Llywodraeth Cymru
Welsh Government

www.cymru.gov.uk

Welsh Implementation Plan for Rare Diseases

February 2015

Digital ISBN 978 1 4734 2130 1

WG23019

CONTENTS

Foreword by the Minister for Health and Social Services

I am pleased, following consultation, to be able to issue this final version of our Implementation Plan for Rare Diseases, which confirms our strong commitment in Wales to improve services for those who live with rare diseases.

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 work with the people and families concerned at every stage, from prevention through to self care and the provision of specialist support. Our approach should reflect our commitment to prudent health care and coproduction. Besides a true partnership approach with those affected, this will involve co-ordinating the contribution of primary care, available everywhere, and various specialised services, which may operate only at regional, national or even international level, and linking also to social services, the third sector, industry and the research community.

To ensure we remain on track with implementation and in touch with developments at all these levels, I am establishing an Implementation Group in Wales to work with the NHS and other agencies in Wales and beyond. Wales will be an active partner with the UK Rare Diseases Advisory Group and the UK-wide Stakeholder Forum will monitor and report on progress in the four countries. This is an important area where I expect to see continued progress in the coming years.

Foreword by the Chief Executive of NHS Wales

This is the first Implementation Plan for Rare Diseases in Wales and it is important that it is considered as a delivery plan on an equal level with others recently issued.

Providing safe, sustainable, high quality care for rare diseases is particularly challenging as they include so many and such varied conditions. The success of this Plan will depend on effective co-ordinating in many areas and the actions in this Plan and the mechanisms it describes aim to ensure far better support for patients and their families and carers.

In this area particularly, we need to see a real partnership approach, across services, agencies, countries and above all between the individuals, their carers, patient organisations and the NHS.

This Implementation Plan will direct our work in Wales. 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 and its partners, providing support as necessary. The expectation now is to see a new and different approach with more clarity to improvement and reporting.

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. Rare diseases can take many and varied forms, including physical and psychiatric disorders. Some 80% are genetic in origin, but that means many are not. Most appear at an early age but again, that means many do not and so services need to be alert to the signs at many different stages in people's lives. It is estimated some 350 diseases account for 80% of cases, but it is also estimated that over 200 new forms are identified each year.

This Rare Diseases Implementation Plan provides a framework for action by the Welsh Government, 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*[1], introduced an ambitious programme for health in Wales with the aim that:

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

Achieving Excellence: The Quality Delivery Plan for the NHS in Wales for 2012-16 [2] 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*[3]. All four countries committed themselves to:

[1] http://wales.gov.uk/topics/health/publications/health/reports/together/?lang=en
[2] http://wales.gov.uk/topics/health/publications/health/strategies/excellence/?lang=en
[3] https://www.gov.uk/government/publications/rare-diseases-strategy

- 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; and,
- 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 many 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 conditions.

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 take a slightly different approach, reflecting local resources, priorities and practices. Accordingly each will separately indicate its own way forward, building on current services, drawing on best practice and aiming to achieve the most effective use of resources. Collectively the

plans will 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. Many agencies and people 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 for Health and 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 disease 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 training and 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.

The **Welsh Rare Diseases Implementation Group** 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.

Everyone in Wales has a contribution to make to improving health. People with rare diseases, like others in Wales, should be taking a responsibility for their own health and contributing to the success of the NHS. They 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). WHSSC will also produce an annual report.

Under new local planning arrangements in the NHS, GP practices will work together and more widely to analyse needs and organise and deliver services. This should lead to better support for people with rare diseases by health services such as health visitors, nurses, dentists, social services and the third sector who should look to work together to develop new skills. It will also be an opportunity to look again at how best to support people who live far from specialised centres or in isolated locations through innovative use of options such as telehealth and telecare facilities and outreach clinics. The planning system is also a way of improving Welsh language services, drawing on the findings of the recent report of the Welsh Language Commissioner on primary care[4].

11. Reporting progress

The actions in this Plan involve a wide range of actors: the Welsh Government (including NISCHR), LHBs, WHSSC, the Wales Screening Committee, All-Wales Medicines Strategy Group, the All-Wales Medical Genetics Service, the Welsh Rare Diseases Implementation Group and the Wales Gene Park.

There are also other stakeholders with an interest including individuals directly affected, their families, patient support organisations and other local service providers. As stated, LHBs will be expected to prepare an annual report on progress, but that will account for only a part of the activity envisaged.

To enable all concerned both to share and assess overall progress it is proposed that the Welsh Government will convene an annual rare diseases meeting at which all the main actors should present a brief public report. Each organisation will present on its achievement over the past 12 months and also on its intentions for the next 12 months, and provide a short written annual report. Such an event will improve transparency and engagement whilst encouraging accountability and the sharing of best practice. The event will be open to external stakeholders and the public. A document drawing on the event will serve as a national annual report on progress.

[4] http://www.comisiynyddygymraeg.org/English/News/Pages/The-Welsh-language-an-integral-part-of-care---.aspx

Section 2 - The Plan

Theme 1 - Empowering those affected by Rare Diseases

The UK Rare Disease Strategy aims to ensure improvements across the whole 'patient journey', from the first contact with the GP through diagnosis to ongoing 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. NHS professionals will therefore need to be very attentive to the views of those affected who may have a much better understanding of the problem and its implications than they have.

A wide range of third sector groups help provide the knowledge and guidance patients need to secure access to health and social care services and to manage and improve their condition. Many are able to act as the patient's advocate when needed and bridge the gap between patients, their families and the health service. The Welsh Government will continue to work closely with patient organisations to support the patient voice in the development and implementation of the rare disease strategy and improving access to information.

Mechanisms for patient involvement in service provision and research need to be strengthened in relation to rare diseases, and formally extended to include consideration of non-medical needs. Those affected will be an important source of information and understanding both on the condition and the service response, which can help the immediate team and the service more widely to perform better. Action will be taken to improve patient involvement in the planning system.

The Welsh Government recently issued guidance - *Agreeing Individual Care with People who have Long Term Conditions*[5]*.* This sets out the Government's expectations about making available to people an individual care agreement, proportionate to the needs and preferences of the individual. It is clear that this can take a variety of forms. The plan is described in the guidance as an agreed summary of discussion and actions, shared by all concerned, promoting supported self care and co-ordinated service support, and containing the essential information on the person concerned and his or her care.

The guidance also notes that: *'Individuals with complex problems, their family and carers greatly value a single point of contact with a professional who is responsible for overseeing and co-ordinating the delivery of their agreed formal care and support. ……..In these cases, the support of a care co-ordinator or key worker, such as a specialist nurse, may be appropriate. It is important that this key contact is agreed with the individual, their family and carers and is communicated to everyone involved in providing care and support and is recorded within the care plan.'* LHBs and WHSSC should review their arrangements in this regard and consider the matter as part of their annual review and reporting process.

[5] The guidance is available on the government website: http://wales.gov.uk/topics/health/nhswales/majorhealth/chronic-conditions/

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

Actions

Who	Action	Timescale
Welsh Government	Consider with the NHS and Genetic Alliance UK how best to monitor patient views on service provision (commitment 1)	By 31/03/15
	Commission Public Health Wales, the NHS and Genetic Alliance UK, drawing on the sections in the reports above from the NHS on patient information, to review and advise on improving information provision (commitment 5)	During 2015/16
LHBs	In order to strengthen meaningful and sustained patient involvement in rare disease service provision and research, review: - their current arrangements for providing information and diagnostic and treatment services to people with rare diseases, including those without computer access, and for facilitating engagement between patients, clinical care teams, researchers and industry wherever practical (commitments 3, 5, 47); - their current arrangements for working with the third sector organisations supporting people with rare diseases and for putting people in contact with them (commitment 1); and, - their arrangements for preparing assessments and care plans for people with rare diseases, and on access for patients (or where appropriate their parents or guardians) to their personal data (commitment 6).	By 31/03/15
	Issue their first annual report on rare diseases, addressing the findings from the above reviews, and setting out how they will in addition: - 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 (commitment 1); - ensure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services (commitment 4); - 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 (commitment 2); and, - recognise the need to encourage effective and timely liaison between the NHS and other public service providers and to encourage providers to consider the effects of rare diseases on people's lives when they are developing and managing services (commitment 3).	By 30/06/15
WHSSC	Establish a clinical evidence reference group for rare	By

	diseases to review evidence for these conditions, its membership to include patient organisation representatives (commitment 1)	31/12/2014
	Make it a required standard within its service specification 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 (commitment 2); - NHS clinical teams engage with patients using patient experience surveys and patient reported outcome measures (commitment 4); and, - there is information 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 (commitment 34),	Initial group agreed by 01/12/14
	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. (commitment 4).	Ongoing
	Issue their first annual report on Rare Diseases	By 30/06/15

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.

The majority of cases of rare diseases are not identified as part of a population-based screening programme and the role of screening and carrier testing is limited. 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 reproductive decisions. The counselling provides accurate information about risk and the availability of prenatal testing options as well as on pre-implantation testing if appropriate.

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.

Consent processes need to be appropriate for both screening and symptomatic pathways to ensure that parents or patients are able to have sufficient information on which to make an informed choice.

General Practitioners need to be informed of babies identified with a rare disease to ensure they are well placed to provide appropriate generalist support to the family.

In future, there will 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.

Actions

Who	Action	Timescale
Welsh Screening Committee	Ensure that stakeholders in Wales are aware of current UK National Screening Committee consultations via networks and websites (commitment 9).	Ongoing

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.

Diagnosis of a rare disease via screening is relatively unusual. 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 offer the prospect of prevention of disease through prenatal or, in the case of IVF, pre-implantation diagnosis and the prevention of disease complications through increased surveillance, prophylactic procedures, chemoprevention or targeted therapy.

Genomic technologies are being used increasingly frequently with newer approaches allowing sequencing of the whole genome (the whole genetic structure of an individual) 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. NHS Wales is exploring ways in which the existing infrastructure for genomic medicine could be further developed as a platform for clinical services, research and innovation.

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.

The principles of prudent healthcare should be used to guide the patient’s journey or pathway through the system; including self-care, diagnosis and treatment processes, with the skills of different clinicians being combined alongside full patient involvement to help achieve the best outcomes. This and the next chapter set out how that should be done.

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; and,
- education and support for professionals.

Developing pathways to diagnosis

a. Primary Care Services

Whilst many rare diseases will be identified at or before birth through specialist services, others first appear later, presenting community services with the challenge of identification and management. A GP will have very limited experience of rare diseases and it is clearly impossible for any individual GP to have an expertise in every 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.

The Royal College of General Practitioners has looked at a number of aspects of the management of patients with suspected or confirmed rare disease, including co-ordination of a clinical network for rare diseases, developing an e-learning package, and computer generated ‘Red Flag’ prompts to aid early diagnosis. Key priorities for primary care services are creating ‘safety nets’ to catch people who have symptoms that form a pattern or do not resolve, and creating effective co-ordination of different aspects of care and continuity of care. The Royal College in Wales will feed progress to primary care clinical leads in LHBs and other material such as the ‘Red Flags’ work will be promoted through the Deanery website.

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; and,
- 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 and of geneticists in; managing rare diseases, education of health professionals, setting standards and developing pathways of care for appropriate use of genetic resources is appropriately reflected in job plans.

c. Specialised Services

The Welsh Health Specialised Services Committee (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 expertise, information and advice, as well as 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[6], the international internet portal for rare diseases and orphan drugs.

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. The right tests at the right time can improve efficiency and reduce anxiety by ensuring quicker diagnosis and avoiding ineffective treatment.

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.

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.

[6] http://www.orpha.net/consor/cgi-bin/index.php

WHSSC is a member of the All Wales Medicines Strategy Group (AWMSG), the Orphan and Ultra Orphan drugs group 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.

A review of access to orphan and ultra-orphan drugs and of the Individual Patient Funding Request process, through which patients can secure access to treatments not routinely approved, is due to be completed in autumn 2014.

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. This will enable the development and validation of diagnostic protocols and specialist bioinformatic analysis, to allow Wales to keep pace with regional genetics services elsewhere in the UK, the AWMGS to be a full partner in the UKGTN, and Welsh clinicians to access appropriate services and laboratory testing that will benefit patients and their families.

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; and,
- 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, healthcare scientists 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 most other specialties such as paediatrics, cardiology and oncology.

Continuing healthcare professional 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; and,
- 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 biomedical and health science 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; and,
- 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.

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; and,
- 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.

The NHS needs to be responsive to rather different ways in which people may need to learn in relation to rare diseases because of rapid changes in technology and biomedical understanding and the need for reflection and patient interaction in relation to particular cases. This may need to be recognised in planning arrangements, contracts and continuing education.

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.

Actions

Who	Action	Timescale
Welsh Government	Agree a plan to: - improve access for primary and secondary care to the information in the Orphanet database by improving usability; - develop with WHSSC a common access page to Orphanet and linkage to specialised services/pathways over one year; - commission the NHS Wales Information Service to report by 31/03/15 on options for supporting the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered, liaising with the National Genetics Education and Development Centre (NGEDC) which is within the National School for Healthcare Science (NSHCS) and other UK countries; and, - discuss the Implementation Plan with the General Practice National Specialist Advisory Group and agree on action (commitment 14).	By 30/11/14
	In relation to Next Generation Sequencing (NGS): - work with AWMGS to review the options to accelerate progress and ensure the best value from recent capital investment (commitments 42,43); and, - 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 (commitment 44).	By 30/11/14
	Improve awareness and education regarding rare diseases, including support for evidence based local counselling for patients and their relatives who receive NGS results, work through the Deanery and Welsh Education and Development Service with other UK countries and NGEDC to review and update curricula for all health professions (commitments 15, 45).	By 30/11/14
LHBs	Work with GPs and other health care professionals to: - develop agreed local care pathways for people with rare diseases (note: where possible, this should be a single all-Wales pathway; and wherever possible, pathways should, minimise avoidable travel); - 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;	Ongoing

	- 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; and, - seek patient and carer feedback to inform service improvement (commitments 11, 12).	
WHSSC	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 and work with clinicians 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 (commitment 13).	Ongoing from 31/03/14
All-Wales Medicines Strategy Group (AWMSG)	Continue to appraise new medicines and develop their processes to do so, taking into account the results of the reviews of access to access to orphan and ultra-orphan drugs and of the Individual Patient Funding Request process, through which patients can seek to secure access to treatments not routinely approved (commitment 13)	Due to be completed in autumn 2014
All-Wales Medical Genetics Group (AWMGS)	In conjunction with **WHSSC** - 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; and, - report on these to the Minister for Health and Social Services (commitment 10,11).	By 30/11/14
	In conjunction with the **LHBs** and **WHSSC** agree a plan to: - formalise and agree care and referral pathways across Wales for access to appropriate expertise and highly specialised services, with an agreed plan; - establish a system of monitoring Welsh genetic testing activity; - develop a clear commissioning model for Welsh genetics services; - monitor and evaluate access to testing; and, - set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support (commitments 11, 12, 26).	By 31/03/15
Welsh Rare	Review arrangements for identifying and supporting	By

Diseases Implementation Group	patients for whom no diagnosis can be agreed and agree future actions (commitments 11, 12).	30/11/14

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.

Care needs will change over time, both as people grow older and as their condition develops. The role of the GP and local services is likely to be lifelong. For children the role of other professionals such as health visitors and paediatricians is likely to be considerable. In the case of children there will be issues around transition from services designed for their age group to those for adults and this needs to be carefully handled. There will need to be arrangements in place to ensure the needs and interests of children and families are properly considered and these must be sensitive to different ages. Children's circumstances change with the passing years and services need to be able to differentiate between and appropriately deal with young children, those who are older, teenagers and young adults, and this needs to be considered in relation to all the sections in this plan.

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.

Service Specification

Commissioning and planning systems for rare diseases will need to ensure equitable access to evidence-based 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 Welsh 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.

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.

Very rare conditions will best be managed and supported by a network of interested and committed clinicians from across the UK, none of whom are likely to see enough patients on a regular basis to meet the criteria for a specialist centre but who,

between them, can initially develop and then follow the appropriate best care pathways.

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.

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. Where patient organisations do not exist, the centre must consider other options for support.

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.

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. There is more on registers in section 5 on research and development.

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 at 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.

Actions

Who	Action	Timescale
Welsh Government	Address the issues around a UK registry (commitment 20)	By 30/11/14
	Ensure that Wales is involved as a member of the UK team that is leading on monitoring the development of ICD-11 in preparation for its adoption (commitments 16).	Ongoing
	Work with WHSSC and colleagues in Europe on the development of the European Orphanet in support of action to: - extend Orphanet listings from genetic testing services to include specialist clinical services; and, - foster better communication between primary and secondary care and support more focused referral using common websites such as Orphanet to improve diagnosis (commitment 17).	Ongoing
	Agree how, as part of work on a new Welsh health information strategy, to address commitments relating to: - standardising data collection and developing standards where they do not exist, increasing the reliability of information for use in providing or commissioning care (18); - exploring options to improve the link between existing patient data and electronic health records (19); - assess the potential for rare disease databases where they do not exist, including assessing the opportunity for extending the Welsh CARIS system (20); - improving systems to record genetic and other relevant information accurately to record the incidence and prevalence of disease and support service planning and international planning (29);	By 30/09/14

	- identifying how to change systems to hold information about rare diseases, including information about the uptake of treatments (30); and, - working across the 4 UK countries to develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways (31).	
	Agree with other UK countries how to move ahead on: - agreeing international standards, building on existing NHS standards (commitment 21); and - supporting international links to UK databases and building on the work of current funded programmes that aim to link rare disease research internationally. (commitment 22)	By 30/11/14
WHSSC	Develop as part of the process of developing the strategy for specialised services, 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 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; and, - work with the lead nation on the development of generic care pathway (commitment 23).	By 31/12/14 for preparation of the Plan with ongoing development activity throughout 2015
	Work with **LHBs** as providers and NHS England to: - develop and agree service specifications; - develop an agreed process for monitoring services against specification; and, - implement and monitor services against service specification including outcome and audit monitoring (commitment 24).	Ongoing
Welsh Rare Diseases Implementation Group	in conjunction with other interests and the other countries of the UK advise on how to: - 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 (commitment 25); - 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) (commitment 27);	Ongoing

	and, - 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 (commitment 28).	

Theme 5 The Role of Research

Improving Research on Rare Diseases

Effective Research and Development (R&D) is essential to improving health and services and there is growing evidence that clinical standards improve in an environment strongly influenced by research. Innovative healthcare is also associated with significant income generation, through the development and exploitation of intellectual property and attracting 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 supported by funding from Government and external bodies.

The Welsh Government's National Institute for Social Care and Health Research (NISCHR) supports efforts to streamline research processes, stimulate excellence and innovation and build research capacity. NISCHR currently funds a broad portfolio of programmes and initiatives of benefit to Wales' health and social care research community, notably (in the context of rare diseases) the Wales Gene Park, the Cancer Genetics Biomedical Research Unit and the Academic Health Science Collaboration. 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, which support the recruitment of patients to high quality research studies, including in rare diseases, in line with demand. Although some of NISCHR's current delivery structures will be changing from 1st April 2015, its commitment to continue to support and facilitate high quality research in areas of strength and to facilitate public and patient involvement in research remains as strong as ever.

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 in existing fora and collaborations, and to forge new ones to increase patient access to research and funding opportunities for Wales-based researchers. 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 aimed at making smarter use of routine data and the creation of a national Citizen's Cohort.

Involving Patients in 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. NISCHR follows INVOLVE definitions of public involvement, engagement and participation in research (http://www.invo.org.uk/) and supports a range of mechanisms designed to place the public at the heart of its funded activities and initiatives.

The Welsh Congenital Abnormalities database CARIS is a comprehensive reporting register that includes many rare congenital and childhood diseases stretching back to 1998CARIS is a member of EUROCAT, the European network of congenital anomaly registers. While EUROCAT restricts its activity to certain structural anomalies coded within the Q chapter of ICD-10, CARIS has a much broader outlook. In addition, basic disease-specific registries for Wales/the UK are maintained locally for several disorders. 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.

Actions

Who	Action	Timescale
NISCHR on behalf of the Welsh Government	Shape its actions in support of research around five key areas: - ensuring proportionate regulatory systems; - optimising engagement and involvement in research; - promoting research and public participation in research; - creating a receptive environment to facilitate industry collaborations; and - investing in infrastructure that facilitates research.	Ongoing
NISCHR	On a continuing basis: - work with the Research Ethics Committee for Wales and NHS R&D Offices in Wales to respond to UK-wide	Ongoing

	initiatives to develop risk-proportional permission systems (commitment 39); - Introduce engagement systems to encourage patient groups to get involved with regulatory bodies. (commitments 36); - provide support and advice to researchers and lay organisations to ensure the provision of high quality and accessible information for research participants and advice on appropriate mechanisms for engaging with and consenting research participants (commitments 8, 40); - work with all stakeholders to develop actions to streamline processes and reduce the time taken to approve and commence health studies in Wales (commitment 41); - 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 (commitment 46); - support the recruitment and training of patients, carers and service users and their engagement in research development and delivery (commitment 8); - work in partnership with NHS Wales to develop mechanisms to ensure that, where appropriate, research opportunities are maximised for patients with rare diseases (commitments 47); - in partnership with all stakeholders encourage and develop the kind of research collaborations required to remove the special barriers to the diagnosis, treatment and management of rare diseases (commitment 48); and, - ensure that appropriate funding for excess treatments costs (ETC) for research in rare diseases is accessible through the NISCHR ETC centralised budget (commitment 33).	
	Through the development of the new NISCHR research infrastructure including developments in the NHS – determine and outline how best to support a cohesive infrastructure for implementation and coordination of rare disease research (commitments 49).	By 31/03/15
	In line with Wales' ambition to put citizens at the heart of creating new knowledge: - enable service providers to involve patients in research (commitments 8, 33); - work with other stakeholders (NHS Wales, patient organisations, funding bodies, industry, researchers and clinicians) to consider how: existing disease registries can be best supported and promoted; sustainable data collection systems can be put in place; and patients of all ages can be supported by their clinical team to join disease registries, (commitment 7); - on a continuing basis, support patients to register on disease registries where they exist (commitment 7); - on a continuing basis, pursue initiatives that connect	On a continuing basis with an agreed way forward by 31/03/15

	patients affected with rare diseases and their families with opportunities to participate in relevant research (commitment 35); - working with the NIHR, explore the use of the UK Clinical Trials Gateway to provide information for patients and their families about research trials and bring forward recommendations by 31/03/15 (commitments 35, 38); - ensure that NISCHR's funding in support of research beyond 31/03/15 helps to create a new and stronger partnership between the population, research funders, service providers, industry and researchers, based on principles of co-ownership and co-production and transparency in health and social care research (commitment 51); and, - in developing its proposals to support research in Wales beyond 31/03/15, and in implementing them, continue to use its influence to ensure that appropriate funding for high quality research into rare diseases can be directed towards meeting the needs of Welsh patients and, as part of this, provide opportunities for NISCHR funding to be deployed in support of high quality research that benefits patients with rare diseases in areas of existing expertise and exceptional future potential (commitment 50).	
All-Wales Medical Genetics Service	Work with other UK nations and specialist and professional bodies (Clinical Genetics Society, British Society for Genetic Medicine and others) 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 (commitment 32).	Ongoing
	Work with WHSSC, other specialties, experts from higher education institutions 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 (commitment 32).	Ongoing
Wales Gene Park	Prepare with the Genetic Alliance and Rare Disease UK a plan for partnership, to help patient organisations and community engagement event organisers develop more formal partnerships with the NHS research-active organisations (commitment 37).	By 31/03/15
	Lead Welsh organisations involved in rare disease research in outlining their approach to supporting structured engagement and partnership with industry to drive research impact (commitment 46).	By 31/03/15

Annex: full list of commitments in the UK strategy and actions under this plan

	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.	See theme 1 – actions by LHBs; Welsh Government; WHSSC
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.	See theme 1 – actions by LHBs; WHSSC
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.	See theme 1 – action by LHBs
4.	Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.	See theme 1 – action by LHBs, WHSSC
5.	Consider how to give all patients with rare disease clear and timely information about their condition and its development; treatment and therapy options; practical support.	See theme 1 – actions by LHBs; Welsh Government
6.	Improve access for patients (or where appropriate their parents or guardians) to their personal data.	See theme 1 – action by LHBs
7.	Support patients to register on databases where they exist.	See theme 5 – actions by NISCHR
8.	Help patients to contribute to research and other activity related to rare diseases.	See theme 5 – actions by NISCHR
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.	See theme 2 – action by the Welsh Screening Committee
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.	See theme 3 – action by AWMGS, WHSSC

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.	See theme 3 – action by LHBs, WHSSC, AWMGS, Welsh Rare Diseases Implementation Group
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.	See theme 3 – action by LHBs, WHSSC, AWMGS, Welsh Rare Diseases Implementation Group
13.	Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.	See theme 3 – action by WHSSC, AWMGS
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	See theme 3 – action by Welsh Government
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.	See theme 3 – action by Welsh Government
16.	Monitor the development of ICD-11 in preparation for its adoption.	See theme 4 – action by Welsh Government

17.	Work with colleagues in Europe on the development of the European Orphanet coding system and in considering the adoption of Orphanet coding and nomenclature.	See theme 4 – action by Welsh Government
18.	Standardise data collection, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.	See theme 4 – action by Welsh Government
19.	Explore options to improve the link between existing patient data and electronic health records.	See theme 4 – actions by Welsh Government
20.	Assess the potential for rare disease databases where they do not exist.	See theme 4 – action by Welsh Government
21.	Agree international standards, building on existing NHS standards.	See theme 4 – action by Welsh Government
22.	Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally.	See theme 4 – action by Welsh Government
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 - 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.	See theme 4 – action by WHSSC
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.	See theme 4 – action by WHSSC, LHBs

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.	See theme 4 – action by Welsh Rare Diseases Implementation Group
26.	Set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support.	See theme 3 – action by AWMGS, WHSSC, LHBs
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 theme 4 – action by Welsh Rare Diseases Implementation Group
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 theme 4 – action by Welsh Rare Diseases Implementation Group
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.	See theme 4 – action by Welsh Government
30.	Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments.	See theme 4 – action by Welsh Government
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.	See theme 4 – action by Welsh Government
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.	See theme 5 – actions by AWMGS
33.	Examine how they can encourage service providers to involve patients in research and to ensure appropriate funding for excess treatment costs for research in rare diseases.	See theme 5 – actions by NISCHR
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.	See theme 1 – action by WHSSC

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.	See theme 5 – actions by NISCHR
36.	Encourage patient groups to get involved with regulatory bodies.	See theme 5 – action by NISCHR
37.	Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.	See theme 5 – action by the Welsh Gene Park
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.	See theme 5 – action by NISCHR
39.	Work with the research community, regulators, providers of NHS services and research funders to develop risk-proportional permission systems.	See theme 5 – action by NISCHR
40.	Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.	See theme 5 – action by NISCHR
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.	See theme 5 – action by NISCHR
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) and similar initiatives.	See theme 3 – action by Welsh Government
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 theme 3 – action by Welsh Government
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	See theme 3 – action by Welsh Government
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 theme 3 – action by Welsh Government

46.	Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.	See theme 5 – actions by the Welsh Gene Park and NISCHR
47.	Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.	See theme 1 – action by LHBs and theme 5 – action by NISCHR
48.	Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders.	See theme 5 – action by NISCHR
49.	Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.	See theme 5 – action by NISCHR
50.	Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.	See theme 5 – action by NISCHR
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).	See theme 5 – action by NISCHR