of the effects that rare diseases can have on a person's education, family, social relationships and ability to work.	• As part of its review (see 1 above) each LHB should ensure that the NHS clinical team leading on the care of each rare disease develops information so that patients will receive adequate and timely information regarding their condition and supporting organisations. WHSSC will ensure that this is a required standard within its service specification.
3. Encourage effective and timely liaison between the NHS and other public service providers, and encourage providers to consider the effects of rare diseases on people's lives when they are developing and managing services.	• LHBs will review their current arrangements for providing information and diagnostic and treatment services to people with rare diseases and use the findings as the basis for their first annual report on rare diseases, to be issued by 31/03/15 and annually thereafter.
4. Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.	• WHSSC will include a requirement in the service specification and contract for the NHS clinical team to engage with patients using patient experience surveys and patient reported outcome measures. • WHSSC will engage with all stakeholders including patient organisations in the development of commissioning policies which provide detail on the referral pathway, clinical indications for the procedure or treatment, and access criteria for the service.
5. Consider how to give all patients with a rare disease clear and timely information about their condition and its development; treatment and therapy options; practical support.	• See action 2 above.
6. Improve access for patients (or where appropriate their parents or guardians) to their personal data.	• LHBs will review their current arrangements for preparing assessments and care plans for people with rare diseases by 01/12/14
34. Make better use of online applications to give patients information about their condition so that they can develop a personalised care path plan with their clinical and social care team.	• WHSSC will include information within service specifications on the model of care including the referral pathway, clinical access criteria, and treatment options, which will be available for patients to access via the WHSSC website with an initial group agreed by 01/12/14 (see also the section on evaluation below).

Theme 2 Identifying and Preventing Rare Diseases

The UK Strategy noted that many rare diseases are present at birth and are either caused by a genetic problem (for example cystic fibrosis) or deficiencies or exposures to substances around the time of conception or during pregnancy. In this context it considered the role of screening and testing.

In fact the majority of cases of rare diseases are not identified as part of a population-based screening programme. In future, there will in future be greater efforts to avoid the emergence of signs and symptoms through preventive therapies aimed at those who are genetically at very high risk. These issues are discussed further later in this Plan. While the role of screening and carrier testing is limited, it is important that Wales has well developed systems for providing such services.

The All Wales Medical Genetics Service has a central role in the ongoing carrier testing and counselling of immediate family and relatives following the identification of rare inherited diseases. The provision of accurate information and carrier testing to families ensures that they are able to access ongoing management and make well informed decisions about future reproductive decisions. The counselling provides accurate information about risk and informs on the availability of prenatal testing options as well as information about pre implantation testing if appropriate.

Case study - Screening

Things have moved on quite significantly since I was diagnosed with cystic fibrosis in the 1980s. I was picked up through a pilot programme in Wales for newborn screening. Screening was randomised on a week on/week off basis.

Only since 2007 has there been universal screening for cystic fibrosis in the whole of the UK. Newborn bloodspot screening is offered for all babies born in Wales at day five to eight days of life for screening for a number of rare conditions. Diagnosing these rare conditions early can significantly alter the outcome for those affected by these rare disorders.

I believe that for me early intervention has enabled me to live as normal a life as possible; I've been able to attend school, university and play an active part in society and work. I have had time in hospital throughout the years and things haven't always been easy but I feel that being picked up early, effective management and starting early interventions gave me the best possible start I could have.

If I had been picked up later in life, which many people with cystic fibrosis had been previously, my lungs and digestive system, would be less healthy and I would have a significantly worse quality of life and decreased life expectancy.

Note this and the following case studies have been based on real patient experiences but constructed to protect anonymity.

Public Health Wales and the Welsh Government are both represented on the UK National Screening Committee (NSC) and Wales has representatives on its subgroups across the screening programmes. The Wales Screening Committee takes advice from the NSC and the Welsh Government takes the decisions on implementing screening policy in Wales.

The Newborn Bloodspot Screening Programme is the most relevant to the Strategy for Rare Diseases. It involves taking a small sample of blood from the baby's heel ideally on day five of life. The sample is screened for rare but serious diseases that respond to early intervention to reduce mortality and/or morbidity.

In Wales, the conditions currently screened for are congenital hypothyroidism (CHT), cystic fibrosis (CF), medium chain acyl-CoA dehydrogenase deficiency (MCADD), phenylketonuria (PKU) and sickle cell disorders (SCD). MCADD screening was implemented in June 2012 and SCD screening in June 2013 as part of a project to implement a safe and sustainable newborn bloodspot screening programme across Wales. The project has developed and disseminated parent information leaflets, developed a website, supplied prepaid envelopes, developed training materials for health professionals and undertaken checks to identify babies (less that one year) who did not have a newborn bloodspot screening result on the child health system.
As part of implementation of the newborn bloodspot screening programmes for these disorders, pathways for babies who are suspected to have the condition have been agreed with lead clinicians in Wales. Work is ongoing to ensure all of the pathways are described and that these are equitable across Wales.

Preparation needs to be undertaken to enable implementation of a possible expansion of the bloodspot screening, for metabolic disorders, dependent on a recommendation made by the UKNSC and Wales Screening Committee

Commitments in the UK Strategy on identifying and preventing Rare Diseases	Actions to be taken forward for Wales
9. Continue to work with the UK National Screening Committee to ensure that the potential role of screening in achieving earlier diagnosis is appropriately considered in the assessment of all potential new national screening programmes and proposed extensions to existing programmes.	• Current links will be maintained. • The Welsh Screening Committee will ensure that stakeholders in Wales are aware of current UK NSC consultations via networks and websites.

Theme 3 Diagnosis and Early Intervention

Introduction

Early and accurate diagnosis of rare diseases is a prerequisite for the best care and for early intervention. Diagnosis of rare diseases often requires recognition of the possibility of a rare disease as a cause for common symptoms and signs and therefore poses a challenge in non-specialist care.

In a small number of scenarios diagnosis of a rare disease may be as a result of screening, as described above, for example following new born and antenatal screening.

Early diagnosis and early intervention may also follow testing because of family history of a rare disease, where no symptoms of disease have yet appeared. Increasingly, early diagnosis and early intervention offers the prospect of prevention of disease through prenatal or, in the case of IVF, preimplantation diagnosis and the prevention of disease complications through increased surveillance, prophylactic procedures, chemoprevention or targeted therapy.

A relatively new discipline in this field is genomics, which involves studying the genome – the whole genetic structure of an individual. New genomic technologies are expected to change how diagnosis of rare diseases is undertaken. The traditional approach to sequencing people's genetic structure has focused on finding one or a few genes that may help understand what the problem is. The new approach is more comprehensive and efficient, using sequencing of the whole genome or part of it (an 'exome') to try to explain an unrecognised pattern of signs and symptoms and support direct genetic diagnosis of a specific (rare) disease. Wales aspires to be at the forefront of this and is committed to developing a proposal for an infrastructure for genomic medicine to meet the clinical, research and economic development needs of Wales.

Some patients may for a long period lack a clear diagnosis or indeed never receive one. It is important that in cases where a patient clearly has an unusual health problem but no diagnosis that he or she receives appropriate support and care and this Plan aims to ensure that happens.

Case study - Management without diagnosis

Patrick came to my Community Paediatric clinic on many occasions during his childhood. I first saw him as a 10-month old baby, following referral by the Health Visitor as he was not meeting his developmental milestones. He was a lovely baby, very smiley, but not sitting up, and was quite 'floppy' to handle. There was no clue in the pregnancy or birth history as to why he was behind in his development and nothing on examination except his general low tone. His parents and I decided to go ahead with blood tests for common conditions such as thyroid problem, Down's syndrome and muscular dystrophy, but these were all normal. Over the next 8 years, I saw Patrick from time to time. He was slow in talking so we checked his hearing and his vision too, and had speech therapy advice. He had walking and co-ordination problems and was helped by exercises and appliances from Physiotherapy and Occupational Therapy. I provided advice to the education department when he was two so they could plan the right supports for him in school.

I asked for a specialised opinion and the geneticists to see him because he seemed to have muscle weakness as he got older, but they were never able to make a diagnosis of anything. Patrick was a little boy who had slow development, walking and speech, and found learning difficult. At the age of eight he loved to play on his skateboard with his friends and they helped him if he fell over. He enjoyed school with some extra help for concentration in class. He still doesn't have any specific diagnosis for his difficulties. His parents still hope to find out why he has problems.

Patrick is not the only patient in my clinic without a diagnosis...

This chapter will look at:

- the interlocking roles of primary, secondary and specialised care and genetic diagnosis in developing pathways to diagnosis
- evaluation of new treatments
- education and support for professionals.

Developing pathways

Case study – a patient journey

I had always walked with a trademark 'wobble' and spoken with a slight slur but this was just part of who I was. I had a career running my own company. One day when I was making sandwiches for work, I found that buttering bread seemed to be taking more and more concentration. I went to my GP, who on numerous occasions simply told me to take time off work and rest. This went on for many years and involved many visits trying to convince GPs that something was definitely wrong.

My condition continued to progress and I had problems with movement, balance and speech. It was only when during one visit I was seen by a locum doctor who had recent knowledge of similar symptoms that I was sent for neurological tests. He also referred me to a general neurologist where I was diagnosed with cerebellar ataxia in my mid-30s. I was told the diagnosis, the probable outcome, that there was nothing that could be done, and 'that was that!'

When I returned to my GP, he said I had a rare condition and that I was unlikely to meet many others with this rare disease. I am determined that information should be made available to patients with all rare diseases and that people realise that support networks are vital so that feelings of isolation do not add to the patients worries. I have received great support and been involved with the charity. Global online support networks for people with rare conditions enable patients and their families to connect with others from across the world who have the same condition, providing an opportunity to share invaluable advice and support.

a. Primary Care Services

Whilst many rare diseases will be identified at or before birth through specialist services, there are particular challenges in identifying and managing rare diseases that first appear later in the context of community services.

A GP will have very limited experience of rare diseases and it is clearly impossible for an individual GP to establish an expertise for each condition. However, since it is estimated that one in 17 people will suffer from a rare disease in the course of their lifetime, it is possible for primary care teams to develop a generic expertise in the management of cases where it appears difficult to reach a diagnosis and where a rare disease might be suspected.

Where there is uncertainty, patients and their carers should be reassured that their concerns are recognised, and a shared understanding of the diagnostic difficulties and appropriate actions should be developed. In many cases GPs will have a long relationship of care for a patient or their family and this may provide particular support at times of uncertainty.

Practice teams should develop their expertise in the management of rare diseases, referencing the UK Strategy and this Plan, and learning from individual case management and the findings of significant event analyses when there have been concerns.

GPs provide continuity of care, which may be particularly important at times of transition between specialist services. GPs will also recognise the potential for conflicting advice when many services are engaged in the delivery of care. Patients should be supported to clarify the advice and make choices that meet their needs and personal preferences.

Primary care teams should also identify and support the needs of carers and should ensure that access to social care advice is provided where appropriate.

b. Secondary Care Services

Secondary care services are an important bridge between the primary care team and the specialist service and their role can include:

- initial consideration of and further investigation in secondary care of a rare disease
- referral for further opinion to specialised services
- ongoing liaison with the specialised services
- provision of routine and emergency care local to the patients home
- access to information and support for patients affected by rare disease and their families.

Each hospital clinician, just as a GP, may have limited experience of a rare disease but unlike primary care will have access to inpatient/outpatient facilities to manage the condition. While it may be possible to develop an expertise in the management of certain rare diseases, patients and their carers need to be reassured that their concerns are recognised appropriately and that there is a clear dialogue between secondary care and specialised services as required. This should include the role of secondary care, in partnership with specialised services, agreeing individual care pathways for patients so they may move between primary, secondary and specialised care appropriately and without delay, receiving treatment in a timely manner. This may include signposting to relevant electronic/web-based systems and urgent contact telephone numbers for patients. It should include the identification of the most appropriate lead clinician for that patient in secondary care to act as an advocate for that patient and co-ordinate shared care.

It may be necessary in some instances for the relationship to be direct contact between specialised services and the patient, but there still needs to be agreement as to the roles of

primary care and secondary care in patient care and advice. Secondary care, like primary care, needs to be aware of the national strategy and the challenges faced by patients with rare diseases.

LHBs will ensure that the role of clinicians in managing rare diseases and of geneticists in education of health professionals, setting standards for healthcare delivery of patients in secondary and primary care settings and developing pathways of care for appropriate use of genetic resources is appropriately reflected in job plans.

c. Specialised Services

WHSSC aims to provide equitable access to safe, effective and sustainable specialised services for the people of Wales, as close to patients' homes as possible, within available resources. WHSSC has contracts in place with specialised service providers that cover the more common rare diseases. While there is provision of specialised services for rare diseases by LHBs within Wales, patients with rare diseases may be referred to specialist providers in England for their care.

Where there are no contracts in place for providers in England or further afield, in order for a patient to access a service, referrers are required to submit clinical information for consideration by the All-Wales Individual Patient Funding Request panel.

WHSSC has established clinical leads in a number of specialities. These leads act as clinical gatekeepers and can authorise referrals to centres in England. Information on the clinical lead and speciality is published on the WHSSC website in a referral management directory accessible here: http://www.wales.nhs.uk/sites3/page.cfm?orgid=898&pid=64292

Feedback from clinicians and patients indicates that at times there can be delays and difficulties in access to specialised services for rare diseases. This is particularly the case where WHSSC does not hold a contract with the specialised service providers. WHSSC is committed to reviewing referral pathways and providing clearer guidance and information on how to access services between primary care, secondary care and regional centres and onto the preferred specialist clinical centres. WHSSC will build on the existing published referral management directory and continue to identify lead clinicians for specific conditions to support with the referral pathway.

WHSSC will continue to work closely with providers in Wales and England to ensure access to services is equitable and the provision of service is of the highest quality. At times when specialised services are only delivered in England WHSSC will work with NHS England and LHBs to identify a lead clinician in Wales to support with the sharing of knowledge, experience, education and research between the four countries.

d. Genetic Diagnosis

The All Wales Medical Genetics Service (AWMGS) provides clinical and genetic testing services for patients across Wales. Genetic testing is a major development area in diagnosing and treating rare diseases. Many genetic tests are provided by the Cardiff laboratory at the Institute of Medical Genetics, while tests for other diseases are accessed via the UK Genetic Testing Network (UKGTN, of which the AWMGS is a member laboratory) or internationally. As an accredited provider, AWMGS delivers some specialist tests to the UK and worldwide, as part of a growing network of laboratories and also delivers international specialist services for some rare disorders linked to the recognised international expertise of clinicians in Wales. The service is also a listed provider with Orphanet, the international internet portal for rare diseases and orphan drugs.

AWMGS also purchases genetic tests for some rare disorders via the UKGTN and from providers internationally. Each year, as knowledge of genetic disorders increases, new

genetic tests are commissioned by UKGTN. These new genetic tests then become available to patients and their families affected by, or at risk of, the associated rare diseases. WHSSC is committed to developing a robust and timely process which takes into account the rapid rate of development of new diagnostic tests, resourcing local genetic and UKGTN services, and providing equity of access for Welsh patients. Next Generation Sequencing (NGS) is discussed in the next section.

Commitments in the UK Strategy on diagnostic pathways for Rare Diseases	**Actions to be taken forward for Wales**
10. Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular tests are evaluated and recommended by UKGTN, is accessible for at risk relatives.	• AWMGS, in conjunction with WHSSC will by 30/09/14: - complete a review of the resources available for local genetic and UKGTN services, and of equity of access - devise a system for reviewing and approving any new tests and agreeing funding systems for any additions - report on these to the Minister for Health and Social Services.
11. Work to achieve reduced times for diagnosis of rare diseases, whilst acknowledging that more needs to be done to ensure that undiagnosed patients have appropriate access to coordinated care e.g. to help disabled children who are thought to have a genetic syndrome or condition that science has not yet identified.	• AWMGS and the LHBs in conjunction with WHSSC will formalise and agree care and referral pathways across Wales for access to appropriate expertise and highly specialised services, with an agreed plan by 31/03/15. • See also action 10 above.
12. Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by professionals and patients, by: – establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and on to specialist clinical centres, as appropriate – putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment – drawing on patients' ability to help inform decisions about referral and diagnosis – creating effective clinical networks to support this process – making high quality diagnostic tests accessible through common, clinically agreed systems or pathways – embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies.	• AWMGS and the LHBs in conjunction with WHSSC will agree by 30/09/14 a plan to: - formalise and agree care and referral pathways for the Welsh population for access to specialised expertise and services - establish a system of monitoring Welsh genetic testing activity - develop a clear commissioning model for Welsh genetics services - monitoring and evaluate access to testing. • LHBs will work with GPs to: - develop agreed local care pathways - help them use significant event analysis to identify diagnostic delays and service barriers to identify and address any learning needs in relation to unusual diagnoses in their personal development plans and share learning across practice teams - ensure that the individual's care plan considers all aspects of health and social care needs, including access to benefits advice where appropriate, and in the case of children transition arrangements - develop services to ensure that consultation times reflect need and allow sufficient time to address complex care management - seek patient and carer feedback to inform service improvement. • The Wales Implementation Group will by 30/09/14 review arrangements for identifying and supporting patients for whom no diagnosis can be agreed and agree future

	actions.

Evaluation of New Tests and Treatments

WHSSC has developed an evidence-based appraisal process and has started to review the costs and benefits of high cost treatments and specialities. The products of this process have been reviewed by clinicians and where relevant patient support groups. The products have identified clear clinical access criteria for services as well as the referral pathway and outcome measures. This information is being used to develop condition specific commissioning policies. WHSSC will continue to use this process to review the costs and benefits of high cost intervention to inform clear clinical access criteria for services as well as referral pathways and outcome measures.

WHSSC is a member of the All Wales Medicines Strategy Group (AWMSG), the Orphan and Ultra Orphan drugs meeting, and the Rare Diseases Advisory Group. WHSSC works closely with the other three nations and also with international partners including the European Union and Committee of Experts in Rare Diseases in relation to rare diseases and the provision of services.

The UK Strategy stresses the importance of the opportunities for improving genetic testing through the development of Next Generation Sequencing (NGS). The Welsh Government has funded the purchase of capital equipment to implement NGS and the challenge going forward is the development and validation of diagnostic protocols and specialist bio-informatic analysis, to enable Wales to keep pace with regional genetics services elsewhere in the UK and allow the AWMGS to be a full partner in the UKGTN as a provider and purchaser of services for the patients of Wales. Even more important, that would allow Welsh clinicians to access appropriate services and laboratory testing that will benefit patients and their families.

Commitment in the UK Strategy on evaluation and new treatments	**Actions to be taken forward for Wales**
13. Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.	• The AWMSG will continue to appraise new medicines and develop their processes to do so. • WHSSC will use its evidence-based appraisal process for reviewing treatments and its membership of technical groups to ensure appropriate evaluation of new genetic and genomic tests and technologies ongoing from 31/03/14. • The Welsh Government will agree by 31/03/14 future arrangements for the approval and managed entry of orphan and ultra-orphan medicines. • Clinicians in collaboration with WHSSC will work to ensure that there are clearly defined and appropriate outcome measures in service specifications and formal processes for evaluation of services to understand the costs and benefits of treatments, including patient and public involvement in the process of evaluation ongoing from 31/03/14.
42. Begin and complete next generation sequencing (NGS) demonstration projects to: evaluate their usefulness, acceptability and cost-effectiveness; develop effective health economic assessments (for example through Health Technology Assessments)	• The Welsh Government will work with AWMGS to review the options to accelerate progress and ensure the best value from recent capital investment by 30/09/14.

and similar initiatives.	
43. Evaluate different NGS platform configurations, for example: NGS for clinical condition-specific sets of genes (such as 100–200 of the 22,000 genes), whole exome sequencing (2% of the entire genome), whole genome sequencing	• See action 42 above.
44. Support the introduction of NGS into mainstream NHS diagnostic pathways, underpinned by appropriate clinical bioinformatics, including clinical bioinformatics hubs supported by high performance computing centres where appropriate	• The Welsh Government will continue to work with Cardiff & Vale UHB (hosts of the AWMGS), Cardiff University and other strategic partners (including Public Health Wales and Swansea University and industry partners) to develop a proposal for development of infrastructure for genomic medicine for action by 30/09/14.

Education and Support for Professionals

There is a lack of awareness and identification of rare diseases amongst healthcare professionals, often resulting in a delayed diagnosis or misdiagnosis of rare disease patients. Education in rare diseases should help all health professionals:

- be aware that common presentations may include a small number of patients with a rare disease
- understand that the pattern and combination of signs/symptoms across the family rather than in just a single individual may suggest a specific rare inherited disease diagnosis
- understand the different modes of inheritance
- be able to take and record an accurate family history of disease
- be aware of clinical genetics services and other services for rare diseases and how these can be accessed
- be aware of sources of reliable and “just in time” information on rare diseases, such as the online resource Orphanet
- be aware of the importance of access to research for those with rare diseases and where information on research opportunities can be accessed (e.g. online via NISCHR and NIHR portfolios).

Additionally, medical undergraduates and specialist clinicians need to be prepared for the clinical application of sequencing technologies that are expected to change the approach to diagnosis in rare disease. AWMGS will have a significant role in the education of health professionals in the information and communication needs of patients in relation to delivery of NGS testing and results. In the future this will not be limited to Clinical Genetics, but will extend to how genomics is linked to clinical practice in other specialties such as paediatrics, cardiology and oncology.

Continuing medical education across the NHS in Wales should also help those providing services for rare diseases through:

- developing and providing education in bioinformatics for laboratory and clinical specialists in rare diseases
- giving laboratory specialists in rare diseases opportunities for education in next generation sequencing technologies
- developing clinical training opportunities in genomic medicine.

In Wales these requirements could be met through:

- engagement of specialists in rare diseases with those developing and delivering undergraduate and taught post-graduate curricula in medical and health schools in Wales
- working with lay organisations and charities representing those affected by rare diseases
- extending the professional education programme of the Wales Gene Park
- inclusion of development and delivery of education and, where required, gaining of education within the job plans of clinical and laboratory specialists working in rare diseases.

As an example of targeted training, there is the case of newborn bloodspot screening. Education and training has been focused on relevant health professionals, providing updates on the conditions screened for and the importance of the screening. Power point presentations have been developed and trainers identified and trained across Wales. A health professional's information pack has been developed and summary sheets on each of the conditions included. The information and presentations are also available via the internet. Two educational sessions on MCADD have been held (one in North Wales and one in South Wales) which were aimed at health professionals who would be involved in the care of babies suspected to have MCADD.

Primary care teams could further be supported to improve care through:

- rapid access to specialist advice
- tools to support more timely diagnosis
- local care pathways including advice about access to specialist services
- clear and timely management plans from specialist services
- access to high quality patient information and support networks
- training on the identification and management of rare diseases
- feedback and shared understanding of diagnostic delays
- feedback on patient experience.

Patients and carers can play an important role in this through their own expert knowledge of their condition. Clinicians in local services must recognise the limitations of their own knowledge and when it is necessary to refer on/seek advice from specialist centres or experts. Patients and patient organisations play a role in education where there is no specialist clinic for a condition.

WHSSC will work with services to develop lead clinicians where service provision is outside Wales and encourage these clinicians to liaise with specialist services. When developing commissioning policies and service specifications, WHSSC will work collaboratively with LHBs to consider the patient pathway and the link between primary care, secondary care and tertiary care.

Commitment in the UK Strategy on education and support for professionals	Actions to be taken forward for Wales
14.Where appropriate support the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered	• The Welsh Government will commission the NHS Wales Information Service to report by 31/03/15 on options for implementing this commitment, liaising with the National Genetics Education and Development Centre (NGEDC) and other UK countries • The Welsh Government will by 30/09/14 agree a plan to - improve access for primary and secondary care to the information in the Orphanet database by improving usability - formalise and agree care and referral pathways across Wales for access to appropriate expertise and highly specialised services - develop with WHSSC a common access page to Orphanet and linkage to specialised services/pathways over one year - discuss the Implementation Plan with the General Practice National Specialist Advisory Group and agree on action
15. Improve education and awareness of rare diseases across healthcare professions, including: - involving patients in the development of training programmes - encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics - ensuring awareness of methods and clinical techniques used in differential diagnosis.	• The Welsh Government, through the Deanery and Welsh Education and Development Service, will work with other UK countries and NGEDC to review and update curricula for all health professions by 30/09/14
45. Ensure that training and education are available to the NHS workforce, highlighting the importance of NGS to all aspects of rare disease care, including support for evidence based local counselling for patients and their relatives who receive NGS results.	• See action 15 above.

Theme 4 Co-ordination of Care

Introduction

This section deals with the situation where an individual may need support from different clinicians possibly far apart or far from home. There are sections dealing with:

- service specification
- specialist centres
- information to support planning and co-ordination of care.

Service Specification

Commissioning and planning systems for rare diseases will need to ensure equitable access to health services and treatments across the UK, regardless of a patient's location. Specialised commissioning and planning bodies across the country therefore need to work

together to improve accessibility to services for rare diseases, as due to the low numbers of patients affected by individual diseases, it is not possible for services to be established for all rare diseases in each of the four home countries. WHSSC will lead on this.

WHSSC has responsibility for developing and agreeing service specifications for some specialised services and highly specialised services. Where services are provided in England WHSSC will work with NHS England to develop these. To support the development of referral pathways and implementation of commissioning policies, WHSSC will work with providers to develop and implement specific service specifications, to include:

- defined providers and conditions treated
- clinical access criteria
- referral pathway including access to genetics and diagnostic support
- measures of quality including clinical effectiveness, patient safety and patient experience
- ensuring there is standardised data collection and use of systems to record genetic and disease specific information
- standards of care which patients can expect
- details on transition and
- details on any relevant patient support groups.

WHSSC as part of its commissioning cycle will aim to review specialist services providers and ensure that they meet the following minimum standards and:

- have a sufficient caseload to build recognised expertise
- where possible, do not depend on a single clinician
- co-ordinate care and ensure all patients have an appropriate care plan
- arrange for co-ordinated transition from children's to adults' services
- involve people with rare conditions and their families and carers
- support research activity
- ensure their expertise is available to families and their healthcare teams and
- ensure that patients have access to their personal data and are included in any related correspondence.

WHSSC will also establish a process for the review of quality and outcome indicators and evidence-based practice. Where providers are in England, WHSSC will work closely with NHS England to review and evaluate services against service specifications. By identifying referral pathways and service specifications WHSSC will be able to establish contracts with all the relevant providers.

The Wales Rare Disease Implementation Group will examine the options for engaging patients, clinical care teams, researchers and industry in discussing how the current service can be improved.

Commitment in the UK Strategy on service specification	**Actions to be taken forward for Wales**
23. Continue to develop service specifications for rare diseases. This will include country specific care pathways and a 'generic' care pathway that sets out best practice that can be applied to all patients with rare diseases in the UK (particularly where there are no disease specific pathways). The generic care pathway will include: - an appropriate care plan for all patients with a rare disease	• WHSSC will by 31/12/14 prepare as part of the process of developing its specialised services strategy a plan to: - develop and agree service specifications for specialised services and highly specialised services - define rare diseases and areas for priority - review existing service specifications for specialised services and highly specialised services used in other areas for

- clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis - the development of seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age - access criteria and measures of quality and outcomes.	appropriateness to Wales - develop service specifications for those identified above incorporating all areas of the commitment - review Heads of Agreements and Service Level Agreements with English providers - work with the lead nation on the development of generic care pathway.

Specialist Centres

A working group will be established to ensure formal links are developed between all those involved in the care and treatment of patients to ensure they receive the best possible care and that it is optimally coordinated. WHSSC through the development of service specifications will ensure that the designated specialist centre meets the standards identified below. As a number of centres will be in England, WHSSC needs to work closely with NHS England to support this commitment.

There will be a systematic programme of designation for centres of excellence for rare diseases supported by networks linking into local services throughout the UK. This will result in the development of centres that have expertise in rare conditions and will be able to centrally coordinate care of patients with that disease. They will have in-depth knowledge of the implications of the condition and know what services are likely to be required and when.

Currently, England has 70 or so Highly Specialised Clinical Service Centres. Research into treatment is an important aspect of these centres. Going forward, Wales should consider the potential to participate more closely with existing Centres (for example virtually), and develop a small number of such centres for rare disorders in which research the UK is leading on research. Examples of involvement in clinical treatment trials led by Wales include the TESSTAL and TRON trials in tuberous sclerosis.

Patient organisations are ideally placed to liaise between newly diagnosed patients and the centre of excellence to ensure that all patients are aware of the centre's existence and that they are accessing the services that the centre is able to provide. There must be a core set of responsibilities agreed between the relevant patient organisations and centres of excellence to optimise this liaison, but there must also be a level of confidentiality maintained within each to ensure that patients feel assured that their information is secure.

Introducing engagement systems so that patients/patient representatives are able to feed into work and developments taking place by regulatory bodies is essential. An example of where this is already taking place in Wales is in the context of the appraisal and approval of new medicines. A group has been established for patients and the public to become involved in the work of the All Wales Therapeutics and Toxicology Committee.

It is essential that all regulatory bodies have open and transparent channels for patients, family members and patient groups to become involved with work that will impact directly on patients. All regulatory bodies with a decision-making role in relation to access to services, support and advice will be required to develop a patient forum to ensure that patients are at the centre of their work.

Commitment in the UK Strategy on specialist centres	Actions to be taken forward for Wales
24. Agree that specialist clinical centres should as a minimum standard: – have a sufficient caseload to build recognised expertise – where possible, not depend on a single clinician – coordinate care – arrange for coordinated transition from children's to adults' services – involve people with rare conditions, and their families and carers – support research activity – ensure their expertise is available to families and their healthcare teams.	• WHSSC with LHBs as providers will work alongside NHS England to: - develop and agree service specifications - develop an agreed progress for monitoring services against specification - implement and monitor services against service specification including outcome and audit monitoring
25. Ensure that the relationship between the specialist clinical centres and science and research is explained to and understood and put into practice by: practitioners delivering local health and social care; the research community; industry; academia.	• The Wales Implementation Group for Rare Diseases will advise on this in conjunction with other interests and the other countries of the UK.
26. Set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support.	• The all Wales genetic laboratory is a member of UKGTN and communicates with other member laboratories and referring clinical centres on the services it provides and receives.
27. Ensure that specialist clinical centres are as concerned with research as with health and social care support and that they develop networks that provide professional-to-professional dialogue and collaboration across a wide range of experts, including internationally (especially for those conditions that are ultra-rare).	• See action 25 above.
28. Work with international partners wherever possible and develop UK-wide criteria for centres to become part of an expert reference network to increase the flow of information between patients and professionals in a range of disciplines.	• See action 25 above.

Information to Support Planning and Co-ordination of Care

At present information on rare diseases is not captured in a consistent and useable way to support planning and treatment and outcome monitoring. There are very few specific registries, though the Welsh Congenital Abnormalities database (CARIS) does capture information on a wide range of cases.

Data around particular individuals can be scattered across GP records and various hospital-based systems, with no routine way of collating data for individuals or for groups with a shared diagnosis. The requirement is collated information for individuals, so that their overall circumstances and treatment record can be viewed as a whole. This might involve information on activity undertaken a different sites in Wales and outside. Better information is needed on overall numbers affected to support planning.

The Welsh Government is about to begin work on a new health information strategy. The commitments to improve information in the field of rare diseases will be taken forward as a part of that work. In doing so, the Welsh Government will work with the other UK countries to establish links necessary to support joint planning and unified records for individuals crossing national borders.

WHSSC will continue to work with the other three countries and international partners in relation to rare diseases and the provisions of services. In particular it will be an active member in the development of generic care pathways and work with colleagues in Europe on the development of the European Orphanet coding system.

CARIS is a comprehensive reporting register that includes many rare congenital and childhood diseases stretching back to 1998. It utilises clinical reporting, data downloads and web reporting of cases, routinely capturing data from sources including cytogenetics, newborn bloodspot screening and medical genetics.

A prevalence table of rare conditions first published in 2009 included genetic, hereditary and syndromic conditions. That list has now be updated and extended and comprises 15 years' data collection in Wales on a birth population of ½ million total births. Prevalence rates are compared with population prevalence estimates from Orphanet, where these are available. CARIS has recently published survival data to five years on a range of conditions (children born 1998-2006) (www.caris.wales.nhs.uk).

CARIS is a member of EUROCAT, the European network of congenital anomaly registers. While EUROCAT restricts its activity to structural anomalies coded within the Q chapter of ICD-10, CARIS has a much broader outlook and registers congenital cancers e.g. Wilms tumour, hereditary blood disorders, e.g. haemophilia, endocrine disorders and in-born errors of metabolism. It also collects data on neuromuscular disorders such as Duchenne muscular dystrophy and sensorineural hearing loss.

EUROCAT membership allows data from over 40 registers across EUROPE to be processed in the same way and for checks to be made for data quality. CARIS consistently performs within the top five for overall prevalence (indicating a good level of ascertainment of cases) and for achieving good quality as measured by the data quality indicators (DQIs). See EUROCAT website www.eurocat.net.eu.

CARIS is well placed to continue to play an important role in rare disease registration in Wales.

Commitments in the UK Rare Diseases Strategy on information for the planning and co-ordination of care	**Actions to be taken forward for Wales**
16. Monitor the development of ICD-11 in preparation for its adoption.	• The Welsh Government will ensure that Wales is involved as a member of the UK team that is leading on this issue.
17. Work with colleagues in Europe on the development of the European Orphanet coding system and considering the adoption of Orphanet coding and nomenclature.	• The Welsh Government will support action to: - extend Orphanet listings from genetic testing services to include specialist clinical services. - foster better communication between primary and secondary care and support more focused referral using common websites such as Orphanet to improve diagnosis.
18. Standardise data collection and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.	• The Welsh Government will agree by 30/09/14 how to take this forward as part of the work on a new Welsh health information strategy.
19. Explore options to improve the link between existing patient data and electronic health records.	• The Welsh Government will agree by 30/09/14 how to take this forward as part of the work on a new Welsh health information strategy.

20. Assess the potential for rare disease databases where they do not exist.	• The Welsh Government will review the options, in discussion with other UK countries including assessing the opportunity for extending the Welsh CARIS system.
21. Agree international standards, building on existing NHS standards.	• The Welsh Government will agree the way forward with other UK countries by 30/09/14.
22. Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally.	• The Welsh Government will agree the way forward with other UK countries by 30/09/14.
29. Improve systems to record genetic and other relevant information accurately to record the incidence and prevalence of disease and support service planning and international planning.	• The Welsh Government will agree by 31/06/14 how to take this forward as part of the work on a new Welsh health information strategy.
30. Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments.	• The Welsh Government will agree by 30/09/14 how to take this forward as part of the work on a new Welsh health information strategy.
31. Look at how the 4 UK countries develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways.	• The Welsh Government will agree by 30/09/14 how to take this forward as part of the work on a new Welsh health information strategy.

Theme 5 The Role of Research

Improving Research on Rare Diseases

Effective Research and Development (R&D) is essential to improving the health and wellbeing of Wales and the quality of services. There is growing evidence worldwide that clinical standards improve in an environment strongly influenced by research. Innovative healthcare is also associated with significant income generation. In addition to the development and exploitation of intellectual property, a strong health science and social care research base fully integrated with the Welsh NHS and social services provides a platform to attract major investment from pharmaceutical, biotechnology and other industries.

Wales has high quality expertise in specific areas of rare disease research, notably, for example tuberous sclerosis and cortical brain malformations. Currently, most rare disease research in Wales is undertaken at Cardiff University/University Hospital of Wales (notably in the Institute of Medical Genetics and Department of Biochemistry). Research is often based on combined local clinical and research expertise and supported by funding from Government and external bodies.

The National Institute for Social Care and Health Research (NISCHR) supports efforts to streamline research processes, stimulate excellence and innovation and build research capacity. That in turn attracts research funding to Wales from the UK Government, charities and the commercial sector. NISCHR funds the Wales Gene Park, which currently focuses the major part of its resources on rare diseases and supports education and engagement for the NHS and the public. NISCHR also funds the Cancer Genetics Biomedical Research Unit which dedicates about 15% of its resources to rare diseases, and the Academic Health Science Collaboration which currently funds five part time Fellowships in rare disease research. NISCHR also supports a skilled all-Wales research professional network of research co-ordinators and nurses with experience of direct patient and service user contact,

who support the recruitment of patients to high quality research studies, including in rare diseases, in line with demand.

Overcoming the barriers to research into rare diseases provides opportunities for (and indeed demands) even greater collaboration between industry and academia, not just in drug discovery but also in supporting and enabling activities such as biomarker identification and companion diagnostics, which will play an increasingly important role in research and innovation in the future. NISCHR has worked to strengthen links with industry and promote fruitful collaborations with the life science sector, including through the establishment of Health Research Wales to provide a one-stop source of information and support for companies wishing to undertake clinical research in Wales, providing partnering and facilitation services promoting the strengths and capabilities of Wales and assisting in linking industry to interested clinicians and academics for collaborative research projects.

It is especially important for Wales to participate actively, and reciprocally, in existing fora and collaborations, and to forge new ones to increase patient access to research and funding opportunities for Wales-based researchers. Rare disease research will benefit greatly from the further development of genomic medicine in Wales. The Welsh Government has agreed that a capital bid should be prepared for infrastructure to capture, store and analyse human genomic information to meet clinical, research and economic development needs. Rare diseases research will also benefit from NISCHR-led initiatives on making smarter use of routine data.

Much of the existing NISCHR-funded infrastructure and programmes designed to support health and social care research in Wales is contracted until March 2015. The principles that will underpin Welsh Government support in this area beyond March 2015 – e.g. creating a culture supportive of R&D in health and social care organisations, increasing capacity to support high quality commercial and non-commercial health research and facilitating strong partnerships with key individuals and organisations in the UK and internationally – will benefit all patients, including those with or at risk of rare diseases.

Commitment in the UK Strategy on improving research	**Actions to be taken forward for Wales**
32. Work together to identify a selection of the rare diseases most suited to the development of best-care pathways and propose other rare diseases for possible pathway development, taking on board the needs of patients and carers and the challenges faced during delivery of the first set of pathways.	• AWMGS will work with WHSSC, other specialties, experts from higher education institution s and patients and families to extend its portfolio of pathways for specific rare diseases, focusing on rare diseases for which there is special expertise in Wales by 31/03/15. • AWMGS will work with other UK nations and specialist and professional bodies (Clinical Genetics Society, British Society for Genetic Medicine and others) ongoing from 31/03/14.
33. Examine how they can ensure appropriate funding for excess treatment costs for research in rare diseases.	• NISCHR will consider whether any adjustments should be made to the current ETC arrangements specifically in relation to rare disease research by 31/03/15. • NISCHR will work with local R&D Offices to ensure adherence to current policy on excess treatment costs associated with research by 31/03/15.
39. Work with the research community, regulators, providers of NHS services and research funders to develop risk-proportional permission systems.	• The Research Ethics Committee for Wales, NISCHR and local R&D Offices in Wales will work together to respond to UK-wide initiatives, ongoing from 31/03/14.

40. Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.	• Ongoing from 31/03/14, NISCHR will continue to provide support and advice to researchers and lay organisations in producing high quality and accessible information for research participants and on how best to engage with and consent research participants, in particular through its Public and Patient Involvement initiatives.
41. Promote good practice and the use of systems which facilitate a consistent and streamlined process to local NHS permissions of publically, charitably and commercially funded research with an aim to reduce timescales.	• NISCHR will work with all stakeholders to develop actions to streamline processes and reduce the time taken to approve and commence health studies in Wales.
46. Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.	• Ongoing, the Welsh Government will continue to develop actions in partnership with other administrations and in collaboration with industry to ensure that Wales is a receptive environment for investment in research and the uptake of innovation. • Welsh organisations involved in rare disease research, led by Wales Gene Park, will develop commercial links using existing Welsh Government, NHS and higher education institution mechanisms by 01/04/15.
48. Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders.	• Ongoing, in partnership with all stakeholders, the Welsh Government will encourage and develop the kind of research collaborations required to remove the special barriers to the diagnosis, treatment and management of rare diseases.
49. Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.	• An external review of NHS R&D offices – due to finish in April 2014 – will be used to determine how functions are managed in the future, beyond 31/03/15 and explore new opportunities to further streamline activities.
50. Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.	• In developing proposals for funding beyond 31/03/15, NISCHR will continue to utilise and invest in existing structures and, where appropriate, help to develop new ones, to ensure that the needs of Welsh patients are adequately addressed and high quality research in rare diseases in Wales can be supported, particularly in areas of existing expertise or with exceptional future potential.
51. Improve engagement between key stakeholders, including: – patients and relatives – main funding providers – healthcare commissioners – NHS hospitals and specialist care units – industry (pharmaceutical, biotechnology, IT, diagnostics).	• NISCHR funding beyond 31/03/15 will put citizens at the heart of creating new knowledge and putting new knowledge into practice and create a new partnership between the population, service users and researchers, based on co-ownership and transparency in health and social care research.

Patient Engagement with Research

The Welsh Government recognises the benefits of involving patients, the public and service users in supporting health and social care research in Wales across all disease areas, including rare diseases. NISCHR currently funds an *Involving People* network as part of its broader Public and Patient Involvement (PPI) work and also provides resources to recruit and train lay people with particular research interests and provide them with opportunities to engage with research. Future action will include the looking at ways to develop useful indicators of progress, including for example the number of Welsh patients consented to relevant studies into rare diseases.

In addition to the register for congenital anomalies in Wales (CARIS), basic disease-specific registries for Wales/the UK are maintained locally for several disorders, for example the UK Rett Syndrome Register and the Wales Registers for Polyposis Syndromes and Von Hippel Lindau Disease maintained at the Institute of Medical Genetics in Cardiff University. There is potential, with support, for these to be developed further.

As Wales can only sustain high quality research in a few rare disease areas, it is important that patients affected by rare diseases for which no research takes place in Wales should be able to access and participate in high quality research outside Wales. Access to information on research into rare diseases for patients and families can be improved. More information about research opportunities needs to be included in care plans.

Recruitment to some types of rare disease research poses special challenges, for example clinical trials. The National Institute for Health Research (NIHR) Genetics National Specialty Group has representation from across the UK and serves as a network co-ordinating support for and recruitment to NIHR/NISCHR portfolio research studies for many rare diseases across the country. International collaboration is also essential for research. Orphanet is a European web-based data resource for rare diseases that lists research studies and trials but does not directly support these. Most clinical trials are also listed (and searchable) at clinicaltrials.gov.

Commitment in the UK Strategy on patient engagement in research	**Actions to be taken forward for Wales**
7. Support patients to register on databases where they exist.	• NHS Wales, patient organisations, funding bodies, industry, researchers and clinicians will consider how existing disease registries can be best supported and promoted and sustainable data collection systems be put in place and how patients of all ages can be supported by their clinical team to join disease registries, agreeing a way forward by 31/03/2015.
8. Help patients to contribute to research and other activity related to rare diseases.	• NISCHR will work with patient organisations to ensure appropriate rare disease representation within its current *Involving People* network by 31/12/14, and in future arrangements to support its Public and Patient Involvement strategy. • See action 7 above.
33. Examine how they can encourage service providers to involve patients in research.	• NISCHR will ensure that the core principles of public and patient involvement in research are embedded in the new research support mechanisms and infrastructure it has in place from April 2015.
35. Use portals to connect patients and relatives to enhance research participation and where appropriate, promote self-enrolment to approved research studies with online consenting, self-reporting and use of social media.	• NISCHR, as part of its broader Public and Patient Involvement work and in partnership with patient organisations and their stakeholders, will develop actions by 31/03/15 to connect patients affected with rare diseases and their families with opportunities to participate in relevant research.
36. Encourage patient groups to get involved with regulatory bodies.	• As part of the action under Commitment 39, the relevant partners will introduce engagement systems for patient groups to work together with Welsh regulatory bodies.
37. Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.	• Genetic Alliance, Rare Disease UK and Wales Gene Park will prepare a plan for partnership by 31/03/15.

38. Explore the feasibility of the UK Clinical Trials Gateway including experimental medicine trials for rare diseases to provide information for patients and their families about research trials.	• NISCHR will work with NIHR and make recommendations by 31/03/15.
47. Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.	• LHBs will as part of their reporting and review arrangements as required under commitments 1-3 review and report on performance on supporting research. • NISCHR will continue to support the recruitment and training of patients, carers and service users and their engagement in research development and delivery in the run up to March 2015 and beyond. • NISCHR will work with NHS Wales to develop mechanisms to ensure that, where appropriate, research opportunities can be included in care plans for patients with rare diseases.

Case Study - Research

My children suffer from an undiagnosed epilepsy syndrome. Along with complex epilepsy that is drug resistant, her children have severe, profound learning difficulties. They also have mobility issues, problems with regulation of body temperature, no awareness of danger and limited life skills.

My family first volunteered to participate in a research study which focused on large epilepsy families and the discovery of new epilepsy genes. The study results identified a novel mutation within my family, which is extremely rare but that provides a specific diagnosis for my family's medical problems.

The experience of participating in research is extremely positive; the researchers provided us with detailed information about what would be expected of us as part of the study and support and encouragement throughout the research. Anna felt at ease during the whole process and feels that others can gain real benefits from being part of research.

Participating in research studies can provide significant benefits to participants, both practically and psychologically but can also carry risks All research involving patients in the NHS is subject to rigorous governance to ensure that the balance of risk and benefit is appropriate.

Annex - The UK Strategy commitments and the Welsh Plan

Commitment Number	Welsh Plan section		Commitment Number	Welsh Plan section
1	1		28	4 centres
2	1		29	4 information
3	1		30	4 information
4	1		31	4 information
5	1		32	5 improving research
6	1		33	5 improving research/ 5 patients and research
7	5 patients and research		34	1
8	5 patients and research		35	5 patients and research
9	2		36	5 patients and research
10	2		37	5 patients and research
11	3 pathway		38	5 patients and research
12	3 pathway		39	5 improving research
13	3 evaluation		40	5 improving research
14	3 education		41	5 improving research
15	3 education		42	3 evaluation
16	4 information		43	3 evaluation
17	4 information		44	3 evaluation
18	4 information		45	3 education
19	4 information		46	5 improving research
20	4 information		47	5 patients and research
21	4 information		48	5 improving research
22	4 information		49	5 improving research
23	4 specification		50	5 improving research
24	4 centres		51	5 improving research
25	4 centres			
26	4 centres			
27	4 centres			

Note the Welsh actions are in the tables in each chapter. Where there is more than one table in a chapter, the additional wording above indicates which is the relevant one for that commitment. For commitment 33 there are actions in two tables.

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