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An Roinn Sláinte
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Northern Ireland Rare Diseases Action Plan 2022/23

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Foreword from Minister

In January 2021, I joined my counterparts across the 4 nations of the United Kingdom to publish the UK Rare Diseases Framework, outlining national priorities for improving the lives of those affected by rare conditions. In this first Northern Ireland Rare Diseases Action Plan we report on progress already made and take the necessary steps to continue building these into meaningful actions to address the priorities of the UK Rare Diseases Framework.

Rare conditions affect fewer than one in 2,000 people, with an estimated 7,000 different conditions. Because of their rarity, people living with rare diseases face specific challenges within the health and social care system and wider public services. The rare disease community includes newborns, children and adults; people living with both genetic and non-genetic conditions; people who have received a diagnosis, but also people who may never be diagnosed. Whilst around 80% of rare diseases have an identified genetic origin, it is important to recognise that other factors such as disordered immunity, infections, allergies, deterioration of body tissues and organs, and disruption to development in the womb can cause rare diseases.

Although rare diseases are individually rare, they are collectively common, with 1 in 17 people being affected by a rare disease at some point in their lifetime. In the UK this amounts to more than 3.5 million people, approximately 110,000 in Northern Ireland. It is therefore important that our health service can provide this large and diverse patient population with the best possible care available under challenging conditions. While the COVID-19 pandemic created unprecedented challenges for our health service, as we begin to emerge from the throes of the pandemic, we have an opportunity to learn lessons and build back better.

The actions described in this plan are far-reaching and diverse, interweaving across and between individual actions and stakeholders. Some may be easier to implement than others, for example where there are opportunities to work smarter and more efficiently with little or no cost. Some are likely to have longer lead-in times where interconnectivity with other government strategies is required.

The Framework's 5 cross-cutting themes of the patient voice; national and international collaboration; pioneering research; digital, data and technology; and, wider policy alignments are implicit within the individual actions of this Plan. From developing a rare disease registry, enhancing the education and training of pre and post registered health professionals, developing rare disease care pathways, to improving access to rare disease specialist teams, the cross-cutting themes will be embedded in the work being taken forward in an effort to improve the lives of people living with rare diseases. The actions in this Plan have

been developed collaboratively with our delivery partners and key stakeholders across the health landscape and in consultation with members of the rare disease community.

This Action Plan is the first in a series of annual updates reporting on progress made and developing new initiatives, and is an important step in addressing the challenges faced by the rare disease community. Over the course of the coming year work will continue to monitor progress, drive change, and step up engagement with organisations who have crucial roles in supporting people living with rare diseases.

The Framework and Action Plan represent commitment to the rare disease community. However it is also important to recognise the strengths across the UK in science and research, and wider initiatives in addressing the challenges of rare diseases. Other UK strategies including Genome UK, the Future of UK Clinical Research Delivery and Northern Ireland's Mental Health Strategy will all support work to continue improving the lives of patients – from those affected by the most common conditions to the very rarest of diseases.

Thank you to all involved in the development of this plan and its future implementation – from officials working across the health and care system through to the clinicians and researchers, the dedicated patient organisations, and most importantly, those personally affected by rare disease.

Robin Swann MLA

Minister for the Department of Health, Northern Ireland

Executive Summary

The UK Government and devolved administrations published the *UK Rare Diseases Framework* in January 2021, setting out a shared vision for addressing health inequalities and improving the lives of people living with rare diseases across the UK. The Framework outlined 4 key national priorities as follows:

- helping patients get a final diagnosis faster;
- increasing awareness among healthcare professionals;
- better co-ordination of care; and,
- improving access to specialist care, treatment and drugs.

To turn this vision into action, each of the four UK nations has committed to developing nation-specific action plans detailing how these will be addressed.

This is Northern Ireland's first Rare Diseases Action Plan to support implementation of the Framework and is for the initial period from April 2022 to March 2023. It has been developed collectively with delivery partners across the health system and in close consultation with the rare disease community. It sets out specific, measurable actions for the next year under each of the four priority areas, including the outcomes that we aim to achieve.

By bringing together many key organisations within the health system to deliver this action plan, we have a unique opportunity to tackle the challenges facing those living with rare diseases. Over the course of the coming year, we will monitor the progress of these actions closely, seeking input from those living with rare diseases to ensure we are measuring the outcomes that matter most.

However, we know that there is more to do, particularly as the health system begins to recover from the effects of the COVID-19 pandemic. While delivery of this first action plan is underway, we will continue to explore future directions and develop new actions, informed by the needs of the diverse rare disease community.

Through this action plan, we will take the first steps in Northern Ireland towards achieving our overarching vision – delivering improvements in diagnosis, awareness, treatment and care, and creating lasting positive change for those living with rare diseases.

The following table lists the 14 high level actions which will be developed during this coming year. The table in Annex A provides further detail for each of these 14 actions.

Table 1: List of 14 Actions for development during 2022/23

	Action
1	Establish an online Rare Diseases Information Hub for Northern Ireland.
2	Agree a new Northern Ireland Genomics Partnership model to deliver against the UK Genome Strategy.
3	Review newborn screening for Northern Ireland, participate in the UK National Screening Committee and engage in UK-wide screening initiatives.
4	Establish a steering group to develop a rare disease registry for Northern Ireland (NIRADCAR), engaging with UK rare disease registries.
5	Establish a Rare Diseases Education and Training steering group to include all relevant stakeholders and develop a cost / benefit analysis to include a dedicated education co-ordinator.
6	Raise awareness generally, but in addition across a range of specialist communities including the newly established All Party Group for Rare Diseases. Working jointly with educators and using all available mediums, including social media platforms.
7	Establish a scoping group to develop an options appraisal for an expert centre for rare diseases, to include a rare diseases specialism information co-ordinator.
8	Establish a working group to develop care pathways / models of care, including developing a methodology to deliver exemplar care pathways for 1-2 individual rare conditions over the next 12 months and engaging with encompass to develop appropriate clinical skills for the new Digital Programme.
9	Ensure the mental health needs of the rare diseases population are included across all appropriate Northern Ireland government strategies and programmes.
10	Explore the potential for a Patient Portal for patients to view their health information; key stakeholders to engage with encompass during the design, build and rollout of the programme. Deliver a rare diseases community update event with encompass.
11	Improve access to rare diseases drugs by producing information resources and developing a clear roadmap for available initiatives to be available in one place. Initiate discussion with delivery partners to consider options for a review of commissioning processes for access to medicines and specialist medicines into the Northern Ireland health system.
12	Improve access to other rare disease specialist teams locally, nationally and internationally by hosting all relevant information centrally and providing clear instruction on pathways to tertiary centre support.
13	Improve awareness of and participation in rare diseases research, including establishing a baseline of current studies actively recruiting patients and regularly reviewing clinical trial registries and research databases to create a clear picture of ongoing studies. This will include development of a communication plan.
14	Explore the potential for a Rare Diseases Champion role.

UK Rare Diseases Framework

Development of the *UK Rare Diseases Framework* (henceforth the 'Framework' throughout this document) was based on the outcomes of the '*National Conversation on Rare Diseases'*, launched in 2019[1]. The Conversation gathered views across the rare disease community on the major challenges faced by people affected by rare conditions across the UK. An impressive 6,293 responses were received, which helped identify four high-level priority areas to bring about real change and forming the basis of the Framework. The four priorities are:

- helping patients get a final diagnosis faster;
- increasing awareness of rare diseases among healthcare professionals;
- better coordination of care;
- improving access to specialist care, treatments, and drugs.

To turn these priorities into a reality, we recognise that significant action is needed across health and social care (HSC). In addition to the four priorities, the Framework identifies five underpinning themes to support the four priorities. These themes are:

- patient voice;
- national and international collaboration;
- pioneering research;
- digital, data and technology;
- wider policy alignment.

The Framework was published on 9 January 2021 and outlines a national vision for how the UK will improve the lives of those living with rare diseases over the next five years. All four UK nations have committed to developing clear and tangible action plans to deliver on our collective Framework.

Northern Ireland's first Rare Diseases Action Plan aims to support implementation of the Framework and is for the initial period from April 2022 to March 2023. It has been developed with stakeholders across the healthcare system, the Northern Ireland Rare Diseases Partnership (NIRDP) representing the rare disease community and rare disease researchers and educators from Queen's University Belfast (QUB) and Ulster University (UU) to bring about specific and measurable improvements for people living with a rare disease.

[1] UK Rare Diseases Framework, Annex B

It is currently estimated that there are more than 7,000[2] rare diseases, with new conditions continually being identified as research advances. While around 80% of rare diseases have an identified genetic origin, they can also be caused by other factors such as disordered immunity, infections, allergies, deterioration of body tissues and organs, or disruption to development while in the womb.

Although rare diseases are individually rare, they are collectively common – with 1 in 17 people being affected by a rare disease at some point in their lifetime. In the UK this amounts to over 3.5 million people. It is important that HSC services provide this large and diverse patient population with the best possible care.

Rare diseases can be both life-limiting and life-threatening. They disproportionately affect children. Around 75% of rare diseases affect children and more than 30% of children with a rare disease die before their fifth birthday. People living with rare diseases, and their families, often face a lifetime of complex care leading to a profound impact on their education, financial stability, mobility, and mental health as well as their clinical and social care needs. It is vitally important that their voices are heard and acted upon when developing wider policy.

UK-wide implementation

The Framework is a UK-wide document. However, each of the four UK nations has its own delivery or implementation group responsible for drafting and monitoring nation-specific action plans. All four nations have committed to publishing their action plans by the end of 2022. Throughout development of the action plans, each nation is engaging with members of the rare disease community to make sure the work is relevant and fit for purpose. Once published, action plans will be updated and reviewed annually to measure progress, update actions, or add new ones. In developing individual action plans there is potential for all four UK nations to benefit by sharing good practice and developments, and learn from each other's experience.

Developing nation-specific action plans involves balancing the specific health needs of the individual UK nations with the Government's commitment to health equity and avoiding disparities. To further help with implementation of the Framework, two UK-wide boards have been created:

1 The UK Rare Disease Framework Board, providing high level co-ordination of rare disease policy and action plans across the four UK nations; and,

[2] UK Rare Diseases Framework – source information for this section

2 The UK Rare Diseases Forum, providing a platform to engage a wide range of stakeholders in the rare disease community for advice and input.

The Forum has two parts: a core membership which meets twice a year, and an online knowledge and collaboration platform for continual engagement with a broad range of stakeholders, both of which feed into the strategic UK Rare Diseases Framework Board.

Northern Ireland Action Plan

The Northern Ireland (NI) Rare Diseases Implementation Group develops, oversees and co-ordinates delivery of NI's Action Plan. It brings together key stakeholders across the healthcare system, including representatives from relevant areas such as commissioning, public health, the rare disease patient voice via the voluntary sector, academia, adult social care, mental health, and genomic medicine. The development of NI's first action plan has been informed by more than 2,000 voices from the rare disease community. Over the course of 2021 and 2022, the Implementation Group met regularly to develop and agree the actions which form the basis of the plan.

The Implementation Group has worked in partnership to develop this action plan, bringing together representatives from the rare disease patient and public voice, and the clinical community, including:

- Department of Health – Secondary Care, Mental Health, Social Care representatives;
- Health and Social Care Board – Specialist Commissioning, Pharmacy, Adult Social Care
- Public Health Agency – Research & Development
- Health and Social Care Trusts – clinicians (paediatric and adult)
- encompass – Northern Ireland digital integrated care record initiative team
- Northern Ireland Rare Disease Partnership – umbrella group representing the rare disease community
- Queen's University Belfast – researchers and educators
- Ulster University – researchers and educators

All of these key stakeholders are committed to working in partnership to co-produce, design and deliver this plan as efficiently and effectively as possible to meet the needs of the rare disease community in Northern Ireland.

Following publication of this Action Plan, the Implementation Group will continue to meet to co-ordinate and report on delivery and develop actions for our second action plan in 2023

Community engagement

In developing the NI Rare Disease Action Plan, we have placed the needs of those living with rare diseases at the forefront. In September 2021, we held an engagement event to offer an opportunity for people living with a rare disease and those working in the field of rare diseases to engage with the action planning process. Attendees included: carers and family members; rare disease charities; healthcare professionals; industry partners; and, researchers. Anyone living or working in the field of rare diseases were offered the opportunity to feed into the action plan in writing at that stage.

In addition, the NI Rare Disease Partnership represents the voice of the rare disease community at every Implementation Group meeting and undertook a final roundtable focus group on the draft action plan in March 2022.

Progress to date

It is now a year since the Framework was published. Alongside the work to produce this action plan, progress has already been made against the priorities of the Framework through collaborative working with local and international colleagues, including individuals in the Department of Health, across NI HSC Trusts, Public Health Agency, Voluntary Groups, Queen's University Belfast, Ulster University, commercial partners, Northern Ireland Rare Disease Partnership, patients, carers, families, clinicians, and researchers, which is helping drive forward positive progress for rare diseases in NI. Some of this progress is summarised below.

Genomics

Significant strides have been made in helping patients get a final diagnosis faster, for example the publication of *Genome UK: The Future of Healthcare* established the strategic direction for the future of genomics across the UK. Genome UK provides a 10 year strategy to create the most advanced genomic healthcare system in the world, underpinned by the latest scientific advances to deliver better health outcomes at lower cost.

Notable Genomic Medicine Service (GMS)[2] achievements to date include the success of the 100,000 Genomes Project, the establishment of a Regional Molecular Diagnostic Service

(RMDS)[3] for NI and the rapid upturn in pathogen genomic sequencing capability in response to the COVID-19 pandemic.

Genomic medicine has greatly improved diagnostics and treatments across many areas of healthcare. Following Genome UK, NI has a shared commitment to ensure equity of service and quality of care for our rare disease population and to further support the public health priorities by continuing to enhance both our pathogen and infectious diseases sequencing capabilities.

New computer tools such as GenOCEANIC (Genomics Open Core Engine for Accelerating Northern Ireland Care) are being developed as a result of the 100,000 Genomes Project to help clinicians streamline rare disease testing for their patients. Experiences from participating healthcare professionals are supporting the practical day-to-day development of the genomic medicine service for NI. Views from patients participating in the 100,000 genomes project have informed recruitment, regular engagement, and delivery of results for whole genome sequencing.

Registry

The initial scoping review on developing NI's rare disease and congenital anomaly register (NIRADCAR) is being progressed in line with the UK Framework agreement and local funding constraints, with five registry scoping meetings held with diverse stakeholders. Several meetings have engaged with Leads of the National Congenital Anomaly and Rare Disease Register (NCARDRS) England, Congenital Anomaly Register and Information Service (CARIS which also specifically captures rare diseases) Wales, and Congenital Anomaly and Rare Disease Registration and Information System (CARDRISS) Scotland, to share best practice and agree common elements across the UK. Several research projects have been conducted to gather preliminary data towards developing NIRADCAR.

Training and Education

Over the past decade, rare disease teaching has been embedded into undergraduate medical, biomedical and nursing curricula at Queen's University Belfast (QUB), additionally providing postgraduate training and education in rare diseases for students and healthcare professionals, with many sessions including patients and their families speaking directly with students. Rare disease sessions have been integrated to personalised medicine and nursing

[3] See Annex C for list of abbreviations and definitions

modules at Ulster University (UU). The NI Rare Disease Partnership and the two NI universities (QUB and UU) have exchanged teaching material and worked together to set up new rare disease teaching sessions and harmonise existing sessions in different courses and schools. There is an evolving programme of local workforce development, including CPD accredited events, to help health and social care professionals use genomics and bioinformatics effectively. This needs to be strategically enhanced and taken forward consistently across NI and is being further considered within the Year 1 Action Plan.

To stimulate interest in developing a career in the rare diseases field and attract nurses, medical doctors or researchers for the future, the three delivery partners developed a collaborative science, technology, engineering and mathematics (STEM) project and engaged with 70 NI schools and 600 Key Stage 5 students. Five additional STEM activities focusing on raising awareness of rare diseases were delivered from March 2021 to February 2022.

Following publication of their quick reference guide for rare diseases[4], QUB explored rare disease perceptions among the NI General Practitioner (GP) community[5], and regularly host continuing professional development events for HSC professionals.

In addition, the NI Rare Diseases Partnership organised training sessions with the Pharmacy Forum and one with the Royal College of General Practitioners in NI, which was delivered by Medics for Rare Disease, a Genetics Clinician from Belfast HSC Trust and QUB.

All party working group

The NI Rare Diseases Partnership actively lobbied on behalf of, and with support from, the rare disease community to successfully establish an All-Party Group (APG) for Rare Disease at the NI Executive. The APG group will act as a mechanism to ensure that all future conversations around rare disease support and services have a direct conduit into our parliamentary structure at the highest level, whilst supporting awareness raising at an Executive and regional level.

[4] A quick reference guide for rare disease: supporting rare disease management in general practice | British Journal of General Practice (bjgp.org)

[5] Perceptions and experiences of rare diseases among General Practitioners: an exploratory study | medRxiv

All-island forum

February 2022 saw a new all-island patient centred North/South Rare Disease Forum which will enable individual rare disease-specific patient groups to come together quarterly to discuss areas of common concern or interest. This Forum will go some way towards unifying the key ‘expert by experience’ voices around the island and working to support identification of services that can be delivered collaboratively to improve quality of life for those with rare conditions.

Access to drugs

The UK aspires to be a world leader for development, testing, access and uptake of new and innovative treatments and technologies. The National Institute for Health and Care Excellence (NICE) is the independent body responsible for providing evidence-based guidance on whether medicines represent a clinically and cost-effective use of NHS resources, ensuring that NHS funds provide the most health benefit for society.

NICE assesses the majority of medicines through its standard technology appraisal (TA) programme, but also operates a separate highly specialised technologies (HST) programme for a small number of medicines for very rare diseases. New commercial flexibilities have resulted in NICE appraising all new medicines. The NI Department of Health has a formal link with NICE where TAs are endorsed for implementation within the NI healthcare system where they are legally and policy applicable.

Currently NICE is updating its processes and a number of the changes adopted will support timely patient access to innovative medicines for patients with rare diseases. For example, NICE has introduced a new severity modifier for committees to consider the severity of the disease or condition under consideration when making recommendations. This reflects evidence that society values more highly the health benefits for people with very severe conditions, and committees will be able to give additional weight to the treatment benefits for rare and severe conditions under consideration.

Where there is uncertain evidence in relation to a medicine, a particular issue for rare diseases where the population is small, NICE will adopt a more accepting, flexible and proportional attitude towards evidence uncertainty within its decision making. In addition, NICE is adopting process changes to help improve participation of patients and clinical experts, and has refined the criteria used for identifying an HST which will provide greater clarity and predictability for stakeholders.

Over the next year NICE plans to implement these changes to make its methods and processes fairer, faster and more consistent – supporting timely patient access to new treatments, including those for rare diseases. NICE will also continue to provide support to the life sciences industry, including companies developing therapies for rare diseases.

Current Context

Progress in developing this action plan has taken place against the backdrop of the current COVID-19 pandemic, which has caused significant disruption to the healthcare service. A focus on COVID-19 has meant that some routine and primary care services have been scaled back, leading to delays or cancellations in diagnostic testing, transfusions, surgeries, scans, and routine appointments – all of which have significantly impacted the rare disease community as well as other patient populations.

Similarly, safety considerations, redeployed staff and travel restrictions have caused additional barriers to rare disease research, where cohort sizes are often already small. The Clinical Research Resilience, Recovery and Growth Programme, which has a focus on managing the recovery of non-COVID research in the UK, with each country having its own delivery group/programme, will contribute to the building of NI's non-COVID research activity.

In the context of rebuilding our healthcare system, the Minister has published a *Strategic Framework for Rebuilding Health and Social Care Services* (June 2020) to guide a regional approach to service rebuilding and transformation which places workforce and technology at the heart of long-term planning across the healthcare service.

Whilst ambitious, the NI Rare Diseases Action Plan is also realistic, recognising that it may take time to implement change within the current context.

Below, we set out our plans for the year ahead, with a clearly identified range of actions and relevant milestones, some of which are achievable within a short timescale and limited resource implications. The Implementation Group is committed to delivery against all areas of work, keeping in mind that some actions will take time to scope, resource and deliver. It is important to recognise that some elements of this plan are subject to available resources during very challenging times for our local health economy.

Framework Priorities: Northern Ireland Actions for 2022/2023

This NI Rare Diseases Action plan should be read in conjunction with the *UK Rare Diseases Framework* which provides further detail on the principles of the Framework and the approach for developing each nation's Action Plan.

The four priorities of the Framework have been highlighted as major challenges by the rare disease community across the UK. Below we describe actions under each of the priority areas that we aim to achieve in the next year. An easy-read table of the actions and milestones is included at Annex A.

This action plan is a high-level outline of the first steps to implement the Framework. It is important to understand that it is not an implementation plan. Delivery partners may need to consider developing implementation plans for individual actions, particularly those that will take longer to progress and are likely to span across several years.

Priority 1: HELPING PATIENTS GET A FINAL DIAGNOSIS FASTER

Getting a diagnosis is of vital importance for people living with rare diseases and their loved ones. An accurate and timely diagnosis can facilitate access to treatment and care, provide a possible prognosis, and offer options for family planning. It can also provide a means of connecting with a supportive community and potential involvement in research, including clinical trials. In some cases, if a condition is diagnosed before the onset of symptoms, it may be possible to limit or even prevent harm.

Importantly, a correct diagnosis also ends what is for many people a "diagnostic odyssey". The complex nature of many rare conditions means that patients may undergo multiple referrals, inconclusive invasive tests and sometimes incorrect diagnoses before a final accurate diagnosis is reached, during which time their condition may deteriorate as well as having a negative impact on mental health and associated care needs of patients and their families.

In developing the following actions to address Priority 1: helping patients get a final diagnosis faster, we have taken into consideration both genomic and non-genomic diagnoses as both are important to our rare diseases community.

Action 1: Information Hub

To establish an online Rare Diseases Information Hub for NI, with a dedicated person employed to collate relevant information, act as a contact point and source of advice for people living with a rare disease in NI and their families, contribute to Orphanet for NI-specific information, and connect those working in the field of rare diseases.

Building on the work previously undertaken, the key milestone for this first year is to establish a scoping group to consider the detail of an information / support hub in light of the UK Framework. To do this we will work with relevant people across other actions within our Year 1 Action Plan to ensure appropriate input is provided. The scoping group will develop and build relationships with the NI Rare Diseases Forum to ensure the patient's voice is represented, and foster links with the All-Island Rare Diseases Forum to ensure all-island information is collated and included where possible.

The expected outputs for Action 1 will include a review paper with recommendations for a best practice model and identification of the resources required to establish an effective Information Hub.

It will also be important to secure funding for a dedicated person/s (to include rare diseases navigators) to take the Information Hub work forward, therefore a relevant business case or funding application will be developed.

Action 2: Agree new NI Genomics Partnership model to deliver against the UK Genome Strategy.

Work with key stakeholders across government, the health service, PHA, academia, industry, patients, carers, and colleagues in the Republic of Ireland to agree the future vision and develop policy decisions on genomics development and provision for NI. This action includes linking and collaborating with all UK nations in line with the shared commitments of *Genome UK: The Future of Healthcare*, the national strategy for genomic healthcare.

The key milestones for this first year are to establish a governance structure and accountability for a new partnership model with stakeholders. As part of the partnership model, work towards establishing a new Genomics Medicine Service (GMS) responsible for advising on the genetic testing capacity and capability offered to patients, as well as considering whole exome and / or whole genome sequencing in specific circumstances, endeavouring to ensure equity of service with the rest of the UK.

Building on lessons learned from our participation in the 100,000 genomes project, we will support multidisciplinary collaborations where researchers and clinicians work together to help resolve complex and novel findings from genome data. We will expand the partnership model to ensure NI meets its shared commitments on all pillars of *Genome UK*. Detailed clinical and genomic data from patients will be combined with clinical decision support to facilitate diagnosis of rare genetic disorders. We will also work to agree an investment and implementation plan by the end of the first year.

Action 3: Review newborn screening

Newborn screening plays an important role in diagnosing rare diseases early, offering opportunities for treatment, management, and support - often before symptoms even develop. This is particularly important because 75% of all rare diseases affect children, accounting for about a third of infant deaths and the majority of childhood mortality in children aged under 15 in the UK[6, 7].

The UK National Screening Committee advises Ministers and the Health Service in all four UK countries on all aspects of screening. Using research evidence, pilot programmes and economic evaluation, the Committee assesses the evidence for national screening programmes against a set of internationally recognised criteria, taking a range of different factors into account. Currently nine rare conditions are screened in newborns through the NHS Newborn Blood Spot Screening Programme.

For our year one action we commit to participating in the UK National Screening Committee and to follow relevant guidance to ensure appropriate use of screening tools in line with UK National Screening Committee recommendations.

We will also establish a task and finish group to scope the newborn screening needs for NI, taking geographical prevalence of conditions on an all-Island basis into account. As an outcome of this review, we will suggest recommendations for optimised screening for conditions in NI.

Additionally, we will actively engage in all new UK-wide screening initiatives, to include exploring NI's participation in the national newborn screening research study which aims to

[6] "*In 2020, the main causes of death among children aged up to 15 years continue to be congenital malformations, deformations and chromosomal abnormalities*" Child and infant mortality in England and Wales - Office for National Statistics (ons.gov.uk)

[7] Orphanet J Rare Dis . 2020 Nov 4;15(1):311. doi: 10.1186/s13023-020-01574-7. A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland Emer Gunne[1], Cliona McGarvey[2], Karina Hamilton[2], Eileen Treacy[3], Deborah M Lambert[3], Sally Ann Lynch[4 3]

evaluate carefully the benefits and risks of implementing newborn genomic screening to accelerate diagnosis and enable earlier access to treatments for rare genetic conditions.

Priority 2: INCREASING AWARENESS AMONG (CURRENT AND FUTURE) HEALTH CARE PROFESSIONALS (INCLUDING POLICY MAKERS AND WIDER SOCIETY)

Raising awareness of rare diseases is crucial to improve the speed and accuracy of diagnoses, as well as ensuring patients receive the best possible clinical care, particularly in medical emergencies. With over 7,000 rare diseases, it is not possible for healthcare professionals to receive comprehensive training on every condition. It is therefore important that they are aware of rare diseases more broadly and are open to considering them as part of the diagnostic process. This includes providing training and resources to enable healthcare professionals to recognise rare diseases in patients and be aware of potential specialist diagnostic options and treatment needs, as well as signposting to support and care pathways.

Action 4: Develop a Northern Ireland Rare Diseases Registry

This action is to establish a Northern Ireland RAre Disease and Congenital Abnormality Registry (NIRADCAR), linked to the new encompass[8] integrated care record, which is a NI strategy introducing a digital integrated care record across the region and supporting the vision to transform HSC and improve patient safety and health outcomes. It is intended that NIRADCAR will link closely to rare disease registries across the UK, and, potentially in the future, in Ireland.

To achieve this action it is vitally important that NI regularly engages with registry colleagues across the four nations to ensure alignment of rare disease registry plans, which will support a UK-wide national rare disease registration facility. Therefore, we will appoint a registry lead and establish a registry steering group to identify how best to obtain a minimum core dataset and standardised coding for the registry. Inclusion criteria and analysis plans will be developed for NI, taking account of stakeholder input from all relevant sectors, specifically ensuring individual condition / medical speciality registry specialists are included to add value from the community and voluntary sector. Part of the work in developing a NI registry will be

[8] http://www.hscboard.hscni.net/encompass/

to scope the logistics for accommodating the registry; a scoping review is expected to be completed and a business case developed as part of this Year 1 Action Plan, to include necessary resources to take the registry work forward.

Alongside the steps above to take forward establishing a registry, we will source registry information for one to two individual rare conditions associated with exemplar care pathways. It is envisaged that these will form a template for the work needed for other rare conditions.

In addition, during our Year 1 Action Plan, we will work with the NI Cancer Registry to generate the first factsheet for rare cancers in NI. Worldwide, rare cancers make up 22 out of every 100 (22%) of cancers that are diagnosed each year[9]. This means that 1 in 5 people diagnosed with cancer has a rare type. Research also shows that about 1 in 3 people with a rare cancer have a particularly rare type.

The overall aim of NIRADCAR is to provide improved identification and monitoring of rare diseases leading to smarter care for individuals living with a rare disease. A summary of the objectives and deliverables can be found in the 2020 registry workshop report.

Action 5: Pre and post registration education and training, including an education co-ordinator

Rare diseases education is already embedded in several undergraduate courses at Ulster University and Queen's University Belfast, with some postgraduate taught modules including specific rare disease sessions. In addition, the NI Rare Disease Partnership delivers awareness raising sessions with health care organisations such as the Royal College of General Practitioners and the Pharmacy Forum, and is developing 'Expert by Experience' modules within the universities.

However, it is important that training for all healthcare professionals across relevant specialities, including general practice and those involved with emergency care, includes information on how to engage, inform, involve and support the diverse rare disease population. Over the coming year we will build on the links already in place with UK and Republic of Ireland training organisations to collaborate on healthcare education and training, from undergraduate education through to continuing professional development (CPD).

To enable regional oversight of healthcare training, our Year 1 Action is to establish a Rare Diseases Education and Training Steering Group to include all relevant stakeholders, with emphasis on Personal and Public Involvement (PPI)/co-production, design and delivery. We

[9] https://www.cancerresearchuk.org/about-cancer/rare-cancers/what-rare-cancers-are Accessed 21 Feb 2022

will develop a cost / benefit analysis to support a bid for additional resources, including a dedicated education co-ordinator to take the education and training work forward.

Until the steering group is in place our two universities will ensure that CPD accreditation is in place for relevant postgraduate training events. The NI Rare Diseases Partnership will continue to scope opportunities outside the university environment, and all three partner organisations will promote and disseminate information on relevant events through respective websites and social media channels.

Healthcare teaching organisations will continue to embed delivery of rare disease teaching sessions, including the patient voice, for all health care professionals to include undergraduate medical, biomedical, nursing and allied health professions (AHP) students, as well as postgraduate taught students.

Alongside the above steps, we will build on existing rare disease awareness sessions and deliver 12 sessions per year.

Action 6: Raise awareness of rare diseases across a range of communities

The difficulties and problems of the rare disease population are wider than healthcare and reach into other domains such as housing, schooling and transport. With this action we commit to raising the profile of rare diseases by increasing public awareness through public events such as Rare Disease Day and other rare disease campaigns.

Our key milestones for year one include providing the NI Rare Diseases Partnership support to the NI Assembly All Party Group for rare diseases and hosting the annual all-Ireland Rare Disease Day Conference on a hybrid basis where possible.

We also commit to hosting at least two rare disease focused science, technology, engineering and math (STEMNI) initiatives this year. Our partner organisations, Queen's University Belfast, Ulster University, the NI Rare Diseases Partnership and other rare disease community and volunteer groups, will promote and disseminate information on relevant events through respective websites and social media channels.

In addition, we will participate in bi-monthly all-Ireland rare disease webinars associated with the All Ireland Students for Rare Disease Network, and plan for quarterly rare disease discovery research meetings with both professional and public components.

To raise awareness across communities we intend to deliver a range of webinars, information sessions and outreach support meetings. Through training we will empower patient group representatives to be active voices for their own discrete groups as well as the broader rare

diseases community, particularly in relation to more complex areas such as research and development.

Priority 3: BETTER CO-ORDINATION OF CARE

Because of the chronic and complex nature of many rare diseases, people living with rare conditions often face multiple hospital appointments and complex condition management involving multiple specialties spread over a number of different hospitals and services. With more than 7,000 rare conditions identified to date, many do not have a dedicated service within HSC. In many cases, the responsibility for co-ordinating appointments and services falls to the individuals themselves or a family member or carer, which can result in a significant care burden. Care co-ordination should be flexible, keeping the patient at the heart of decision making. People living with rare diseases and their carers should have the support they need to navigate the healthcare system and should be empowered to do so if they wish.

Action 7: Develop an expert centre for rare diseases, to include a rare diseases specialism information co-ordinator

To develop a national dedicated rare disease care centre with a co-ordinator acting as a central point of contact for rare diseases across NI has consistently been one of the top priorities for stakeholders, including healthcare professionals, rare disease patients and their carers or family. This expert centre has the potential to develop a referrals process for rare diseases to enable patients and families to get the specialist help they need.

Key milestones for year one include establishing a scoping group to capture the care co-ordination landscape in NI, and further afield, and develop an options appraisal. This will include working with teams across all other actions of this Year 1 Action Plan. One of the outcomes will be to explore the requirement for a physical space and developing a business case and implementation plan for an expert centre by the end of year one.

Action 8: Develop care pathways / models of care

For a small country like Northern Ireland it is imperative that patients with rare diseases can access services across the province, GB and elsewhere. Therefore developing care pathways and providing patient information on available services, including the transition from paediatric to adult care and accessing care outside the region, is crucially important.

There should be clear patient information to enable better understanding, for example by using infographics and layman's terms / non-medical terminology.

Improving access through established care pathways across the many and varied disciplines related to rare diseases is essential to ensure care is effectively managed, the burden on patients and carers is minimised, and that healthcare professionals are working together to provide the right care at the right time. This includes defining what a better model of care looks like, and collaboration is required across all working groups detailed in this Action Plan.

Therefore the key task for this first year is to establish a working group to agree and develop a number of individual pathways and models. These will include developing a pathway for individuals with suspected, but not yet diagnosed rare diseases, for example through regular gene discovery clinics. Aligned to Action 4 (registry), the working group will also develop a methodology to deliver exemplar care pathways for one to two individual rare conditions in the first year, with a view to using these as a template for other rare conditions into the future. The group will work with paediatric and adult rare disease clinical leads as well as other individual clinical leads to progress work on agreed pathways / defined service specifications.

This action is interconnected with Action 10 and will require liaising with the encompass core team to familiarise potential users (clinicians and patients) with the patient facing functionality of MyChart (the operating mechanism) and ensure appropriate information is available for rare disease patients who wish to avail of this service. Importantly, there will be a need to engage with and recruit rare disease specialty clinicians to participate in the 'clinician builder programme' to develop the skills to enable clinical input to the building of rare disease pathways, including 'rare disease flag' signposting and other needs.

The working group will also continue to improve the virtual and face-to-face mixed model for consultations to connect patients with the clinicians they need, and scope the suggested pathways of existing individual rare disease voluntary organisations.

Action 9: Ensure the mental health needs of the rare diseases population are included across all appropriate Northern Ireland Government strategies and programmes.

From waiting for a diagnosis to navigating multiple avenues of care, individuals and families living with the realities of a rare disease often find the burden impacting on their mental health and wellbeing. This may be in addition to some specific conditions having a clinical mental health impact. In some cases, the impact of this has been exacerbated by the COVID-19 pandemic, which has affected the lives of those more vulnerable in society, as

well as causing disruption to routine care services. Evidence suggests that groups which had the highest risk of mental ill-health before the pandemic, including those living with pre-existing conditions, have been worst affected.

This action rightly looks first to our mental health services and the needs of the rare diseases population, with a focus on enhancing wellbeing and providing counselling support. However, it also emphasises the broader need for support across all relevant government departments. This includes, for example, the Department for Communities in relation to employment, housing and other accessibility issues, the Department of Education in relation to the Education Strategy and wellbeing needs of rare disease children with complex needs, and the Department for the Economy in relation to further and higher education, and job skills.

We recognise that much of the work in relation to this action will largely be included in other actions such as the Information Hub, the Expert Centre and developing pathways and models of care. However, this standalone action will remain as a prompt to ensure that the mental health and wellbeing of rare disease patients is considered appropriately across relevant areas of this Action Plan and is aligned with the Department of Health's Mental Health Strategy. This may include scoping appropriate links through other actions to ensure that clear pathways are identified between all relevant support organisations.

Action 10: Patient Portal

This is potentially a "one stop shop" where patients can view information, with access to all communications, healthcare professionals, summary care record and emergency care record. This action, similar to the rare disease registry, is dependent on the new encompass programme being rolled out across the province. It will take time to achieve and early exploration steps are required to determine the scope of the work. Additionally, this has the potential for UK-wide benefit and further discussion across the four nations will be required.

Taking the action forward will require discussion and liaison with relevant people across other Year 1 Actions to ensure appropriate input is provided. The action owner will need to ensure that the UK Rare Diseases Framework is appropriately considered by the NI Digital Strategy (encompass) during the design, build and rollout of the programme by establishing links with the design team to ensure maximum benefit to the rare diseases community.

This action will include exploring the potential to carry out a rare diseases specific public engagement event with the Encompass team to provide an update on the current position of encompass and how it may benefit the rare diseases community in terms of a patient portal.

Priority 4: IMPROVING ACCESS TO SPECIALIST CARE, TREATMENT AND DRUGS

Many rare diseases do not have established treatments, but where they do exist they can be life-changing and lifesaving, significantly improving prognoses and an individual's quality of life. Providing access to safe, high-quality specialist care and treatments presents challenges across the four nations of the UK, with some patients needing to travel significant distances to access specialist centres. The small number of patients affected by each rare condition means that the scale of clinical trials typically used to assess the safety and efficacy of medicines may not be possible. Assessment of, and access to, rare disease medicines can also require additional consideration from health technology assessment bodies due to limited and uncertain data.

Action 11: Improve access to rare disease drugs

The UK is a world leader in science and technology, with a world-class research infrastructure, and an increasing number of innovative drugs and treatments being developed for rare diseases. Ensuring rapid, safe and equitable access to these treatments as they become available will improve the lives of those living with rare diseases and foster an environment that will attract substantial investment in the high-value life sciences products of the future, leading to continued innovation in the field. However, there are clear challenges with promoting equitable access, with multiple treatments being developed for some rare conditions, whilst others do not receive the same levels of research activity, investment or attention.

We recognise that ensuring equity of access to rare disease medicines locally, regionally and nationally will improve by increasing health professional and patient awareness of the existing mechanisms to access available initiatives. Working with delivery partners, our aim is to produce information resources and a clear roadmap for available initiatives and will include exploring options for hosting all information digitally in one locality.

This action is likely to remain active for the life of the Action Plan as new and more effective medicines become available. Over the coming year, work will be undertaken to review the Managed Entry of New Medicines process including consideration of the current arrangements and the Individual Funding Request policy. Consideration will also be given to augmenting current processes in areas such as access to innovative medicines and participation in other procurement opportunities in conjunction with wider NHS colleagues.

Action 12: Improve access to other rare disease specialist teams locally, nationally and internationally

Whilst mechanisms are in place to access specialist teams, there is a need to develop clearer pathways and guidance to increase awareness of existing referral mechanisms for UK, all-island and wider/international rare disease specialist teams. Increasing awareness of the available processes for access to local, national and international specialist teams will have the added benefit of supporting local health professionals and patients to access tertiary specialist centres where local specialists are unavailable. Therefore a platform to hold and access this information is key to raising awareness amongst health professionals and patients.

Hence a key action for year one is to explore options for hosting all information regarding rare disease specialist teams digitally in one locality and providing clear instructions on pathways to tertiary specialist centres, making sure all relevant options are clearly detailed and accessible online by the end of the year. This action interconnects with links to the Information Hub, Expert Centre and Patient Portal, all of which may have long lead-in times, however given that the mechanisms and information already exist, the task for year one is to collect it digitally into one place until other platforms are enabled to migrate the information into their systems. Again, this action may be considered using a four nation approach.

Action 13: Improve awareness of and participation in rare disease research

Clinical trials and research studies are the means by which the safety and efficacy of new medicines, interventions and care approaches are tested, and hence are a route to early access to the newest treatments. Rare disease studies are often delivered through specialist centres and eligible patient numbers may be low in any geographical region, resulting in challenges for patients in terms of access to studies, and for study investigators reaching eligible patients. Innovative approaches to participation in studies are therefore needed.

Our NI rare disease clinical and academic communities have contributed to key research studies such as the Deciphering Developmental Disorders study in which 705 children from NI participated and 280 received a definitive diagnosis, and the 100,000 Genomes Project in which 440 individuals with rare disease participated, with 87 diagnosed on first pass analysis.

Overall management of information on the number of rare disease studies to which NI clinicians are recruiting patients could be improved, as well as the number of NI patients able to access studies and knowledge of available studies. A better understanding of current

levels of participation is a key first step towards maximising awareness, opportunity and participation.

It is important that clinicians and patients can access publicly available information about ongoing research, such as clinical trial registries and other research databases. Increased awareness has the potential to encourage uptake of suitable clinical trials and research studies amongst healthcare professionals. It is also important to create more transparency for patients around the challenges, complex mechanisms and limited entry opportunities for rare disease clinical trials and research studies.

Over the next year we will review and establish a current baseline for the number of rare disease studies openly recruiting patients in NI through the Northern Ireland Clinical Research Network, Northern Ireland Cancer Trials Network and beyond. We will also review participation of NI patients in clinical trials and research studies elsewhere to gain further understanding of the models by which participation is facilitated. On a regular basis we will also review clinical trial registries and research databases to create a clear picture of ongoing rare disease research studies, so that current opportunities for participation are known. Through other actions detailed in this plan, the information will be disseminated to healthcare professionals and patients (actions 7 and 10), and patients potentially suitable for ongoing studies will be actively identified (action 4). This will create a more co-ordinated approach to maximising opportunities for our patients to participate in clinical trials and other studies.

Through delivery of a communications plan, individual research studies will be highlighted on a regular basis, and case studies of participation in research will also be developed in partnership with patients. These will increase awareness of opportunities for both clinicians and patients, and provide exemplars of models of recruitment and participation which could be employed to facilitate an increased level of involvement in more studies. In preparation for medium-term actions beyond Year 1, other planned developments in the research infrastructure will be monitored in terms of increasing capacity and capability to deliver rare disease studies.

Whilst it is recognised that our healthcare system continues to meet significant resource challenges, opportunities for research training to increase the capability and capacity of our HSC rare disease workforce will be actively identified and disseminated, and support provided in identifying and securing appropriate funding for rare disease research studies.

A short term working group will be convened to review, refine and agree these activities for the next 12 months.

Action 14: Northern Ireland Rare Diseases Champion

There may well be many advantages to creating a Rare Diseases Champion role for NI to highlight and raise awareness of rare diseases both throughout the province and internationally. Recognising the good work being taken forward by, and replicating the success of the NI Mental Health Champion appointed in June 2020 in terms of advocacy, policy influencing and acting as a central point of contact.

Further exploration and consideration of this role is required and therefore this action will initiate a scoping exercise to explore / consider such a role, either standalone or jointly between statutory, university and voluntary representatives and any requirement for additional resources. The aim will be to develop outline proposals with an options appraisal.

Monitoring progress

The *2021 UK Rare Diseases Framework* commits to working closely with the rare diseases community to ensure that the Action Plans developed are actionable, measurable, and regularly reviewed.

Following publication of this Year 1 Action Plan, which has been co-developed with a diverse range of stakeholders including the NI Rare Diseases Partnership, the Northern Ireland Rare Diseases Implementation Group will continue to meet regularly to report on progress and over the next year will work on proposals for new and / or updated actions.

It is important to note that it will take time to make progress across all of the actions in this Year 1 Action Plan; some may be delivered in this first year and others should be considered as longer-term plans that need early work to fulfil the vision.

Progression of a number of the actions will require development of business cases or funding applications, all of which will be subject to normal approval processes, available funding and value for money.

We will also work closely with the four nation delivery/implementation groups to ensure that outcomes are identified and best practice shared throughout the duration of the plan.

Most importantly, we will continue to work with patient organisations and charities already performing valuable work under the four priorities of the Framework, and ensure through ongoing dialogue that people with rare diseases remain at the heart of the decision-making process.

Summary of actions

Annex A

Framework Priority	Action	Key milestones & measures Year 1	Related Year 1 Action Plan actions
1. Helping patients get a final diagnosis faster	**1. Information Hub** An online Rare Disease Information Hub established for NI, with a dedicated person employed to: • collate relevant information; • act as a contact point and source of advice for people living with a RD in NI and their families; • connect those working in the RD field.	• Scoping Group established. • Scoping review with recommendations for a best practice model. • Identification of resources / budgets required. • Secure funding for a dedicated person/s (to include RD navigators) to take the Hub work forward through relevant business case or funding application to appropriate funding organisation.	7. Expert centre 9. Mental health needs 10. Patient portal
	2. Agree a new NI Genomics Partnership model to deliver against the UK Genome Strategy Genomics Medicine Service established in NI, working with delivery partners across government, HSC, academia, industry, patients, and ROI counterparts, to agree future vision and policy decisions on genomics development and provision in NI, linking and collaborating with the UK nations in line with the commitments of Genome UK.	• Develop governance structure for Regional Genomics Medicine Service, in partnership with key delivery partners, including links with ROI. • Genomics working group agree governance structure and accountability. • Ministerial approval to commence with new Service; • By end of year one, to have held the first meeting of the new NI Genomics Medicine Partnership.	8. Care pathways/models 11. Improve access to RD drugs 12. Improved access to other RD specialist teams locally, nationally and internationally
	3. Review newborn screening Newborn screening to be reviewed in line with the population needs of NI.	• Participate in UK National Screening Committee (NSC) - follow guidance to ensure appropriate use of genetics in line with NSC recommendations. • Establish Task & Finish Group (including patient voice) to scope new-born screening needs for NI, taking geographical prevalence of conditions on an all-Island basis into account. • Produce a scoping review to include recommendations for optimised screening for conditions in NI. • Ensure NI representation in all new UK screening initiatives.	

2. Increasing awareness of RD among healthcare professionals	**4. Develop a Northern Ireland Rare Diseases Registry** A NI RAre Disease and Congenital Abnormality Registry (NIRADCAR) to be established and linked to the new Encompass Integrated Care Record.	• Quarterly devolved nations meetings with registry colleagues to work towards a UK-wide national rare disease registration facility. • Registry Lead appointed and Registry Steering Group established. • Identify how best to obtain a minimum core dataset and standardised coding for the registry. • Develop inclusion criteria and analysis plans, taking account of stakeholder input from all relevant sectors, ensuring individual condition registry specialists are included. • Scope logistics for accommodating the NI rare disease registry • Work with the NI cancer registry to generate the first factsheet for rare cancers in NI. • Source registry information for 1-2 individual RD conditions associated with exemplar care pathways. • Scoping review completed and business case developed to include necessary resources to take registry work forward.	1. Information Hub 7. Expert Centre 8. Care pathways/models 9. Mental health needs. 10. Patient portal 13. RD research awareness/ participation
2. Increasing awareness of RD among healthcare professionals	**5. Education & Training - pre & post registration. Education co-ordinator** Build on links with external organisations providing training.	• Education & Training Steering Group established to include relevant stakeholders, with emphasis on PPI/Co-production, design, and delivery. • Secure funding for a dedicated person to take the E&T work forward. • Cost / benefit analysis completed to support a bid for additional resources. • Ensure CPD accreditation for postgraduate training events. • Representatives to promote/disseminate CPD event. information through websites and social media channels. • Build on existing RD awareness sessions and deliver 12 sessions per year in partnership with relevant bodies. • Embed delivery of RD teaching sessions, including the patient voice, to all health care professionals, including undergraduate medical, biomedical, nursing and AHP students, as well as postgraduate taught students.	1. Information Hub 4. Registry 6. Increase awareness across communities 7. Expert Centre 8. Care pathways/models 9. Mental health 10. Patient portal 11. Improve access to RD drugs 12. Improved access to other RD specialist teams 13. RD research awareness/ participation

2. Increasing awareness of RD among current and future healthcare professionals, policy makers and wider society.	**6. Raise awareness of rare diseases across a range of communities** Increase the profile and public awareness of rare diseases through public events such as Rare Disease Day, RD Campaigns, and establishment of an All-Party Group on Rare Disease.	• NIRDP support to the All-Party Group for RD. • Host Annual All-Ireland Rare Diseases Day Conference. • Host at least 2 rare disease focused STEM initiatives in 2022. • Promotion/ dissemination of information on events through websites and social media channels. • Bi-monthly All-Ireland RD webinars through All Ireland Students for RD network. • Quarterly RD discovery research meetings planned with both professional /public components. • Deliver 12 webinars, information sessions or campaigns to raise awareness amongst the wider community. • Implement training initiatives to empower patient group representatives to be active voices for their own groups and broader RD community in all rare areas, but particularly for more complex areas, e.g. research and development.	5. Education & Training Other Actions links as per Action 4
3. Better co-ordination of care	**7. Develop an expert centre for Rare Diseases to include RD specialisms co-ordinator** Develop a national rare disease care centre acting as a central point of contact for rare disease across NI. Appoint a rare disease specialisms information co-ordinator.	• Scoping group captures the NI landscape plus further afield and develop an options appraisal. • Determine requirement for physical space – consider hybrid model. • Develop a Business Case / implementation plan for developing an expert centre and specialism coordinator.	1.Information hub 2. Genomics 3. Registry 8. Care pathways/ models 9. Mental health needs 10. Patient portal

3. Better co-ordination of care	**8. Develop care pathways / models of care.** Access to services in the UK & elsewhere. Define "what a better care model looks like". More patient information on all available services, including transition from paediatric to adult care. Clear patient information to enable understanding e.g., infographics & lay terms/non-medical terminology.	Establish a working group to: • Develop a pathway for individuals with suspected, but not yet diagnosed RDs, for example through regular gene discovery clinics. • Develop a methodology to deliver exemplar care pathways for 1-2 individual RD conditions in the first year with a view to use as a template for other RD conditions. • Engage with/ recruit RD clinicians to participate in the 'clinician builder programme' to develop skills for clinical input to the building of RD pathways, including 'red flag' signposting and other needs. • Recruit and work with Paediatric, Adult RD Clinical Leads as well as other individual clinical leads to push forward work on agreed pathways/ defined service specifications. • Continue to improve a virtual & face to face mixed model for consultations to connect patients with the clinicians they need. • Scope existing individual RD voluntary organisation work in this area to determine best practice examples.	Action 8 is important to all other Actions and collaboration is required with all working groups, depending on the individual item under consideration.
3. Better co-ordination of care	**9. Ensure the mental health needs of rare disease patients and carers are included across all appropriate NI Government strategies and programmes.** This includes provision of support in areas wider than the health service, for example social care, housing and education. Within Mental Health services, to focus on enhancing wellbeing and providing counselling support for people with rare diseases. To consider CAMHs being open to patients through the hub. Clinicians enabled to refer directly into Mental Health services.	• Largely included in the Information Hub, the Expert Centre and developing pathways and models of care, and should be embedded in those actions. However, given the importance of MH for RD and Action 22 of the MH Strategy, this will remain as a standalone Action to ensure it is appropriately addressed. • This may include scoping appropriate links through other Actions to ensure that clear pathways are identified between all relevant support organisations. • Identify what support functions already exist within healthcare provision and the community & voluntary sector, and determine where gaps exist (gap analysis).	1.Information hub 2.Registry 5. Education & Training 7. Expert centre 8. Pathways/models 14. RD Champion

3. Better co-ordination of care	**10. Patient portal** A "one stop shop" where a patient can view information, with access to all communications, healthcare professionals, summary care record and emergency care record.	• Work / liaise with relevant people in working groups across other Actions to ensure input is provided. • UK RD Framework is considered by the NI Digital Strategy (encompass) team during the design, build and rollout of the programme. • Liaise with encompass to familiarise potential users (clinicians and patients) with the functionality of MyChart. • Ensure appropriate information (leaflets/online instruction) is available for RD patients. • encompass team to carry out a RD specific public engagement event to provide an update on the current position of encompass and how it may benefit the RD community in terms of a patient portal.	1. Information hub 4. Registry 7. Expert centre
4. Improving access to specialist care, treatments, and drugs	**11. Improve access to Rare Disease drugs** Ensure equity of access to RD medicines locally, regionally, and nationally by increasing health professional and patient awareness of the existing mechanisms to access available initiatives.	• Ensure appropriate RD representation and contribution (including patient voice) during the development of policy processes around access to, and managed entry of medicines, including the Individual Funding Request mechanism, into the NI healthcare system. • Working with delivery partners, produce information resources and a clear roadmap for available initiatives. • Explore options for hosting all information digitally in one locality. • Over the coming year, work will be undertaken to review the Managed Entry of New Medicines process and augmenting current processes such as access to innovative medicines and participation in wider procurement opportunities.	7. Expert centre 8. Care pathways/ models 10. Patient portal
	12. Improve access for clinicians and patients to other rare disease specialist teams – locally, nationally, and internationally Improve awareness of available processes for access to local, national, and international specialist teams.	• Explore digital hosting options for all information. • Ensure all relevant options are clearly detailed and accessible online by end of year. • Provide clear instruction on pathway to tertiary specialist centres.	1. Information hub 7. Expert centre 8. Care pathways/ models 10. Patient portal

	Provide support for local health professionals and patients to access tertiary specialist centres where local specialists are unavailable. Develop clear mechanisms and/or increase awareness of existing referral mechanisms for UK, all island and wider/international.		
	13. Improve awareness of and participation in rare disease research Increase health professional awareness of NI Clinical Trials Network and other UK clinical trial databases. Encourage uptake of suitable / available clinical trials amongst healthcare professionals. Create transparency for patients around the mechanisms and entry opportunities for clinical trials. Advise suitable NI patients about current clinical trials in NI.	Establish a short working group to refine and review the following tasks by the end of the year: • Establish a baseline of current rare disease studies recruiting NI patients. • Review participation of NI patients in clinical trials/research elsewhere and understand the models for participation. • Regularly review clinical trials and research databases for opportunities to participate. • Develop a communication plan to disseminate information on opportunities to participate. • Where possible, work with other UK nations to scope and map information and data related to rare disease clinical trials and research studies.	1. Information hub 7. Expert centre 8. Care pathways/ models 10. Patient portal
	14. Champion for Rare Diseases in NI	• Scoping exercise to explore/ consider role and requirement for additional resources. Develop outline proposals with an options appraisal. • Consider joint role between statutory/university/voluntary sector reps.	5. Education & Training 6. Increase awareness across communities

Annex B

Northern Ireland Rare Diseases Implementation Group members / core delivery partners

Department of Health

- Chief Scientific Advisor (Chair)
- Secondary Care Directorate (Deputy Chair, policy lead and secretariat)
- Adult Mental Health Unit
- Social Services Policy Group

Health and Social Care Board

- Specialist Commissioning
- Pharmacy
- Adult Social Care

Public Health Agency

- Research & Development
- Child Health Partnership

Health and Social Care Trusts

- Clinicians (paediatric and adult) across a range of specialties

encompass

- Northern Ireland digital integrated care record initiative team

Northern Ireland Rare Disease Partnership

- Chief Executive
- Chair
- Patient representatives
- Board member/s

Queen's University Belfast

- Researchers
- Educators

Ulster University

- Researchers
- Educators

Abbreviations and definitions

APG	All party group	A cross party group of all the main political parties based at the Executive in Stormont
CARDRISS	Congenital Anomaly and Rare Disease Registration and Information System	The Scottish rare diseases and congenital anomaly registry
CARIS	Congenital Anomaly Register and Information Service	The Welsh rare diseases and congenital anomaly registry
CPD	Continuing personal development	Process of maintaining & enhancing knowledge, skills and experience relating to professional activities following completion of formal training
encompass		A health and social care initiative that will introduce a digital integrated care record for every person in Northern Ireland
Framework	*UK Rare Diseases Framework*	Published in January 2021
GB	Great Britain	England, Scotland and Wales
GenOCEANIC	Genomics Open Core Engine for Accelerating Northern Ireland Care	A tool which helps streamline genetic testing
GMS	Genomic Medicine Service	Doctors and genetic counsellors who see people with suspected genetic conditions
GP	General Practitioner	Family doctor
HSC	Health and social care	The health service in Northern Ireland
HST	Highly specialised technology	HST evaluations are NICE recommendations on the use of new & existing highly specialised medicines and treatments within the NHS
Key stage 5	Advanced level student	A'level or 6th Form students (16-18 year old)
NCARDRS	National Congenital Anomaly and Rare Disease Register	The English rare diseases and congenital anomaly registry
NI	Northern Ireland	
NICE	National Institute for Health and Care Excellence	NICE provides national guidance and advice to improve health and social care
NIRADCAR	NI RAre Disease and Congenital Anomaly Register	The Northern Ireland rare diseases and congenital anomaly registry
NIRDIG	NI Rare Diseases Implementation Group	Responsible for developing, agreeing and monitoring implementation of a Rare Diseases Action Plan for Northern Ireland. Led by the Department of Health with representatives from commissioning and delivery partners and stakeholders to ensure a unified approach.
NIRDP	NI Rare Diseases Partnership	Umbrella group representing rare diseases community
Orphanet		Online portal providing high quality information on rare diseases, experts networks, & associated support groups
PPI	Personal & Public Involvement	Term used to describe active and meaningful involvement of service users, carers, their advocates and the public in the planning, commissioning, delivery and evaluation of HSC services

PHA	Public Health Agency	A multi-disciplinary, multi-professional organisation with four key functions: health & social wellbeing improvement; health protection; public health support to commissioning and policy development; and, health and social care research and development
QUB	Queen's University Belfast	
RoI	Republic of Ireland	
RMDS	Regional Molecular Diagnostic Service	Laboratory undertaking genetic testing for Northern Ireland
STEM	Science, technology engineering and mathematics	An education program which encourages young people to understand the opportunities that a STEM-based career can offer them
UK	United Kingdom	England, Northern Ireland, Scotland & Wales
UKNSC	UK National Screening Committee	Advises Ministers and the NHS in the 4 UK countries about all aspects of population screening and supports implementation of screening programmes.
UU	Ulster University	
TA	Technical appraisal	TAs are recommendations on the use of new and existing medicines and treatments within the NHS.
100,000 Genomes Project		Transformational NHS research project evaluating new genetic tests for rare diseases

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