Department of Health, Social Services and Public Safety

Providing High Quality Care for people affected by Rare Diseases – *The Northern Ireland Implementation Plan for Rare Diseases*

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The Northern Ireland Rare Diseases Implementation Plan -

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Ministerial Foreword

This Northern Ireland Rare Diseases Implementation Plan sets out the commitment of my Department, the Health and Social Care sector, the voluntary sector, and education and research stakeholders to address the needs of people living with the impact of a rare disease regardless of their age, where they live and whatever their circumstances.

The experience of people living with a rare disease, their carers and wider family network, often provide the most challenging of circumstances to health and social care professionals involved in the diagnosis, treatment and care of these patients. At the same time, advancements made through screening, research and innovation have been and will continue to be at the edge of groundbreaking medical science with the potential for significant benefit for patients, families and the community, so that people may live their lives as ordinarily as possible.

My Department, Health and Social Care commissioners and providers, and the Northern Ireland Rare Disease Partnership (NIRDP), have worked together to ensure that service user and carer groups, who share our aim to make further progress in this important area, have been included in developing this implementation plan for rare diseases in Northern Ireland.

The plan identifies a range of actions to be taken forward in Northern Ireland during the period 2015-2021 in respect of the 51 Commitments in the UK Rare Diseases Strategy that the Health Departments from the four UK countries were pleased to endorse. These Commitments are set out under five key themes in the UK Strategy which together provide a holistic approach to caring for people with a rare disease and to maximise the available resources for research, innovation and collaboration across the UK to benefit the entire rare disease community.

The Northern Ireland Implementation plan for rare diseases has a sixth theme, setting out our commitment and associated actions which reflect the opportunity to work with our neighbours in the Republic of Ireland to realise the mutual benefits of cross-border collaboration on rare diseases.

This plan has taken into account the views of service users, carers, their wider families and stakeholders who participated in a consultation exercise which ran from 27 October 2014 until 19 January 2015. I am delighted that so many individuals and organisations responded to the consultation on the draft plan; their views and comments have been

invaluable in the production of this five year plan to drive forward improved services for those living with the impact of a rare disease.

Simon Hamilton MLA
Minister for Health, Social Services and Public Safety

Introduction by the Chief Medical Officer for Northern Ireland

This Northern Ireland Implementation Plan for Rare Diseases, referred to below as "the Northern Ireland Plan", has been published by the Department of Health, Social Services and Public Safety, referred to below as "the Department" or "DHSSPS", as a response to the 51 Commitments set out in the United Kingdom Strategy for Rare Diseases published in November 2013 referred to below as the "UK Strategy".

On 8 June 2009, the European Council issued a Recommendation on an Action in the field of rare diseases (2009/C 151/02), obliging Member States to put in place a plan or strategy on rare diseases by 2013 to integrate all current and future initiatives at local, regional and national levels in the field of rare diseases.

Following UK-wide public consultation on proposals for a UK Strategy for Rare Diseases, the Department of Health in London (DHL) published a summary of the consultation responses in November 2012. Following consideration of the responses a Working Group, chaired by DHL, finalised the UK Strategy for Rare Diseases, in conjunction with a Rare Diseases Stakeholder Forum, which included representatives from Northern Ireland. The UK Strategy was published in November 2013 following approval by the Health Ministers from the four countries of the UK who have agreed to work to achieve the Commitments in the UK Strategy by 2020.

Following publication of the UK Strategy, Scotland[1] and Wales[2] have published their respective Implementation Plans for Rare Diseases. This document is Northern Ireland's response. Each country is detailing how they plan to address the Strategy's 51 Commitments and strategically direct local services in future years. England is setting out its intentions across its high level frameworks and strategies[3].

The Department will work with the Health and Social Care sector, other Government Departments, local government, voluntary and community organisations, researchers and industry to progress these commitments for people living with a rare disease in Northern Ireland.

[1] It's not rare to have a rare disease, http://www.scotland.gov.uk/Publications/2014/07/4751/0

[2] http://www.wales.nhs.uk/sites3/documents_range/898/Agenda%20Item%2019%20Annex%20%28i%29%20rarediseaseplanen.pdf

[3] NHS Five Year Forward View, http://www.england.nhs.uk/wp-content/uploads/2014/10/5yfv-web.pdf

The Northern Ireland Plan has been drafted by the Department and been informed by the Northern Ireland Rare Diseases Stakeholder Group, an ad-hoc group established and chaired by the Department to help steer the development of the NI Plan, which comprises representatives from the:

- DHSSPS policy directorates with responsibility for policy related to rare diseases;
- Health and Social Care Board (HSCB) which is responsible for the planning, commissioning and delivery of health and social care services, and support to people living with a rare disease and their carers;
- Public Health Agency (PHA) which is responsible for health and social wellbeing improvement; health protection; public health support to commissioning and policy development and HSC research and development; and,
- Medical professions, universities and service user and carer groups who provide care for and conduct research into rare diseases and provide support to those affected by rare diseases.

The above stakeholders play key roles in the policy, planning, commissioning, delivery, research into rare diseases and the provision of support to people living with a rare disease.

The Northern Ireland Plan identifies short, medium and long term actions designed to improve services for people living with rare diseases in Northern Ireland during the period 2014 to 2020/21.

The actions address the five main objectives in the UK Strategy:

- empowering those affected by rare diseases;
- identifying and preventing rare diseases;
- diagnosis and early intervention;
- coordination of care, and
- the role of research.

The sixth theme of the Northern Ireland Plan focuses on the opportunities for cross-border collaboration on rare diseases initiatives with our neighbours in the Republic of Ireland.

This Northern Ireland Plan provides an agenda to progress the 51 Commitments in the UK Strategy in a way that takes account of our local Northern Ireland context whilst

encouraging and enabling us to learn and benefit from best practice in health and social care provision nationally and at the European and international levels. The Northern Ireland Plan also aims to ensure that Northern Ireland can contribute to and benefit from information sharing and research which are important disciplines for better understanding and developing preventative measures and treatments for rare diseases; and that our universities, clinicians, patients, researchers and industry can participate in and benefit from the medical research and development process across the UK in Europe and internationally.

Throughout the plan where rare disease is mentioned this should be taken to refer to all rare diseases both genetic and non-genetic. The term genetic is used when actions and comments are specifically for only genetic conditions.

Work Programme

Annex A provides priority actions for implementation requiring new investment in the initial work programme for the period 2015/16 and 2016/17. This work programme will be rolled forward by the Department annually within the context of available resources on a year basis to implement the overall plan. All actions set out below under the six activity themes will be progressed over the lifetime of this plan. Their implementation will mostly be funded from within existing DHSSPS related administrative and programme delivery budgets to be prioritised within the annual financial planning cycle. However, several initiatives will require new investment and these are identified in Annex A below together with the estimated cost. Implementation of these initiatives will therefore be subject to securing available funding and business case approval.

Dr Michael McBride

1. Strategic Context

A rare disease is a life-threatening or chronically debilitating disease, also referred to as an orphan disease which affects a small percentage of the population. There are between 6,000 and 8,000 rare diseases, and each one affects less than 0.1% of the UK's population.

The European Union's definition of a rare disease is one that affects not more than 5 per 10,000 persons in the European Union. The European Council's estimate that one in seventeen of the population will be affected by rare disease leads to a figure of approximately 106,000 people affected by rare disease in Northern Ireland.

80% of rare diseases are genetic; the remainder are non-genetic and include conditions arising from a wide variety of causes, including some rare forms of cancer. Genetic illnesses present throughout the person's entire life, however, for some non-genetic conditions, there are possibilities of a cure.

The Northern Ireland Plan aligns with the Northern Ireland Executive's Programme for Government's[4] priority which seeks to address the challenges of disadvantage and inequality that afflict society and the relatively poor health and long-term shorter life expectancy of our population.

The Northern Ireland Plan also aligns with 'Transforming Your Care' (TYC)[5] which describes a new model for the delivery of integrated health and social care services in Northern Ireland focused on prevention initiatives and earlier interventions, and on promoting health and well-being. TYC also highlights that more services should be provided in the community, closer to people's homes where possible and that there should be more personalised care. TYC also highlights that services which have such a low volume that they cannot be sustained to a high quality in Northern Ireland, even

[4] Priority 2: "Creating opportunities, tackling disadvantage and improving health and wellbeing"; in the Northern Ireland Executive's Programme for Government (2011-15)

[5] 'Transforming Your Care' (TYC) (2011)

without networking to other tertiary centres, should continue to be delivered outside of Northern Ireland. These include for example a range of organ transplantations and some highly specialist elements of rare disease management.

Therefore there is a clear link between this Northern Ireland Plan and the related strategic policies of the NI Executive and DHSSPS which support the vision and mission in this plan.[6]

2. Vision

Our vision is to ensure that people living with a rare disease have access to the best evidence-based healthcare, treatment and life opportunities that Northern Ireland can provide.

The Department fully supports the Vision in the UK Strategy. 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;

[6] Quality 2020 – 10 year strategy to protect and improve quality in Health and Social Care
Making Life Better – A whole system framework for Public Health 2013-2023
Equality, good Relations and Human Rights Strategy and Action Plan
NI Children and Young Peoples Strategy 2006-2016
Physical and Sensory Disability Strategy
Living with Long Term Conditions
Who Cares – Future of Adult Support
10 year Strategy for Social Work in Northern Ireland 2010-2020
Strategy for Allied health Professionals 2011-16

- 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.

3. Mission

Our mission is, within available resources, to further improve the delivery of health and social care services and support for people living with the impact of a rare disease, aiming to ensure that these are of high quality, are safe and sustainable.

4. Why Providing High Quality Healthcare for Patients Living with

Rare Disease is a Priority

The UK Strategy aims to ensure no one gets left behind just because they have a rare disease. The Health Ministers from the four countries have clearly stated that they want to put the patients' needs first. To do this, they have committed to bring together the talent, skills and professionalism of all relevant sectors. This will bring real, positive change in how we deal with rare diseases and how we help people living with complex conditions.

5. Our Journey So Far

The UK 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 health and social care organisations, 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 or high level strategies 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, aiming to improve access to information, services and research and to bring real, positive change to the lives of people affected by rare diseases and their families.

At present, generally services for people living with a rare disease in Northern Ireland are provided within the Health and Social Care sector by relevant medical specialties depending on the particular symptoms they have. There are no specialist treatment units dedicated to individual rare diseases. Patients may, therefore, be treated in a number of different settings across Northern Ireland's Health and Social Care services. As the majority of rare diseases are genetic-based, services are also provided by the Northern Ireland Regional Genetics Service. In addition, where appropriate, the Health and Social Care Board will consider funding the referral of patients for specialist services elsewhere in the UK, Republic of Ireland, or further afield.

6. What We Want to Achieve

Given the unique nature of each of the diseases concerned and the relatively small numbers of people affected by each individual condition in Northern Ireland, we envisage that the way forward will be to work to improve cooperation in terms of diagnosis, treatment, codification, research, development of specialised drugs and centres of expertise between policy makers across Government Departments, Health and Social Care, clinicians, universities and industry, in Northern Ireland and with our counterparts in the rest of the UK, the Republic of Ireland, the rest of the EU, and internationally.

7. Working Together

The Northern Ireland Plan promotes a partnership approach working to ensure that the HSC and all stakeholders work together to understand and address the needs and experiences of patients, their families and carers by supporting people to manage their own condition within their individual set of circumstances.

8. Measuring Success

The Department proposes to prioritise the implementation of the 'Actions to be Taken', set out under the six themes below, within the context of the Department's annual Business Plan, the annual Commissioning Plan Direction, business case approval, available resources and existing accountability and governance responsibilities within the HSC in Northern Ireland. The Department proposes to publish an annual statement reporting progress in implementing the 'Actions to be taken'.

9. Equality Screening

Human Rights and Equality Implications

Section 75 of the Northern Ireland Act 1998 requires Departments in carrying out their functions relating to Northern Ireland to have due regard to the need to promote equality of opportunity:

- between persons of different religious belief, political opinion, racial group, age, marital status or sexual orientation;
- between men and women generally;
- between persons with a disability and those without; and
- between persons with dependants and those without.

In addition, without prejudice to the above obligation, Departments should also, in carrying out their functions relating to Northern Ireland, have due regard to the desirability of promoting good relations between persons of different religious belief, political opinion or racial group. Departments also have a statutory duty to ensure that their decisions and actions are compatible with the European Convention on Human Rights and to act in accordance with these rights.

DHSSPS has carried out a preliminary screening of the Plan and as part of this screening process has concluded that an Equality Impact Assessment is not necessary.

Human Rights

Article 8 of the European Convention on Human Rights guarantees a right to privacy which can only be interfered with when it is necessary to meet specified legitimate needs. The Department recognises that the proposals in relation to patient databases will broaden the use of personal information provided to the HSC, but would stress that this will only be considered in prescribed conditions, and in circumstances which clearly have a legitimate need and a social benefit, and when supporting legislation is in place.

Privacy

The Department acknowledges that the proposals in relation to sharing of patient information may raise concerns but believes that the societal benefits, required safeguards and control of access should mitigate the concerns and risks involved.

Rural Proofing

Due to the small number of patients who are affected by each individual rare disease and the highly specialised expertise required to treat them services tend to be provided on a regional basis within Northern Ireland or supra regionally within the UK or further afield. In this context, the whole of Northern Ireland suffers from geographic isolation. Patients in rural areas may have more difficulty in travelling to receive their treatment but the implementation plan aims to deliver better quality services for all those affected by a rare disease and to reduce the administrative and logistical barriers to highly specialised diagnosis, assessment, and treatment.

Health Impact

It is considered that these proposals will have a positive impact on the health of all those affected by a rare disease, and go some way to address the imbalance between the care provided to these people and the general population which was identified through the development of the UK Strategy for Rare Diseases.

Sustainable Development

It is considered that there are no negative impacts on sustainable development opportunities. Better health outcomes for people living with rare diseases may, in the longer term, result in added social and economic benefits. Improved opportunities for Northern Ireland universities and industry to participate in research and innovation may lead to additional economic benefits, as recognised in the UK Strategy.

Regulatory Impact Assessment

The Department does not consider that a Regulatory Impact Assessment is required as the Implementation Plan does not have any adverse regulatory impact on local business.

The Northern Ireland Rare Diseases Implementation Plan

Theme 1: Empowering those affected by rare diseases

The UK Strategy provides an opportunity for people living with a rare disease, their families, carers and representatives (the stakeholders) to increase the role they can play in a) the day-to-day management of specific conditions and b) to influence the development of services which are of benefit to patients.

Empowered patients, who are engaged in their treatment and in decision-making, tend to be more responsive to treatment. Increased involvement of patients and their families can realise benefits in terms of resources and improved outcomes. One aspect of empowerment is the availability of relevant information which is provided to individuals to make them aware of the choices available to them and to help them to take balanced decisions. Another aspect is to show that their views are important and to provide suitable mechanisms for engagement of clinicians and policy makers with patients or their representatives.

Another aspect of the plan which incorporates aspects of empowerment is the need for this plan and related strategic policies of the NI Executive, the DHSSPS and other Departments to support the actions and vision of this plan to provide a holistic approach to meet the needs of those impacted by rare disease.

The Department and the HSC in Northern Ireland are committed to listening to the views of patients and stakeholders who have first-hand experience of the services we provide in order to further develop and enhance services which meet patients' needs and expectations. While the Department and the HSC want to afford every opportunity to learn about the needs of individual patients, this is not always possible due to constraints on resources and the time which the HSC community can make available. Therefore we are committed to ensuring that patients' representatives can play a full and meaningful role on their behalf in having access to the Department and HSC to ensure that patients' views and needs are properly represented.

We propose to further secure this engagement by building on our existing strong working relationship with the Northern Ireland Rare Disease Partnership (NIRDP). The

NIRDP was established in March 2011 by a group of organisations representing people living with rare diseases, representatives of the Northern Ireland Regional Medical Genetics Service and Rare Diseases UK (RDUK). It brings together those living with a rare disease and organisations representing them; clinicians and other health professionals; researchers and producers of specialist medicines and equipment; health policy makers, and academics.

The roles of the NIRDP are as advocates, educators, innovators and supporters of people affected by a rare disease, their dependents and carers.

Consultation avenues are already in place with NIRDP, Rare Diseases UK, the Rare Disease Advisory Group (a four nations group supported by NHS Commissioning and the Department of Health England), the UK Rare Disease Forum (which is established on a UK-wide basis by the Department of Health England as the UK Lead Department for Health matters internationally) and other key interest groups to ensure that the views of people living with a rare disease are taken into account in policy development and in the planning and delivery of services. The HSCB/PHA meets with NIRDP at key stages in the commissioning cycle to inform key priorities for this group of patients and the annual HSCB /PHA Commissioning Plan. The HSCB and PHA will continue to work in partnership with NIRDP and other key interest groups to ensure that feedback from service users and carers underpins the identification of the commissioning priorities for rare diseases. This will include involving patients in the development of service models and service planning, and in the evaluation and monitoring of service changes or improvements. DHSSPS will seek to raise awareness of the need to liaise with patients living with rare disease with other Government Departments.

The Department, HSCB and PHA will continue to participate in NIRDP events and support them to provide opportunities for people living with rare diseases and their carers to give a first-hand account of the impact that rare diseases have on their lives.

In addition to engagement with the NIRDP, wider consultation will be facilitated when significant changes to services for the rare disease community are being explored.

The following Commitments and Actions to be taken are designed to provide patients suffering from a rare disease and their representatives the opportunity to influence the development of services over the period of the final NI Plan to 2020/21 and beyond.

UK Strategy Commitment		**Actions to be taken**	**Completion Date**	**Owner**
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 – including in the development of the four country plans to implement the Strategy.	• 1.1 The Department and HSC will maintain and enhance the existing avenues of communication with key stakeholders for people living with the impact of a rare disease and their representatives. • 1.1.a. Complete a communications review and introduce enhanced methods of communication.	Monitoring and progress reports to be published annually no later than 30 June. 30/06/2016	DHSSPS DHSSPS
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.	• 2.1 The HSCB and PHA through their membership of the UK Rare Disease Advisory Group (RDAG) will work with other nations to develop care pathways and protocols to support referrals to highly specialist centres. This will provide a better understanding of the needs of patients being cared for with a rare disease. New learning from this will be incorporated into HSCB/PHA commissioning processes. • 2.2 The Department will incorporate awareness of rare	Monitoring and progress reports to be published annually no later than 30 June. See Action 15.1	HSCB PHA DHSSPS

		diseases into training for its clinicians and service providers		
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.	• 3.1. Within the commissioning planning cycle (2015/16 Commissioning Plan) the HSCB and PHA will continue to work on improving the patient experience for those patients travelling outside Northern Ireland for specialist care. • 3.2. DHSSPS will promote to other NI Executive Departments the need to liaise with patients living with rare disease in order to understand and respond to their needs.	Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA DHSSPS
4	Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation	• 4.1.During the period 2015 to 2020/21 the HSCB and PHA, as members of the RDAG, will participate in the development of	Monitoring and progress reports to be published annually no later than 30	HSCB PHA

	and monitoring of services.	service specifications and pathways for specialist care and will require providers of specialist care to demonstrate their consideration of the effects of rare diseases on people's lives.	June.	
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; and practical support.	• 5.1 The HSCB and PHA through working with the NIRDP will develop a model of support for people with rare diseases and their families to access services and information.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
6	Improve access for patients (or where appropriate their parents or guardians) to their personal data.	• 6.1 The Department will work with the HSC to assess the existing Northern Ireland Electronic Care Records (NIECR) to explore the potential to contain appropriate information on rare diseases. • 6.2 The Department will review its current policy on patients having access to their own data to establish whether this needs to be customised to meet the requirements of patients with a rare disease.	Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS DHSSPS
7	Support patients to register on databases, where these exist.	• 7.1 This Commitment will be taken forward in conjunction with the Actions for Commitment 6.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS

8	Help patients to contribute to research and other activity related to rare diseases.	• 8.1 The HSC Research and Development Directorate will seek to identify patients with rare diseases, their carers and representatives who would wish to join the HSC Research and Development Personal and Public Involvement Panel.	Monitoring and progress reports to be published annually no later than 30 June.	PHA

Theme 2: Identifying and preventing rare diseases

The UK Strategy for Rare Diseases highlights the World Health Assembly resolution in 2010[7] which emphasised that birth defects are still significant and that early diagnosis and action can prevent complications and illness.

Many rare diseases are present at birth and are either caused by a genetic problem or deficiencies or exposures to substances around the time of conception or during pregnancy. Non-genetic rare diseases present at different stages of life.

For some rare diseases screening programmes have an important role to play in achieving early identification and intervention. Screening refers to the application of a medical procedure or test for people who as yet have no symptoms of a particular disease, for the purpose of determining their likelihood of having the disease. The goal of screening is to reduce morbidity or mortality from the disease by detecting diseases in their earliest stages, when treatment is usually more successful.

The UK National Screening Committee (UK NSC) advises Ministers and the NHS in the four UK countries about all aspects of screening and supports implementation of screening programmes.

Using research evidence, pilot programmes and economic evaluation, it assesses the evidence for programmes against a set of internationally recognised criteria covering the condition, the test, the treatment options and the effectiveness and acceptability of

[7] http://apps.whoint/gb/ebwha/pdf_files/WHA63/A63_R17-en.pdf

the screening programme. The UK NSC regularly reviews policy on screening for different conditions in the light of new research evidence becoming available.

Northern Ireland has two representatives on the UK NSC. Since its inception, the UK NSC has developed policies on screening for a range of conditions and has overseen the successful introduction of a number of national screening programmes including the addition of new conditions to the newborn blood spot screening programme.

The following Commitments and Actions to be taken will serve to continue and strengthen existing arrangements in Northern Ireland to deliver the benefits through early identification and prevention of rare disease. Commitments 9 and 10 facilitate Northern Ireland's representation and participation in national screening programmes and on the UK Genetic Testing Network.

UK Strategy Commitment		**Actions to be taken**	**Completion Date**	**Owner**
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.	• 9.1 The Department and the Director of Public Health, PHA, will continue to be represented on this Committee. • 9.2. A Working Group on Primary Prevention of Congenital Anomalies, including all relevant stakeholders should consider the recommendations of EUROCAT / EUROPLAN in Northern Ireland.	Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS PHA PHA
10	Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular tests are evaluated and recommended by UK Genetic Testing Network (UKGTN), is	• 10.1 The HSCB will aim to secure routine access for patients and their families (where appropriate) to UKGTN approved panel tests on much the same basis as that provided for National Institute for Health and Care Excellence	Monitoring and progress reports to be published annually no later than 30 June.	HSCB

	accessible for 'at risk' relatives.	(NICE) recommended Technical Appraisals (TAs), subject to available funding.		

Theme 3: Diagnosis and early intervention

Early diagnosis of a rare disease can allow for timely intervention which can, in turn, provide better outcomes for patients. Many GPs will never see an example of many of the conditions which are termed collectively as rare diseases. Whilst training on rare diseases is provided at undergraduate and postgraduate levels for doctors, the nature of a rare disease is such that the symptoms which present are common with other conditions; this can cloud the issue and add difficulty in coming to a timely, conclusive diagnosis. In many cases the rare disease can only be identified by a person with specialised expertise and laboratory tests after many alternative avenues have been explored.

The Department believes that the effective diagnosis and intervention in the treatment of rare diseases can be further strengthened in Northern Ireland by ensuring that the medical workforce is effectively trained in this field and given access to robust information and patient screening programmes in order to improve overall awareness.

All medical specialties and multi-professional care teams should therefore have a general awareness of rare diseases, so that they can make rapid referrals to specialists in the appropriate field. Common protocols to identify patients at risk of rare diseases, or who have no diagnosis should also be in place.

In terms of training, there is already some emphasis on training in rare diseases in Northern Ireland. For example, the Queen's University medical school provides teaching on rare diseases and the Royal College of Nursing (RCN) includes the subject in the nurse training curriculum.

There is the potential for further progress to be achieved through the Modernising Scientific Careers (MSC), an acclaimed UK-wide government initiative to address the training and education needs of the whole healthcare science workforce in the National Health Service (NHS). This aims to introduce flexibility, sustainability and modern career pathways for the healthcare science workforce to meet future needs of the

Health Service. It sets out an education, training and career framework for all healthcare scientists, including a strong focus on genetics.

In respect of providing access to information in Northern Ireland, the existing data collection system (NIECR) runs on software provided by Orion Health on secure servers in the HSC data centres and is available to authorised users for the delivery of care throughout the HSC and all GP practices across Northern Ireland. The potential for strengthening this system, to collate information and provide clinicians with a comprehensive history of treatment and facilitate more informed decision-making in a shorter period of time, should be considered.

There is excellent potential in Northern Ireland to consider establishing rare disease database(s) where they do not exist. Expertise and experience in epidemiology, statistics, informatics, genetics, data management, and data protection are all readily available within Northern Ireland. Consideration should therefore be given to the potential for Northern Ireland to develop a comprehensive rare disease registry and information hub that could potentially set high standards within the UK and beyond.

The effective use of screening, carrier testing and cascade testing for severe genetic conditions can all help to reduce the impact of a disease by escalating the point at which a rare disease is identified. The advent of genomics offers new possibilities for the diagnosis process. We need to ensure that where possible speedy diagnosis can be provided for people with a rare disease, both genetic and non-genetic.

The HSCB and PHA will continue to secure access to effective clinical and social care for people with rare diseases including NICE Technology Appraisals and Highly Specialised Technology evaluations (recommendations on the use of new and existing highly specialised medicines and treatments within the NHS) and UKGTN tests.

The following Commitments and Actions are aimed at speeding up the time taken to come to a definitive diagnosis and having a positive impact on the current gaps between the first symptoms being noted and diagnosis of a patient's condition.

UK Strategy Commitment		Actions to be taken	Completion Date	Owner
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.	• 11.1 The HSC will aim to ensure that Northern Ireland patients receive test results in a timely manner comparable with best practice in other parts of the UK and to ensure that testing is available through the extra contractual referral (ECR) process for patients whose condition is so rare that tests need to be sent abroad to specialist laboratories.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB
		• 11.2 Where new genetic tests are endorsed by the UKGTN, HSC service providers will aim to work with the Regional Genetics Service to ensure undiagnosed patients can access testing where appropriate. The Genetics Service itself routinely reviews patients who have no diagnosis to make sure they can avail of new technologies.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB
		• 11.3 The HSCB will aim to secure routine access for patients and their families (where appropriate) to UKGTN approved panel tests on much the same basis as that provided for NICE recommended Technical Appraisals (TAs). This is subject to business case approval and available funding.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB
12	Work with the NHS and clinicians to establish appropriate diagnostic pathways which: - are accessible to,	• 12.1 The HSCB and PHA will take into consideration, in relation to service provision in Northern Ireland, the Generic Rare Disease Annex for all service specifications which is being	Monitoring and progress reports to be published annually no later than	HSCB PHA

	and understood by, professionals and patients, by - establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and 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	developed by NHS England. • 12.2 The HSCB and PHA will work with other UK nations through the Rare Disease Advisory Group to develop care pathways and protocols to support referrals to specialist centres. • 12.3 The HSCB and PHA have identified genetic services as a priority in making progress with the NI Rare Disease Implementation Plan. A sub group was established in 2014 specifically to develop a service specification for medical and clinical genetics. When completed this will deliver a documented description of the services, including: - care pathways; - service model; - service user / carer information; - referral processes / criteria; - interdependencies with other services; - service standards including test reporting times; and, - key service outcomes.	30 June. Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA HSCB

	diagnostic studies.			
13	Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.	• 13.1 The HSCB and PHA will participate in processes to be established by the UK Rare Disease Advisory Group for commissioning highly specialised services.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
		• 13.2 The HSCB and PHA will, where appropriate, continue to take account of the specific treatment needs of patients with a rare disease. This is supported through their internal processes for Extra Contractual Referrals (ECR) /Individual Funding Requests (IFR) and arrangements for the managed entry of new medicines.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
		• 13.3 As highlighted in C11 above the HSCB will aim to secure routine access for patients and their families, (where appropriate) to UKGTN approved panel tests on much the same basis as that provided for NICE recommended Technical Appraisals (TAs). This is subject to business case approval and available funding.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB
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.	• 14.1 A new system called Experior Medical is currently being trialled to support specialist learning in radiology. As part of the Northern Ireland eStrategy, consideration will be given by the HSC to incorporate rare diseases into this or a similar type of platform.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB

15	Improve education and awareness of rare diseases across the 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.	• 15.1 The Department will work with the Northern Ireland Medical and Dental Training Agency (NIMDTA), universities, Royal Colleges and patient groups to deliver appropriate training. • 15.1.a. – Complete a scoping exercise (training needs analysis) and produce a training action plan Estimated cost £20K. • 15.2 The Department will consider committing HSC funding for implementation of the Modernising Scientific Careers (MSC) initiative in Northern Ireland subject to available resources. • 15.3 Health Education England (HEE) is establishing a group to set out education / training pathways on which Northern Ireland is represented. Consideration will also be given to committing HSC funding for implementing its recommendations subject to available resources.	Monitoring and progress reports to be published annually no later than 30 June. 30/06/2016 Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS DHSSPS DHSSPS DHSSPS
16	Monitor the development of ICD-11 in preparation for its adoption.	• 16.1 This is a commitment which has a UK-wide application. The Department will liaise where necessary with other UK Health Departments in associated development work.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
17	Work with colleagues in	• 17.1 This is a commitment which has a UK-wide	Monitoring and	DHSSPS

	Europe on the development of the European Orphanet coding system and considering the adoption of Orphanet coding and nomenclature.	application. The Department will liaise where necessary with other UK Health Departments in associated development work.	progress reports to be published annually no later than 30 June.	
18	Standardise data collection, building upon existing NHS data standards, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.	• 18.1 This is a commitment which has a UK-wide application. The Department will liaise where necessary with other UK Health Departments in associated development work.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
19	Explore options to improve the link between existing patient data and electronic health records.	• 19.1 See Commitment 6. The Department will explore whether the NIECR system might be developed (within the context of provision for rare diseases patients) to give HSC professionals a single, secure overview of key information about their patients or clients in order to aid better, faster, safer decision-making and improve the quality and efficiency of HSC services.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
20	Assess the potential for rare disease databases where they do not exist.	• 20.1 The Department will work with all stakeholders to improve coordination of existing and additional rare disease registries/resources/databases to facilitate the creation of a Northern Ireland register for Rare Diseases. • 20.1.a. – Complete a database review and produce a fully costed action plan to implement a	Monitoring and progress reports to be published annually no later than 30 June. 30/06/2016	DHSSPS DHSSPS

		Northern Ireland register for Rare Diseases. Estimated cost £20K. • 20.2 In conjunction with the development of supporting legislation (expected in 2016), the Department will develop policy guidance to cover the use of rare disease databases.	31/12/2016	DHSSPS
21	Agree international standards, building on existing NHS standards.	• 21.1 This is a commitment which has a UK-wide application. The Department will liaise where necessary with other UK Health Departments in associated development work.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
22	Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally.	• 22.1. This is a commitment which has a UK-wide application. The Department will liaise where necessary with other UK Health Departments in associated development work.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS

Theme 4: Coordination of Care

The Health and Social Care sector in Northern Ireland is committed to ensuring that the diagnosis and care provided for people with rare diseases is effectively coordinated and connected at local, regional, national and international levels.

A key premise in commissioning health services for Northern Ireland is providing local services where possible and seeking specialist treatment where this is not available in Northern Ireland; this will be a key driver in shaping the commissioning profile for specialist care. With a population of only 1.8m Northern Ireland is however unlikely to ever be in a position to independently support highly specialist elements of care for rare diseases locally and will need to continue to link with

tertiary and quaternary centres in Great Britain (GB) and elsewhere to secure access to the full range of care needed.

Northern Ireland already has a strong relationship with many of the specialist units in GB and there are many examples of innovative models of 'shared' care ranging from visiting 'in reach' services to inclusion of Northern Ireland clinicians in regular Multi-Disciplinary Teams (MDTs) with specialist centres. This supports care for patients locally and also helps enhance the skills and resilience in local clinical teams.

Work undertaken in the last few years, particularly in the area of specialist paediatric services has allowed for these links to be strengthened. A Network Manager for specialist paediatrics was appointed in September 2012 to review current referrals to specialist providers and to develop and document opportunities for clinical engagement and networking where possible.

The multi-disciplinary approach to deliver effective care to people living with a rare disease requires high quality communication, coordination and planning skills to ensure that patients receive the right treatment at the right time and to ensure they are fully involved in the determination of an appropriate care plan which meets their needs.

Work has also commenced in developing service specifications for specialist paediatric care to allow more focused commissioning and clearer direction to providers regarding HSCB/PHA expectations and standards for specialist care. There are already a number of initiatives ongoing which support the care pathway and transition issues including those highlighted under Commitment 5 above.

Health and Social Care recognises the importance of documented pathways to guide appropriate and timely diagnosis and onward referral to services. We use this theme to consider the multi-disciplinary approach to well coordinated care where several specialists and hospital areas work together to provide a complete care package for patients.

It is especially important to ensure care provided by different service areas complement each other and provide a package which uses resources effectively to provide the best outcomes and meets the needs of individuals. The decision-making process requires input from appropriate health care professionals and patients.

Treatment provided by primary, secondary, tertiary and quaternary health care services should deliver services through a joined-up approach. The services of specialist centres (either virtually or remotely) should be available to allow patients to be referred in a timely manner and minimise delays in treatment.

In the last three years there has been significant investment in specialist paediatric services. There are less than 0.4m children aged 0 to 15 years in Northern Ireland which creates significant challenges in providing specialist care for relatively very small numbers of children with rare or specialist conditions. Initial investments aimed to deliver resilience to the existing services, with a number of sub-specialty areas only able to sustain one or two person consultant teams. Efforts have focussed on increasing staffing where it is sustainable to do so, building formal network links with clinical colleagues in GB, establishing joint MDTs meetings and clinical case conferences, and participation in clinical audit and peer review.

Work commenced in 2012 within the area of specialist paediatrics which included the appointment of a dedicated Network Manager to review current specialist care for children and develop network arrangements between the regional children's hospital in Belfast and paediatric services in other Trust areas. Clinical links with paediatricians across Northern Ireland are now in place for specialist gastroenterology and epilepsy. Referral protocols, pathways and service standards are under development. It is expected that the output of this work will establish useful templates with components which will be generically applicable to a number of conditions.

More of the clinical linkages between specialist and local care are planned and the model will be further supported by the Department's 'Paediatric Hospital and Community Based Services Strategy' which is in the final stages of development.

The following Commitments and Actions aim to promote and support the coordination of care for people with a rare disease.

UK Strategy Commitment		Actions to be taken	Completion Date	Owner
23	Continue to develop service specifications for rare diseases. This will include country specific care pathways and a 'generic' care	• 23.1 As a member of the Rare Disease Advisory Group (RDAG), HSCB and PHA will actively participate in	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA

	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, and - access criteria and measures of quality and outcomes.	confirming the service standards, developing commissioning specifications for highly specialist care and agreeing the accreditation of individual units as specialist providers by service areas. Pathways for referral and care for specialist providers will also be agreed through this process. These will then be linked as appropriate to local pathways for Northern Ireland informing what, if any, elements of care can be provided locally, for example, diagnostics, work ups, follow up work and ongoing care. • 23.2 The HSCB and PHA will further develop clinical links across Northern Ireland and with GB to identify separate commissioning specifications for specialist services for adults as required. • 23.3 The HSCB and PHA will take into consideration the Generic Rare Disease Annex for all service specifications which is being developed	Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA HSCB/PHA

		by NHS England.		
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, and - ensure their expertise is available to families and their healthcare teams.	• 24.1 The Department will liaise with other UK Health Departments in development work related to this commitment as it is more likely to have a UK-wide application. • 24.2 The HSCB and PHA will continue to progress a process mapping exercise for patients with chronic conditions moving from paediatric to adult care. In 2014/15 funding was made available to support a metabolic and genetics nurse specialist to support transition of young people to adult services. • 24.3 As a member of the UK Rare Diseases Advisory Group (RDAG), the HSCB and PHA will establish the parameters within which highly specialist care can be delivered. The HSCB and PHA will commission services only from those providers able to meet the standards established by RDAG and identified as national centres of expertise for	Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS HSCB PHA HSCB PHA

		specified services. • 24.4 See Commitment 23 above, final bullet point.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
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.	• 25.1 The Northern Ireland response to Modernising Scientific Careers includes proposed training for senior healthcare scientists to support communication of knowledge to patients. As with Commitment 24, however, this commitment is more likely to have a UK-wide application and the Department will liaise with other UK Health Departments in related	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS

		development work.		
26	Set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support.	• 26.1 Patients with rare diseases will continue to be referred to specialist centres in line with either the agreed pathway for that condition and/or the Extra Contractual Referral process.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB
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).	• 27.1 The Department will encourage HSC employers to address the need for allocated clinical staff time for research.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
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.	• 28.1 See Commitment 27 above.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
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.	• 29.1 See Commitment 20 above.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS

30	Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments.	• 30.1 See Commitments 20 and 29 above. • 30.2 The Department will work with HSC organisations to explore how this Commitment can be achieved by building on the potential sources of information provided by the HSCB's Extra Contractual Referral process and the UK Genetic Testing monitoring of molecular testing rates.	Monitoring and progress reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS DHSSPS

Theme 5: The role of research

The role of research into rare diseases has a UK-wide dimension and we have used the above theme to suggest opportunities for Northern Ireland based research to contribute to international understanding of rare diseases. The following Actions therefore aim to strengthen Northern Ireland's: contribution to UK-wide policy; and, the capacity to build on Northern Ireland's existing research base.

With regard to the research role of specialist clinical centres, this is already included in the Regional Genetics Service draft service specification and is part of its practice. Clinical Research Networks in place in Northern Ireland are compatible with networks across the UK. Northern Ireland's stable population, tightly knit structure and local expertise makes it an attractive option for a base for rare disease research. In addition, opportunities exist for Northern Ireland based academic and clinical researchers to participate in research with colleagues from across the UK or globally.

The Republic of Ireland Health Research Board also offers funding in partnership with charities, many of which support rare disease patients and carers. Funding for projects may include research partners in Northern Ireland. The HSC Research & Development Division (PHA) also offers funding in partnership with several charities and the opportunity exists to do so with 'rare disease' charities.

With regard to engagement with industry on research for rare diseases, industry engages with research funding organisations on a UK-wide or European basis. Northern Ireland based academic and clinical researchers are encouraged and supported to participate in relevant networks. The HSC R&D Division provides support for HSC researchers to work with industry colleagues, for example through research infrastructure such as the Northern Ireland Clinical Research Networks and Northern Ireland Clinical Research Facility, and ensures any intellectual contribution made by HSC researchers through collaborative projects is captured and protected through the HSC Innovations service.

Northern Ireland based clinicians and researchers are encouraged to participate in UK and international networks and centres. In particular, Northern Ireland based oncologists are leading the development of international (UK, Europe and US) clinical trials of potentially beneficial treatments for certain rare cancers. Although at present many people with rare diseases cannot participate in clinical trials, as they do not have sufficient follow-up data for inclusion, the unique distribution of services and electronic datasets in Northern Ireland offers good scope in the future for fully integrated population-based information and research.

The following Commitments and Actions are aimed at promoting and supporting the role of research in the field of rare diseases.

UK Strategy Commitment		**Actions to be taken**	**Completion Date**	**Owner**
31	Look at how the 4 UK countries develop, change or expand information systems to capture, connect and analyse data about	• 31.1 This is a commitment which has a UK-wide application. The Department will liaise where necessary with other UK Health	Monitoring and progress reports to be published annually no later than 30	DHSSPS

	clinical and social care pathways.	Departments in associated development work.	June.	
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.	• 32.1 See Commitments 23 and 24.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
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.	• 33.1 Costing and funding mechanisms for clinical research are in place in Northern Ireland to enable participation in UK-wide studies. The HSC will explore how these can be further developed.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
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.	• 34.1 The HSCB and PHA will consider and adapt, where appropriate, the prototypes to be developed by NHS England.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
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.	• 35.1 The HSC will work with patients, their relatives, clinicians and patient groups to determine how this commitment may be taken forward in Northern Ireland, taking account of the size of our population, based on the best	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA

		possible advice and support from clinical and academic researchers.		
36	Encourage patient groups to get involved with regulatory bodies.	• 36.1 The Department will work with the Northern Ireland Rare Disease Partnership, and other patient groups, to support their involvement with regulatory bodies.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
37	Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.	• 37.1 The Department will support patient organisations to build upon their existing engagement with the HSC and NHS research-active organisations and develop this further.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
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.	• 38.1 The Department will work with Northern Ireland stakeholders to build upon the existing UK Clinical Trials Gateway, which includes clinical trials that recruit patients from and in Northern Ireland, and explore the inclusion of experimental medicine (EM) trials.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
39	Work with the research community, regulators, providers of NHS services and research	• 39.1 The Department will work with UK-wide partners to further	Monitoring and progress reports to be published	DHSSPS

	funders to develop risk-proportional permission systems.	develop risk-proportional permission systems.	annually no later than 30 June.	
40	Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.	• 40.1 The Department will encourage local researchers to build upon and develop the existing guidance process in parallel with practice UK-wide.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
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.	• 41.1 The Department will encourage local researchers to promote good practice in relation to this Commitment in parallel with practice UK-wide.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
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.	• 42.1 The HSCB and PHA will contribute as required to this Commitment and will look to the Northern Ireland Regional Genetics Service to interface with national assessment bodies to inform Northern Ireland service configurations and referral pathways.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA
43	Evaluate different NGS platform configurations, for example: - NGS for clinical condition-specific sets of genes (such as 100–200 of the 22,000	• 43.1 See Commitment 42.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA

	genes) - whole exome sequencing (2% of the entire genome) - whole genome sequencing.			
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.	• 44.1 The HSCB and PHA will, supported by DHSSPS policy, contribute as required, appropriate to local circumstances, to this UK-wide policy area.	Monitoring and progress reports to be published annually no later than 30 June.	HSCB PHA DHSSPS
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.	• 45.1 The Department will work with medical training and education bodies, commissioners, providers and patient groups to ensure that appropriate training is provided for NGS with regard to the relevant protocols.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
46	Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.	• 46.1 The Department will encourage local researchers and industry to engage with research funding organisations on a UK-wide, European or international basis in order to promote collaborative research.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
47	Support initiatives to facilitate engagement	• 47.1 See commitment 46	Monitoring and progress	DHSSPS

	between patients, clinical care teams, researchers and industry wherever practical.	above. • 47.2 The Department will work with Genomics England to establish a Genomics Medicine Centre in Northern Ireland and participate in the 100,000 genome project. • 47.2.a – Produce a proposal and business case for a NI Genomics Medicine Centre and secure the funding. Estimated cost 2015 – 2018 is £3.3m. • 47.2.b – Submit a funding application to the UK Medical Research Council for a £1m grant contribution towards establishing the NI Genomics Medicine Centre	reports to be published annually no later than 30 June. Monitoring and progress reports to be published annually no later than 30 June. 30/07/2015 30/07/2015	DHSSPS DHSSPS DHSSPS
48	Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders.	• 48.1 The Department will work with local researchers and industry to set out the benefits of collaboration for all stakeholders.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
49	Continue to build a cohesive infrastructure	• 49.1 The Department will	Monitoring and progress	DHSSPS

	for implementation and coordination of rare disease research in the NHS.	contribute as required, appropriate to local circumstances, to this UK-wide policy area.	reports to be published annually no later than 30 June.	
50	Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.	• 50.1 The Department will contribute as required, appropriate to local circumstances, to this UK-wide policy area.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
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).	• 51.1 The Department will contribute as required, appropriate to local circumstances, to this UK-wide policy area.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS

Theme 6: Collaboration between Northern Ireland and the Republic of Ireland

The Northern Ireland Plan commits the Department to identifying opportunities to work with healthcare policy makers, commissioners and providers in the Republic of Ireland and organisations involved in research in the field of rare diseases. The intention of this would be to maximise the potential benefits arising from collaboration to meet the respective needs of both jurisdictions.

The following Action is aimed at identifying and developing opportunities to collaborate with the Republic of Ireland in cross-jurisdictional initiatives to support people with rare diseases.

UK Strategy Commitment		Actions to be taken	Completion Date	Owner
	Not Applicable.	• 52.1 The Department will work with the Department of Health and other Agencies in the Republic of Ireland, to identify opportunities for collaboration in the field of rare diseases to meet the respective needs of the populations of the two jurisdictions.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS
		• 52.2 The Department will work with the Department of Health in the Republic of Ireland to identify opportunities for collaboration and funding within European Union initiatives and programmes in the field of rare diseases.	Monitoring and progress reports to be published annually no later than 30 June.	DHSSPS

Annex A

PRIORITY ACTIONS FOR IMPLEMENTATION REQUIRING NEW INVESTMENT IN THE INITIAL WORK PROGRAMME (2015/16 and 2016/17)

All actions set out above under the six activity themes will be progressed over the lifetime of this plan. Their implementation will mostly be funded from within existing DHSSPS related administrative and programme delivery budgets to be prioritised within the annual financial planning cycle. However, several initiatives will require new investment and these are

identified below together with the estimated cost. Implementation of these initiatives will therefore be subject to securing available funding and business case approval.

Theme 1: Empowering those affected by rare diseases

UK Strategy Commitment		Actions to be taken	Initial Work Programme/Estimated Cost	Completion Date	Owner
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 – including in the development of the four country plans to implement the Strategy.	1.1 The Department and HSC will maintain and enhance the existing avenues of communication with key stakeholders for people living with the impact of a rare disease and their representatives.	1.1.a. – Complete a communications review and introduce enhanced methods of communication. Estimated cost £30K.	30/06/2016	DHSSPS

Theme 3: Diagnosis and early intervention

UK Strategy Commitment		Actions to be taken	Initial Work Programme/Estimated Cost	Completion Date	Owner
15	Improve education	• 15.1 The	15.1.a. – Complete a scoping exercise	30/06/2016	DHSSPS

	and awareness of rare diseases across the 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.	Department will work with the Northern Ireland Medical and Dental Training Agency (NIMDTA), universities, Royal Colleges and patient groups to deliver appropriate training.	(training needs analysis) and produce a training action plan Estimated cost £20K.		
20	Assess the potential for rare disease databases where they do not exist.	• 20.1 The Department will work with all stakeholders to improve coordination of existing and additional rare disease registries / resources / databases to facilitate the creation	20.1.a. – Complete a database review and produce a fully costed action plan to implement a Northern Ireland register for Rare Diseases. Estimated cost £20K.	30/06/2016	DHSSPS

		of a Northern Ireland register for Rare Diseases.			

Theme 5: The role of research

UK Strategy Commitment		Actions to be taken	Initial Work Programme/Estimated Cost	Completion Date	Owner
47	Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.	• 47.2 The Department will work with Genomics England to establish a Genomics Medicine Centre in Northern Ireland and participate in the 100,000 genome project.	47.2.a – Produce a proposal and business case for a NI Genomics Medicine Centre and secure the funding. Estimated cost 2015 – 2018 is £3.3m. 47.2.b – Submit a funding application to the UK Medical Research Council for a £1m grant contribution towards establishing the NI Genomics Medicine Centre.	31/07/2015 31/07/15	DHSSPS DHSSPS

ANNEX B

ACRONYM	DEFINITION
DHL	Department of Health London
DHSSPS	Department of Health, Social Services and Public Safety
ECR	Extra Contractual Referral
EM	Experimental Medicine
GB	Great Britain
HEE	Health Education England
HSC	Health and Social Care
HSC R&D Division	Health and Social Care Research and Development Division
HSCB	Health and Social Care Board
ICD-11	International Classification of Diseases 11th Revision
IFR	Individual Funding Requests
MDTs	Multi-Disciplinary Teams
MSC	Modernising Scientific Careers
NGS	Next Generation Sequencing
NHS	National Health Service
NICE	National Institute for Health and Care Excellence
NIECR	Northern Ireland Electronic Care Record
NIMDTA	Northern Ireland Medical and Dental Training Agency
NIRDP	Northern Ireland Rare Disease Partnership
PHA	Public Health Agency
R&D	Research & Development
RDAG	Rare Disease Advisory Group
RDUK	Rare Diseases UK
RCN	Royal College of Nursing
TA's	Technical Appraisals
TYC	Transforming Your Care
UK NSC	UK National Screening Committee
UKGTN	UK Genetic Testing Network

ANNEX C

Glossary of Terms

Word/Phrase	Definition
Tertiary centre	A hospital or centre that provides tertiary care, which is health care from specialists after referral from primary care and secondary care.
Primary Care	General health care traditionally provided by GPs, and is the first contact for patients within the healthcare system.
Secondary Care	Medical care provided by a physician who acts as a consultant at the request of the primary physician.
Equality Impact Assessment	A process designed to ensure that a policy, project or scheme does not discriminate against any disadvantaged or vulnerable people.
Regulatory Impact Assessment	The role of an RIA is to provide a detailed and systematic appraisal of the potential impacts of a new regulation in order to assess whether the regulation is likely to achieve the desired objectives.
EUROCAT	A European organisation which aims to improve the collection of data about congenital disorders.
EUROPLAN	The European Project for Rare Diseases National Plans Development (EUROPLAN) is a project co-funded by the EU Commission (DG-SANCO) to promote and implement National Plans or Strategies to tackle rare diseases, to share relevant experiences within Countries, linking national efforts with a common strategy at European level.
NICE	The National Institute for Health and Care Excellence (NICE) provides national guidance and advice to improve health and social care.
Epidemiology	The science that studies the patterns, causes, and effects of health and disease conditions in defined populations.
Extra Contractual Referral	This occurs when the Health and Social Care Board approves a consultant's request to transfer a patient to a provider outside Northern Ireland for assessment or treatment which the consultant considers necessary but which is not available through HSC facilities locally.
Individual Funding Request	An individual funding request is a request for an individual to have access to a specific treatment (the majority of which are for a specialist drug treatment) within the acute programme that is not normally commissioned or funded within Northern Ireland.
ICD-11	The International Classification of Diseases (ICD) is the foundation for the identification of health trends and statistics globally. It is the international standard for defining and reporting diseases and health conditions. It allows the world to compare and share health information using a common language. The 11th revision process is underway and the final ICD-11 will be released in 2017.
Nomenclature	The terminology used in a particular science, art, activity, etc

Quaternary Centre	A Centre which provides Quaternary care which is considered to be an extension of tertiary care - even more specialised and highly unusual. The types of care that might be considered to be quaternary would be experimental medicine and procedures, and highly uncommon, specialised surgeries.
UK Clinical Trials Gateway	The UK Clinical Trials Gateway provides easy to understand information about clinical research trials running in the UK, and gives access to a large range of information about these trials. It is designed to enable you and your clinician to locate and contact trials of interest to you.
Next Generation Sequencing	This is a methodology which allows millions of fragments of DNA from a single sample to be sequenced in unison, using special technology which allows an entire genome to be sequenced in less than one day.