Rare Disease Priorities for Progress

Within Northern Ireland: Workshop Report

AJ McKnight & Grace Henry with multidisciplinary stakeholders: 28th May 2020

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Executive Summary

Rare diseases are a major public health concern, cumulatively affecting ~6% of the population. In 2013, the UK strategy for rare disease was published[1] followed by the Northern Ireland rare disease implementation plan (NIRDIP) in 2015[2]. In response to priority actions identified within the Northern Ireland Rare Disease Implementation Plan, three complementary (confidential) commissioned reports were submitted to the Northern Ireland Rare Disease Implementation Group (January 2020) describing recommendations for action.

1) Rare disease communications and information review[3]. **Communications and Information Review: NI Report for Rare Diseases**

2) Rare disease education and training review[4]. **A rapid scoping review of rare disease education needs for NI**

3) Research towards building a rare disease registry[5]. **Perspectives on a NI Rare Disease Registry**

A focused, knowledge exchange workshop was held with multidisciplinary stakeholders to help prioritise existing recommendations across all three documents and generate a report to inform policy and practice for future rare disease progress. A NI rare disease implementation / action plan is being discussed for beyond 2020, which may be informed by the outcomes of this workshop.

Initially, individual recommendations were consolidated to minimise duplication between each report. It was accepted that the consistently top-ranked recommendations remain the top 3 priorities for rare disease progress in NI: (1) to develop a national registration service, (2) to develop a dedicated online information hub, and (3) to develop a role to appoint a dedicated information coordinator. Six recommendations already have work underway, so workshop participants ranked the remaining recommendations as priorities for action within the short, medium and longer term prior to workshop discussions. This provided a framework for initial discussions during the workshop.

Following considerable discussion as to the best way to prioritise recommendations, workshop participants ranked 34 recommendations by focused themes with a focus on 'registry', 'information and communication', 'training and education', as well as a cross-cutting focus. Prioritisation included ranking on short-, medium- and longer-term goals. However, it was proposed that there may be a better approach to generate progress for our rare disease community.

The final approach taken by workshop participants was to consolidate recommendations under the proposed NIRADCAR (Northern Ireland RAre Diseases & Congenital Abnormalities Registry) and information hub within a Rare Disease Centre. Streamlined priorities with suggested deliverables have been suggested to support rare disease progress within Northern Ireland.

[1] Department of Health (2013). The UK Strategy for Rare Diseases. Retrieved from https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/26056 2/UK_Strategy_for_Rare_Diseases.pdf

[2] 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 2015. https://www.health-ni.gov.uk/sites/default/files/publications/dhssps/ni-rare-diseases-implementation-plan-oct-2015.pdf (accessed 01/01/2018).

[3] McKnight, A. J., McMullan, J., Walker, R., & Collins, C. (2020, Jan 31). Communications and Information Review: NI Report for Rare Diseases. DOI 10.17605/OSF.IO/PUH5B

[4] McMullan, J., Moore, K., & McKnight, A. J. (2020, Jan 31). A rapid scoping review of rare disease education needs for NI. DOI 10.17605/OSF.IO/UJ9KR

[5] McMullan, J., Crowe, A., Kerr, K., Bailie, C.... McKnight, A. J. (2020, Jan 31). Perspectives on a NI Rare Disease Registry. DOI 10.17605/OSF.IO/XU7P

Workshop Participants

Amy Jayne McKnight (host)

Dr Amy Jayne (AJ) McKnight has a keen interest developing excellent research and improving resources for individuals living and working with rare diseases. AJ has worked for rare disease communities for ~40 years, more recently extending this interest into formal research at QUB where we are building an enthusiastic and productive rare disease research team.

Grace Henry (facilitator)

Grace Henry has a background in nursing and health visiting and has worked at a senior level in the voluntary sector for over 30 years, with a particular interest in older people's issues. She was formerly a member of the board of trustees for Huntington's Disease Association Northern Ireland where she developed an interest in rare diseases. She has had the pleasure of facilitating workshops and supporting several projects in relation to rare disease strategy development over the last number of years.

Caitlin Bailie

Caitlin Bailie (Montgomery) is currently undertaking her PhD with Prof Maxwell and Dr McKnight. She began her PhD looking at molecular biomarkers of kidney disease. After taking some time out with her growing family, Caitlin is now extending her interests to rare disease issues and evaluation of the 100,000 genomes project with the hopes of moving towards a career in genetic counselling.

Tanya Boggs

Former Stronger Together Project Officer for the Western and Northern Trust areas. Tanya is a nurse with over 25 years' experience, most recently working in paediatric palliative care at Great Ormond Street Hospital for Children NHS Foundation Trust. Returning to NI after a long absence to undertake a job with NIRDP has been lifechanging, and she looks forward to continuing to champion healthcare for all, but especially those most vulnerable.

James Caldwell

James is the treasurer of NIRDP – responsible for finance and company secretarial matters. While NIRDP is an umbrella group for all rare diseases, his particular interest is rare cancers. James is a rare cancer patient, diagnosed in 2014 with cancer of the appendix. In 2014 he had major surgery and systemic chemotherapy. He is also a member of the European Cancer Patient Coalition (ECPC) working group on rare cancers and hasattended a number of patient advocacy events throughout Europe. He is a patient advocate to EURACAN, the ERN on Rare Adult Solid Cancers. James is quite active on a number of social media groups that provide advice and support for appendix cancer patients and those with PMP (Pseudomyxoma Peritonei). By profession, James is a Chartered Accountant and Registered Nurse.

Sandra Campbell

Sandra is currently Lecturing in NWRC in Early Years and Special Needs. She has worked across many sectors of Education for 30 years. Sandra's interest in rare disease began when her beloved father was diagnosed with PSP in 2000. She worked as a Volunteer for PSPA for five years then took up post as PSPA Development Officer for NI & ROI for six years. During this time, she worked with other Rare Disease Charities which then officially launched as NIRDP with Christine Collins' leadership. She has been involved with NIRDP since then and now Volunteers in NW to facilitate a local group.

Christine Collins

Christine Collins MBE retired from the Civil Service in 2005 and since this time has immersed herself in the world of human rights, equality and advocacy; with a particular focus on those living with disability and/or rare disease. Christine was the founding Chair of the Northern Ireland Rare Disease Partnership; and a Patient Public Voice representative on the UK Rare Disease Advisory Group from its inception until April 2018. She was a Northern Ireland Member of the UK Rare Disease Forum. Christine has been involved in rare disease for many years and is actively involved in the development and implementation of both the UK and Northern Ireland RD plans. Christine was a Commissioner at the Northern Ireland Human Rights Commission from 2011 to 2017. She is currently Chair of the Patient and Client Council and a member of the Duty of Candour Work Stream, part of the Inquiry into Hyponatremia Related Deaths Implementation programme.

Ashleen Crowe

Ashleen Crowe is undertaking her PhD with Dr McAneney & Dr McKnight, which is titled, 'Improving communication and identifying priorities for modernising rare disease services with healthcare practitioners, patients, families and advocacy groups'.

Tabib Dabir

Dr Dabir has been a Consultant in Clinical Genetics since 2007 and Clinical lead for Northern Ireland Regional Genetic Centre since 2016. He has recently been appointed as Clinical Director for Molecular Diagnostics and Microbiology. Dr Dabir has extensive clinical and academic experience of working as Paediatrician and Clinical geneticist in India, Saudi Arabia, Dubai and UK. He was instrumental in setting up regional clinics for Von Hippel- Lindau (VHL) Syndrome and 22q 11.2 deletion syndrome patients at Belfast City Hospital

William Duddy

My interest in rare diseases stems from the experience of having a brother with the muscle wasting disease, Duchenne muscular dystrophy. This experience pushed me towards a career in science and research. After some 18 years at research institutes in Scotland, the US, and France, I recently returned home to join the University of Ulster's Northern Ireland Centre for Stratified Medicine. As part of the neuromuscular research team, I am looking for new ways to understand and treat a range of rare conditions affecting the body's ability to control and maintain its muscle mass. Our focus is on the role of muscle in motor neuron disorders such as Amyotrophic Lateral Sclerosis (ALS). I learned about the NIRDP first through fellow researchers in the area, and I think that the NIRDP's work is important because rare diseases are not rare at all when you add them all together!

Stephanie Duguez UU

The first time I heard about rare disease was as a teenager, watching the French Telethon. The Telethon introduced me to neuromuscular conditions, and how genetics could help us to understand these diseases and identify therapeutic targets. Ever since then, I have wanted to study neuromuscular disorders, and work as a scientist in this field. During my career, I have worked in different institutes (LPEH Saint Etienne, France, Genethon France, CNMC Washington DC, Institute of myology France) on muscle cellular physiology, Duchenne Muscular Dystrophy and then motor neuron diseases. I recently joined the University of Ulster's Northern Ireland Centre for Stratified Medicine, where I lead the research group in neuromuscular health. Our group focuses on how the muscle communicates with other cells in the body, and its role in motor neuron disorders such as Amyotrophic Lateral Sclerosis (ALS), spinobulbar muscular atrophy (SBMA or Kennedy disease) and SMA-IV. Having worked for the associations created by the French Telethon, I'm very much aware that rare diseases collectively are frequent, and that solidarity is a key point to fight rare diseases. I also understand the necessity to make people aware of rare conditions. I have been introduced to NIRDP through fellow researchers in the area and I'm convinced that the work done by this group is crucial to improving patient care in Northern Ireland.

Alex Duncan – with apologies due to ill health.

Claire Kerr

Dr. Claire Kerr is a Senior Lecturer in Rehabilitation and a physiotherapist. Her research aims to improve the health, well-being and provision of clinical services for children and young people with physical disabilities, in particular, those with cerebral palsy. Her work explores the efficacy of therapeutic interventions, the use of measurement tools and service provision for this population, as well as knowledge translation with allied health professionals that work with children with cerebral palsy and their families.

Fiona McLaughlin: Fiona lives in Belfast with her family, a cairn mix terrier, and Myalgic Encephalomyelitis (M.E.) She had to give up work in the voluntary sector in 2007, and has been volunteering when able since then. Her late mother had a rare neurological illness, which meant she learned a lot about how our health and care systems do and don't work. She has been involved with the PCC since 2010, working on issues around rare disease, neurology and M.E. She is a former Chair of Northern Ireland Rare Disease Partnership (NIRDP).

Julie McMullan

Dr Julie McMullan is a post-doctoral research fellow carrying out research around communication and education of rare diseases. Julie enjoys meeting the people 'on the ground' and hearing their stories and experiences. Julie is keen that the work she does raises awareness of rare diseases as well as contributing to improving care in Northern Ireland and further afield.

Kerry Moore

Information Project Officer of NIRDP (funded by The Rank Foundation). Kerry is a research scientist who has worked with Dr McKnight as a PhD student, research associate and post-doctoral research fellow. Kerry has extensive experience of working with patients, overseeing research projects and teaching. She has also worked in diagnostic genetics and was a research associate at the Northern Ireland Cancer Registry. She aims to help build an informative research resource on the NIRDP website while developing a library of practical information for patients, carers, scientists and healthcare professionals alike. Kerry has three young children to keep her busy in her spare time.

Julie Power

Prior to diagnosis in 2005 with a rare disease called Granulomatosis with Polyangiitis Vasculitis, Julie worked for 17 years as an Occupational Therapist, specialising in Neurology and Palliative care. Following diagnosis, her life and that of her family's was totally changed. The uncertainty and isolation Julie experienced on her patient journey has fuelled her interest in raising awareness, improving care and research. Julie believes knowledge is power and that patient involvement throughout the entire treatment and research process is mutually beneficial to the Researchers, Clinicians, Service Providers and Patients.

In 2010, Julie founded Vasculitis Ireland Awareness, an All-Ireland support group for anyone affected by Vasculitis in Ireland, liaising closely with Vasculitis UK and the Vasculitis Foundation. Julie is a patient representative in the Irish Rare Kidney Disease Registry and Bio bank Steering Committee, the recently formed Vasculitis Ireland Network (VINE) and in planning for a pilot NI Vasculitis service. She graduated as a EUPATI (European Patient Academy in Therapeutic Intervention) fellow in 2016 after completing the 14month intensive research and development course. Julie has been a member of the Northern Ireland Rare Disease Partnership (NIRDP) board of directors since 2015. This year, she was invited onto the Irish Platform for Patient Organisations Science and Industry board (IPPOSI) and is actively involved in improving care for those affected by rare disease both in NI and ROI.

Sam Robinson

Amy Jayne got me involved with her work within The Rare Disease arena even before I knew I had a rare disease myself (Periventricular Nodular Heterotopia). I still like to live life to the full, enjoying fishing, dog walks, walking on the beach in all sorts of weather, and even like my 'day job' running my own business that involves many sorts of work. I've been involved in running Rare Disease Day events and helping with lots of other meetings, particularly bringing awareness or rare diseases to the lower Ards and North Down. Looking forward to new challenges in the future.

Rhoda Walker

Rhoda came to volunteer with the Northern Ireland Rare Disease Partnership nearly two years ago, four years after her son Robbie was diagnosed with Ehlers Danlos Syndrome. So little support was provided to them following diagnosis and so their journey to be more knowledgeable about the condition was quite a frustrating one. Her passion for the Partnership's vision that "no one is disadvantaged because of the rarity of their condition" stems from seeing first-hand the day to day difficulties that her son faces. Whilst his condition is rare, his struggles are not! As every patient with a rare condition has detailed, they all face the same problems within the existing medical and benefit systems, systems that have limited mechanisms to handle complex issues. Rhoda has over twenty years' experience in community development and has been working as a freelance community development facilitator since 2016, with volunteering experience across a range of organisations. Whilst with the Partnership she would like to help identify ways to improve user involvement in every aspect of programming which supports their lives from research and development, design and production through to delivery. By improving sharing information and effective communication we can support the professionals to simply the issues that affect the lives of those living with a rare condition. She is keen to continue the good work of the partnership in developing collaborative working to ensure that all available resources are best used to meet the community's needs.

Summary of the three individual reports

A rapid scoping review of rare disease education needs for NI

Individuals living and working with rare diseases often voice concerns about healthcare and non-medical professionals not being sufficiently informed and educated about rare diseases. While professionals will have considerable expertise within their own specialist areas, few have resources to thoroughly research rare diseases and many report being overwhelmed as patient's attend with complex health and social care needs. While this may involve the challenge of not knowing where to find accurate, timely information, it is complicated by rapid technological advances and mainstreaming of genomic medicine across the UK.

Rapid laboratory and computational advances are providing significant reductions in the time for many individuals to receive an accurate diagnosis of rare diseases with a genetic cause. However, such tools will not be effective unless there is the right level of accompanying education to enable our existing NHS workforce to consent individuals in this era of genomic medicine, interpret whole genomes sequencing results, and deliver best outcomes for patients. Training for healthcare professionals using genomics and bioinformatics in the NHS will be an evolving programme of workforce development and transformation across multiple levels. Urgent training is required in the short-term for five distinct workforce groups:

(1) clinical and research staff involved in recruiting patients / consenting individuals for whole genome sequencing as part of Northern Ireland's participation in the 100,000 genomes project and beyond, capturing core information sets, and sharing data within relevant ethical and governance guidance.

(2) specialist healthcare scientists such as NHS clinical genetic scientists, molecular pathologists, and bioinformaticists to enable them to handle genome sequencing data and generate interpretive clinical diagnostic reports;

(3) clinical staff involved in delivering results to enable them to interpret clinical diagnostic reports, deliver results to patients that may include unexpected findings, and take appropriate action to maximise patient care;

(4) general healthcare professionals who are not presently consenting individuals or delivering whole genome sequencing results to raise awareness of genomic medicine, red flags to consider rare disease investigations, and improved patient care.

(5) non-medically trained health and social care professionals supporting individuals living and working with rare diseases.

The development of ENCOMPASS as Northern Ireland's digital integrated care record has significant potential to improve rare disease diagnosis and patient care, but customised training will be required to maximise outcomes for rare diseases from this initiative. In the longer-term, ongoing training is essential to accommodate increasing numbers of individuals eligible for whole genome sequencing data, multidisciplinary genomic interpretation, the implementation of multiomic (transcriptomics, proteomics, metabolomics, epigenomics) analysis, and the incorporation of developing models of care for rare diseases. Developing education and training tools should be flexible to allow for convergence of individual specialisms and provide future healthcare professionals with an appropriate skill mix to improve diagnosis and care for rare diseases.

Communications and Information Review: NI Report for Rare Diseases

Rare diseases are characterised by their relatively low prevalence individually, but cumulatively they are common, affecting one in seventeen persons at some point in their lives. It is estimated that 450 million people globally have a rare disease with approximately 110,000 people affected by a rare disease across Northern Ireland. Being diagnosed with a rare disease can lead to a significantly reduced lifespan and quality of life for patients and their families. Patients, carers, health and social care professionals consistently describe challenges accessing accurate information about individual rare diseases and accessing appropriate services. Individuals living with rare conditions often describe feeling 'isolated' and 'dismissed' so improved communication strategies are urgently required. Improved provision of information has been embedded in many national plans for rare diseases, including the Northern Ireland Rare Disease Implementation Plan.

While there are many charities with dedicated patient advocacy services, members of the rare disease community often struggle without a dedicated charity for their rare disease. Many rare disease patients (and / or their family or carers) need to source information and promote best care for themselves, without any training in how to do so. They frequently report healthcare professionals and service providers being unfamiliar with their rare disease or relevant rare disease resources for information and support. This report reflects feedback from people diagnosed with rare disease(s), people still searching for a diagnosis, their families, carers, and support teams.

Beyond the initial communications review envisaged in the NI Rare Disease Implementation Plan, information has been developed for healthcare professionals, patients living with rare diseases (Stronger Together; RARDTAC), and those helping to care for them. However, there is a strong and continued call for a NI Information Hub for Rare Diseases, in the form both of an online guidance tool, and of "a person at the end of a telephone", to stabilise information provision and provide an element of sustainable coordination for this isolated and fragmented community.

Perspectives on a NI Rare Disease Registry

Rare Disease registries are essential components of improving healthcare for the rare disease community. Historical challenges of developing such registries are minimised with technological advances, interoperable systems, and collaboratively shared expertise. This report was generated to help inform the NI rare disease stakeholder implementation group, including developments from ROI, England, Scotland and Wales. The need for a coordinated and cohesive approach to information on rare diseases was identified as a priority in the consultation process leading to the development of Northern Ireland's Implementation Plan; and this is reflected in the Priority Action "To complete a database review and produce a costed action plan to implement a Northern Ireland register of Rare Disease".

The request for a sustainable, NI rare disease registry has been consistently reported in the top three priorities from healthcare professional, industry, public and patient stakeholder / open meetings. Work has been completed to identify existing data sources (31 rare disease clinical / research 'lists', several of which are in danger of being permanently lost), and scoping how a Rare Disease Registry for Northern Ireland can be progressed in line with NI's unique legislation and health and social care information systems. A rare disease registry for Northern Ireland with clinical and administrative data linkages were requested by multiple groups, but many have insufficient resources (funding, people, skills, or computational) to create this themselves.

Many names were suggested for such a NI registry, with the majority voting for, **'NIRADCAR: Northern Ireland RAre Diseases & Congenital Abnormalities Registry'.** From the >8,000 rare diseases described in the literature, ~80% have a molecular cause. While the majority of rare diseases (84.5%) are very rare with a prevalence less than one million individuals, a recent review of Orphanet revealed that more than 98% of people diagnosed with a rare disease have one of the 390 less unusual rare diseases, with ~80% of the population burden of rare disease believed attributable to ~150 rare diseases. Almost 70% of rare diseases have an exclusively paediatric onset with one-third of rare disease patients dying before their fifth birthday. Approximately 15% of individuals with a rare disease have a congenital abnormality; the majority of congenital anomaly phenotypes reported in UK registers are considered rare diseases. Collecting data on rare diseases and congenital anomalies is challenging due to the often diverse, fragmented sources of original information. This paper describes initial consultations supporting the design of a rare disease registry that collects data along a longitudinal care pathway in near-real time, with an emphasis placed on the ability to link data from a wide range of sources.

Background to recommendations

Recommendations were provided within each individual report (Appendix 2: Original recommendations from the three reports), but there was considerable overlap in the issues and options discussed. Below tentatively summarises these recommendations into ongoing, short, medium, and longer-term recommendations in advance of the workshop. Several of these recommendations were suggested in 2017 and are already being successfully delivered. Several require minimal financial investment while others require major investment of resources. Within each timeframe, recommendations are also group under 'education and training', 'information and communication', 'registry', and 'multi-foci' themes. Under each recommendation, green text highlights work ongoing in this area while red text has been identified as a mechanism for potential delivery.

Education and training focus

The Northern Ireland implementation plan for rare disease reiterates the UK-wide strategy to, *"support education and training programmes that enable health and social care professionals to better identify rare diseases to help deliver faster diagnoses and access to treatment pathways for patients*". Reflecting the importance of improved education for healthcare professionals, national rare disease plans in England, Scotland, Wales and the Republic of Ireland[6,7,8,9,10] all support commitments to improvements in this area.

Northern Ireland Implementation Plan for Rare Disease[3]: education of healthcare professionals components

"Commitment 2.2: *the Department will incorporate awareness of rare diseases into training for its clinicians and service providers*

Commitment 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*

Commitment 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".*

[6] Department of Health Northern Ireland. Providing high quality care for people affected by rare diseases – the Northern Ireland implementation plan for rare diseases. 2015. https://www.health-ni.gov.uk/sites/default/files/publications/dhssps/ni-rare-diseases-implementation-plan-oct-2015.pdf (accessed 01/01/2018).
[7] Department of Health and Social Care UK. The UK Strategy for Rare Diseases: 2019 update to the Implementation Plan for England In: Department of Health and Social Care UG, editor.; 2019.
[8] Government Scotland. It's Not Rare to Have a Rare Disease: The Implementation Plan for Rare Diseases in Scotland. 2014.
[9] Government Wales. Welsh Rare Disease Implementation Plan. 2017.
[10] Department of Health Ireland. National Rare Disease Plan for Ireland: 2014-2018. https://www.gov.ie/en/publication/a4ac1b-national-rare-disease-plan-for-ireland-2014-2018/ (accessed 01/01/2018)

Information and communication focus

The UK Rare Disease Strategy published in 2013 sets out the following recommendations describing information, emphasising the importance *'making sure that patients, their families and professionals can get the information they need; e.g. how to give all patients with a rare disease clear and timely information about: their condition and its development; treatment and therapy options; practical support'*.

UK Strategy recommendations:

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

5. consider how to give all patients with a rare disease clear and timely information about: their condition and its development; treatment and therapy options; practical support

6. improve access for patients (or where appropriate their parents or guardians) to their personal data

12. work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients, by – establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and 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 pathway – embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies

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

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

30. identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments

31. look at how the 4 UK countries develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways

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

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,

The Northern Ireland Rare Diseases Statement of Intent was released in 2014, with our Northern Ireland Rare Diseases Implementation Plan published by the Department of Health, Social Services and Public Safety (DHSSPS) in October 2015[11]. Recognising that accurate information, effective timely information, and engagement of healthcare professionals, patients, carers, practitioners, and policy makers are cornerstones of progress for rare diseases, a communications review was conducted by the Northern Ireland Rare Disease Partnership and researchers within the Centre for Public Health at Queen's University Belfast. National rare disease plans all support improved information sharing, communication, and more effective patient / public / participant engagement.

Priority Action from the NI Rare Diseases Implementation Plan:

a) *To 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.*

b) *To complete a communications review and introduce enhanced methods of communication*

In the Republic of Ireland, a priority recommendation from the National Clinical Programme for Rare Diseases in line with the National Rare Disease Plan for Ireland (2013) was to establish a National Rare Diseases Office, which was launched on 4th June 2015, along with a developing rare disease website. The Irish National Rare Diseases Office primarily provide information about specific rare diseases, relevant clinical expertise available, and research opportunities, acting as the central point of contact for Orphanet[12]. They also connect people with clinical networks, patient support groups, and social care support contacts. They are helping develop national rare disease care pathways and regularly consider the development of rare disease registries.

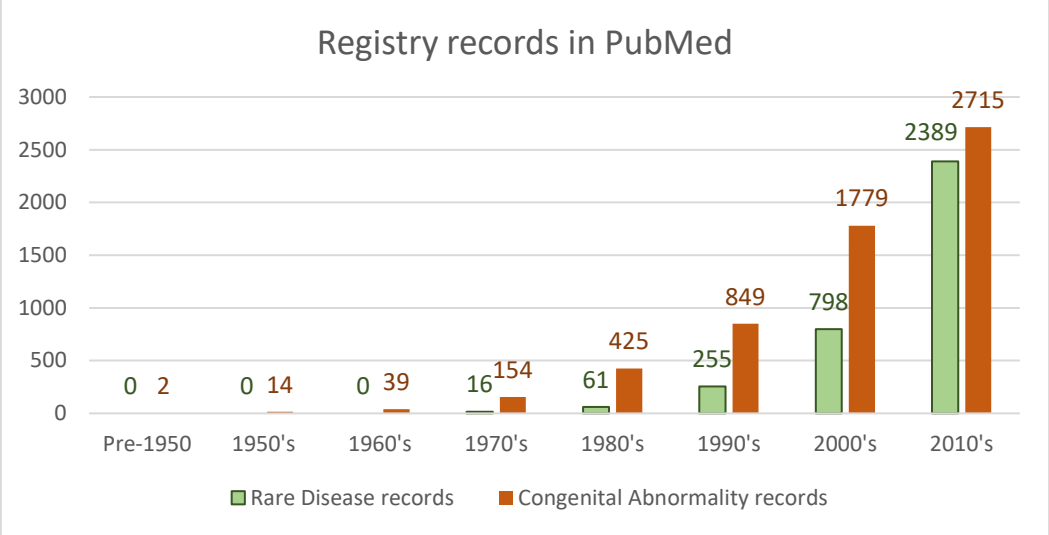
The need for a rare disease coordinator has been a consistent theme, but the defined role of such a person varies. In previous discussions, a substantial proportion of participants defined a 'rare disease coordinator' as a healthcare professional who coordinates complex clinical care needs, or as a personal care coordinator to help individuals with rare diseases manage their health and social care needs. Such a care coordinator would sit clearly within the medical teams, where some current provision exists but would benefit from increased clarity and improved pathways. A non-clinical rare disease coordinator was primarily requested who would be focussed on providing information and support for individuals in their interactions with their medical and social environment.

[11] Department of Health Northern Ireland. Providing high quality care for people affected by rare diseases – the Northern Ireland implementation plan for rare diseases. 2015. https://www.health-ni.gov.uk/sites/default/files/publications/dhssps/ni-rare-diseases-implementation-plan-oct-2015.pdf (accessed 01/01/2018).

[12] Orphanet is a centralised European resource that collates high-quality, validated information on rare diseases to improve the diagnosis, care and treatment of patients with rare diseases. Orphanet also provide specific, stable rare disease nomenclature (ORPHAnumber in ORPHAdata), which is being incorporated to many rare disease registries as an essential component to improve the recording and identification of individual rare diseases in electronic healthcare systems.

Registry focus

Researching rare diseases is not a new concept; the first mention of, 'rare disease' in the US National Library of Medicine National Institutes of Health occurs in 1864[13], while the first mention of 'congenital abnormality' occurs in 1887[14]. Many countries across the world have developed clinical information systems for rare diseases in the form of registries, databases, and spreadsheets; the chart below illustrates the trajectory of such developing resources being published.



Recent technological advances, such as the genomics and digital revolutions, are facilitating the development of effective rare disease registries. Northern Ireland is developing a new initiative to introduce a true digital integrated care record to every person born and / or living in Northern Ireland – encompass. Encompass is envisaged to transform health and social care across Northern Ireland, standardising data that is currently held in lots of different systems that do not easily communicate with each other. Encompass will manage electronic care records including medical notes across specialties, prescribing information, tests, referrals, and appointment bookings. Encompass will minimise duplication and facilitate improved care for patients with rare diseases as their medical records will be centrally stored and be readily accessible to a range of clinical care providers. Multiple patient portals derived from the NI electronic care record are currently being piloted with service user groups within Northern Ireland; for example, My Care Record (Orion Health) and the patient portal integrated to Encompass (EPIC). Other patient portals are accessible to service users in Northern Ireland; for example PatientView and patientMpower. In the longer term, NI may consider the feasibility of a patient portal / linking to existing patient portals and web applications to enhance the NI rare disease registry, for example by enabling patients to self-register, subject to validation and information governance issues, but this would be time consuming, expensive, and outside the current scope. The environment is now optimal to develop a rare disease registry for Northern Ireland; early meetings with the encompass team are essential to efficiently progress this component of the NI rare disease implementation plan.

[13] Hutchinson J. Clinical Lecture on Rare Forms of Disease Consequent on Constitutional Syphilis. *Br Med J* 1864; **1**(173): 440-2.
[14] Lane WA. The Causation of Several Variations and Congenital Abnormalities in the Human Skeleton. *J Anat Physiol* 1887; **21**(Pt 4): 586-610.

Northern Ireland does not presently have a consolidated rare disease research infrastructure and does not have a national rare disease registry. There have been consistent calls for a 'rare disease registry' in Northern Ireland, but the distinction between routine electronic healthcare records, patient self-reported registries, disease-focused registries held by charities, databases and spreadsheets held by clinical teams or individual researchers has not been clearly defined. For convenience and simplicity today, the term 'registry' is used to mean any version of a clinical information resource that contains demographic and phenotypic information on participants. There is scope for synergies with each of these different data resources, given appropriate ethics, governance and data security mechanisms in place. The aim of a Northern Ireland Rare Disease Registry is to provide improved identification and monitoring of rare diseases leading to smarter care for individuals living with a rare disease(s). Key tasks include:

- ✓ Enabling epidemiology and monitoring of incidence, prevalence, nature and cause of rare diseases
- ✓ Improving diagnosis and information on the natural history of disease including clinical outcomes and survival rates.
- ✓ Improving patient care and treatment options
- ✓ Empowering patients and their carers by enabling provision of information relevant to their rare condition.
- ✓ Identifying individuals eligible for clinical trials and supporting participant recruitment for clinical research under an appropriate consent model
- ✓ Providing a resource to monitor, evaluate, and audit services, including outcomes from screening programs and medication provided under managed access schemes.
- ✓ Enabling the creation of a network of professional collaboration for data collection and interpretation
- ✓ Supporting high quality research as a resource with a core minimum dataset and harmonised quality standards.
- ✓ Informing health and social care service planning and commissioning.
- ✓ Scope to connect with local, national, and international rare disease registries

Key indicators for outputs and benefits include:

- Accurate incidence and prevalence data linked to mortality data. Standardising terminology and coding systems is essential.
- Population health monitoring and informing primary prevention alongside congenital anomalies
- Evaluating performance and impact of antenatal screening and non-invasive prenatal testing
- Resource planning for health and social care services
- Resource planning for rare disease patients and their families
- Prioritising service development and information needs
- Supporting research – epidemiology studies, outcome data, facilitating more individuals living with a rare disease(s) to participate in clinical trials, new treatment options, improved information and understanding of rare diseases

Recommendations

*** Consistently top-ranked as priorities for NI

A: Develop a national registration service for rare diseases (Rank 1)
B: Dedicated online NI Rare Disease Information Hub (Rank 2)
C: Lead Rare Disease Information Coordinator (Rank 3)

A: NIRADCAR (Rank 1)

Summary

Recommendation	Key points within the recommendation
(A) Develop a national registration service for rare diseases	**NIRADCAR:** Northern Ireland RAre Diseases & Congenital Abnormalities Registry name proposed Incorporate results from the Newborn Bloodspot Screening Program Formal 'launch' of a rare disease registry *plan* within the next 12 months. Correct coding and accurate data input is essential A working group created with the ENCOMPASS team Aligned with coding systems such as Orphanet, ICD and SNOWMED CT A collaborative steering committee with representation from universities, clinical and public health professionals, patient representatives, existing national registry leads, industry representatives, and encompass representatives should be created within 6 months. Registry must align with, and link to, the developing NI patient and service user information infrastructure. Annual Report A multi-source approach should be employed including secure access to routine HSC data feeds The Registry must be capable of sharing information Longer-term, consideration should be given to complementary direct patient input to research registries Taking advantage of planned computational developments across Northern Ireland

Full background

Rank 1 - Recommendation A: NI should support the unanimous request from patients, as identified from public consultation, voluntary groups and healthcare providers, to develop a national registration service for rare diseases. A centralised NI rare disease resource (**NIRADCAR:** Northern Ireland RAre Diseases & Congenital Abnormalities Registry name proposed) should strive to collect high quality data on every child identified with a congenital anomaly and every person living with a rare disease. This NI rare disease registry should incorporate results from the Newborn Bloodspot Screening Program. There should be a formal 'launch' of a rare disease registry *plan* within the next 12 months.

Correct coding and accurate data input is essential for a successful registry. Rare disease diagnoses should be aligned to Orphanet codes and a working group created with the ENCOMPASS team and the registry lead to discuss how this can be integrated with the digital integrated care records within Encompass[15], aligned with other coding systems such as ICD and SNOWMED CT.

There is a need for robust mechanisms to be developed to ensure developmental milestones are established and met with relevant stakeholder involvement initiated and maintained through an inclusive steering committee. A **collaborative steering committee with representation from universities, clinical and public health professionals, patient representatives, existing national registry leads, industry representatives, and encompass representatives should be created within 6 months.** The registry must align with, and link to, the developing NI patient and service user information infrastructure. Patient representatives are essential members of the registry stakeholder team. An annual report should be generated reporting outcomes and progress with the registry.

A multi-source approach should be employed including secure access to routine HSC data feeds with a model developed for accurate case ascertainment. There are multiple diverse information sources that capture local information on rare diseases, but at present these do not have an adequate coding system recording relevant information for a comprehensive congenital abnormality and rare disease registry. The NI rare disease registry should be developed to derive information primarily from encompass, with a 'yes/no' field included in the patient record asking if each individual has a rare disease. An open source framework should be used so that data is not limited to a specific vendor and can be extensively reused.

The Registry must be capable of sharing information (subject to information governance requirements) with Registries elsewhere in the UK, in the Republic of Ireland, and internationally. Data sharing is critical for rare disease progress. It is essential that a core information dataset is recorded for rare disease patients in NI and harmonised to national / international registries to facilitate anonymised data sharing subject to local governance approvals. A data access committee should be established to consider requests for sharing identifiable data – sharing identifiable data for research purposes should be based on a fully consented opt-in model in order to comply with NI's current legislative requirements. Clarity is required for implementation of the HSC control of data processing legislation in terms of a fully consented / opt-out model for patient information.

Longer-term, consideration should be given to complementary direct patient input to research registries with possibility of case confirmation by interested clinicians and potential for long-term data collection, support forums and groups, and information resource linked to a NI rare disease information hub. A working group to evaluate this option should include utility, validation, and information governance issues.

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

Taking advantage of planned computational developments across Northern Ireland a reliable, flexible 'Clinical Trial Ready' Rare Disease Registry that provides accurate disease data would enable complementary innovative analytics to be developed that could be embedded into routine care and used to inform clinical decision-making tools and health services planning, thus positioning the NI rare disease registry as a 'go-to' local resource for rare disease research, pending regulatory approvals.

B: Online information hub (Rank 2)

Summary

Recommendation	Key points within the recommendation
(B) Dedicated online NI Rare Disease Information Hub	Provides access (hosted, or signposted where possible) to reliable rare disease specific and generic clinical and non-clinical information. Should link with ongoing programmes such as NI Direct, Digital Innovation Hubs through HDRUK, The UK Clinical Trials Gateway through NIHR, and Orphanet. Disease specific information for individuals living and working with rare diseases; signposting to expert standards of care guidelines Access to referral resources e.g. physio, genetics, counselling, treatment options, and emergency telephone numbers (e.g. to resolve wheelchair, hoist, malfunctions) A directory of accredited educational resources for rare diseases A diary of events, conferences, workshops and experts invited to NI Access to work, educational support, and benefit related information A list of local and national experts and charities / support groups for many rare diseases Map of key services e.g. accessible hydrotherapy and accessible leisure facilities List of research and a map of local rare disease research projects / developments Regular email updates / newsletter with updates from the Hub Discussion forum / network facilitating improved communication and the development of strong inter-professional networks (MDTs, ERNs, Project ECHO, etc.) Online training modules; generic and specific Specialised rare disease training for professionals

Full background

Rank 2 - Recommendation B: To develop a **dedicated online NI Rare Disease Information Hub** that provides access (hosted, or signposted where possible) to reliable rare disease specific and generic clinical and non-clinical information. This resource should link with ongoing programmes such as NI Direct, Digital Innovation Hubs through HDRUK, The UK Clinical Trials Gateway through NIHR, and Orphanet.

It is recognised that this will be an evolving process, but the following components were repeatedly requested within an online Rare Disease Hub:

1) Disease specific information for individuals living and working with rare diseases, including signposting to expert standards of care guidelines
2) Access to referral resources e.g. physio, genetics, counselling, treatment options, and emergency telephone numbers (e.g. to resolve wheelchair, hoist, malfunctions)
3) A directory of accredited educational resources for rare diseases
4) A diary of events, conferences, workshops and experts invited to NI
5) Access to work, educational support, and benefit related information
6) A list of local and national experts and charities / support groups for many rare diseases
7) Map of key services e.g. accessible hydrotherapy and accessible leisure facilities
8) List of research and a map of local rare disease research projects / developments
9) Regular email updates / newsletter with updates from the Hub
10) Discussion forum / network facilitating improved communication and the development of strong inter-professional networks (MDTs, ERNs, Project ECHO, etc.)
11) Online training modules; generic and specific
12) Specialised rare disease training for professionals

C: Lead rare disease information coordinator (Rank 3)

Summary

Recommendation	Key points within the recommendation
(c) Lead Rare Disease Information Coordinator	A person who maintains a list of information and advises on accessible health and social care resources. Non-medical role Sign-posting and advising Keeping a record of ongoing clinical trials, research opportunities, rare disease events in NI' Advising on content within the online information hub Developing networks and contact points Acting as an Orphanet representative for Northern Ireland.

Full background

Rank 3 – Recommendation C: To appoint a **Lead Rare Disease Information Coordinator** to fulfil the critical role of*, 'A person who maintains a list of information and advises on accessible health and social care resources. This rare disease information coordinator should have a non-medical role directing / sign-posting individuals to appropriate health and social care resources. This includes keeping a record of ongoing clinical trials, research opportunities, rare disease events in NI'.* This is a demanding role with a substantial commitment that should include advising on content within the online information hub (ranked priority recommendation 2), developing networks and contact points within each 'system' to efficiently advise patients, carers, professional colleagues, and importantly acting as an Orphanet representative for Northern Ireland.

Recommendations already achieved, but requiring ongoing commitment

Education and training focus

D: NI participate in national RD panels
E: Dedicated RD teaching in curricula
F: Staff supported to attend RD training
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Summary

Recommendation	Progress to date
(D) NI participate in national RD panels *(NI should maintain updated knowledge to ensure NI training is aligned to that in GB. HEE is working with a range of stakeholders to ensure genomics is embedded in training curricula and revalidation requirements to ensure sufficient genomics understanding by all staff involved in clinical decisions)*	NI representatives already participate on relevant national panels supporting genomic education such as the UK Faculty of Genomic Medicine, NHS Genetics Training Leads, and MSc Genetics Training Managers Networks as well as ongoing clinical and biomedical accredited training courses.
(E) Dedicated rare disease teaching in curricula *(Should continue to be incorporated to undergraduate medical, nursing, and biomedical university curricula, including providing signposting to useful rare disease resources such as Orphanet. Teaching methods and clinical techniques for genomic medicine should be formally incorporated to taught curricula. Patients should be supported to be involved as educators in the development and delivery of rare disease training programs. Patients are ideally placed to communicate their lived experience as a fundamental message to health and social care professionals at undergraduate and postgraduate levels)*	NI has several rare disease teaching sessions embedded into the existing undergraduate medical school and a single session into the biomedical curricula at QUB; these include students hearing directly 'the patient voice'. Opportunities are being scoped to include further rare disease teaching into the C25 curriculum being developed for medical students at QUB. 'Patient voices' videos are being developed where rare disease patients may not be able to attend scheduled teaching sessions due to the nature of their condition. Further videos are required. Individuals living with rare diseases have been supported to attend external training sessions such as the European Patient' Academy on Therapeutic Innovation (EUPATI) program. Work is underway as part of the Transformation Programme to establish a Patient and Public Involvement infrastructure, including the development of a capacity building framework for participants, ongoing management and support arrangements. Collaborative partnerships between NIRDP, PCC, PHA and local Universities (QUB, UU) are proving

	productive in this area, especially the capacity building and training elements, drawing on the EURORDIS/EUPATI experience in rare diseases. **NOTE:** Should explore how this can be embedded in the UU medical and nursing curriculum.
(F) Staff supported to attend RD training *(Due to the number of individuals needing training locally consideration should be given to developing an accredited MSc compatible with the UK scientist training program (STP). HSC staff should continue to be supported to undertake clinically relevant rare disease PhD research, for example through HSC or ICAT schemes)*	Individuals from NI have been supported to attend external training locally (by attending MSc components at Queen's University of Belfast) and in GB (e.g. by attending MSc components at University of Manchester) as part of upscaling the workforce planning through the NI genomic medicine centre. Clinical bioinformaticists employed in the Regional Genetics Centre were both trained at Queen's University of Belfast. Drs Donnelly and McKnight have been scoping potential for a dedicated MSc / postgraduate certificate hosted by QUB to support training of registrars, genetic counsellors, clinical scientist trainees, and other students. Also, Genomics workshops for healthcare professionals have been held as part of the 100,000 genomes project. Existing staff have received specialist training, with two additional trainee Clinical Scientists appointed to support next generation sequencing in Northern Ireland. Two bioinformaticians were also appointed as part of the local 100,000 genomes project team and are now permanently funded as part of the NI next generation sequencing service development. These individuals are also contributing to core teaching of undergraduate and postgraduate medical and biomedical students within Queen's University Belfast. As a result of the initial workshops with healthcare professionals considering participating in the 100,000 genomes project, a non-genetics-specialist training presentation was developed with an explicit pathway for recruitment and consent.
(G) CPD accreditation promoted *(Should be sought for rare disease conferences and training days, which should be circulated on Trust and University websites with individuals*	Many training sessions and conferences held locally have secured CPD accreditation for attendees.

supported to attend where possible. Dedicated training events for specific rare diseases / rare disease groups with input from international experts were welcomed – but online training is preferred. Dedicated rare disease training for local professionals, using a mixed approach of interactive sessions, seminars, training courses, printed material, and online training modules. Ideally such events would be posted on a centralised rare disease 'events' website (could be hosted within ranked priority recommendation 2; online information hub).	

Full background

D: NI participate in national RD panels

Recommendation D: NI should maintain updated knowledge to ensure NI training is aligned to that in GB. HEE is working with a range of stakeholders to ensure genomics is embedded in training curricula and revalidation requirements to ensure sufficient genomics understanding by all staff involved in clinical decisions; **NI representatives should be supported to participate on relevant national education and training panels.**

1) NI representatives already participate on relevant national panels supporting genomic education such as the UK Faculty of Genomic Medicine, NHS Genetics Training Leads, and MSc Genetics Training Managers Networks as well as ongoing clinical and biomedical accredited training courses.

E: Dedicated RD teaching in curricula

Recommendation E: Dedicated rare disease teaching sessions should continue to be incorporated to undergraduate medical, nursing, and biomedical university curricula, including providing signposting to useful rare disease resources such as Orphanet. Teaching methods and clinical techniques for genomic medicine should be formally incorporated to taught curricula. Patients should be supported to be involved as educators in the development and delivery of rare disease training programs. Patients are ideally placed to communicate their lived experience as a fundamental message to health and social care professionals at undergraduate and postgraduate levels.

1) NI has several rare disease teaching sessions embedded into the existing undergraduate medical school and a single session into the biomedical curricula at QUB; these include students hearing directly 'the patient voice'. Opportunities are being scoped to include further rare disease teaching into the C25 curriculum being developed for medical students at QUB.

2) 'Patient voices' videos are being developed where rare disease patients may not be able to attend scheduled teaching sessions due to the nature of their condition. Further videos are required.

3) Individuals living with rare diseases have been supported to attend external training sessions such as the European Patient' Academy on Therapeutic Innovation (EUPATI) program. Work is underway as part of the Transformation Programme to establish a Patient and Public Involvement infrastructure, including the development of a capacity building framework for participants, ongoing management and support arrangements. Collaborative partnerships between NIRDP, PCC, PHA and local Universities (QUB, UU) are proving productive in this area, especially the

capacity building and training elements, drawing on the EURORDIS/EUPATI experience in rare diseases.

4) Should explore how this can be embedded in the UU medical and nursing curriculum.

F: Staff supported to attend RD training

Recommendation F: Consideration should be given to supporting individuals from the existing workforce in NI to **attend taught/research postgraduate training courses in genomic medicine**. Due to the number of individuals needing training locally consideration should be given to developing an accredited MSc compatible with the UK scientist training program (STP). HSC staff should continue to be supported to undertake clinically relevant rare disease PhD research, for example through HSC or ICAT schemes.

1) Individuals from NI have been supported to attend external training locally (by attending MSc components at Queen's University of Belfast) and in GB (e.g. by attending MSc components at University of Manchester) as part of upscaling the workforce planning through the NI genomic medicine centre. Clinical bioinformaticists employed in the Regional Genetics Centre were both trained at Queen's University of Belfast. Drs Donnelly and McKnight have been scoping potential for a dedicated MSc / postgraduate certificate hosted by QUB to support training of registrars, genetic counsellors, clinical scientist trainees, and other students. Also, Genomics workshops for healthcare professionals have been held as part of the 100,000 genomes project.

2) Existing staff have received specialist training, with two additional trainee Clinical Scientists appointed to support next generation sequencing in Northern Ireland. Two bioinformaticians were also appointed as part of the local 100,000 genomes project team and are now permanently funded as part of the NI next generation sequencing service development. These individuals are also contributing to core teaching of undergraduate and postgraduate medical and biomedical students within Queen's University Belfast.

3) As a result of the initial workshops with healthcare professionals considering participating in the 100,000 genomes project, a non-genetics-specialist training presentation was developed with an explicit pathway for recruitment and consent.

G: CPD accreditation promoted

Recommendation G: CPD accreditation should be sought for rare disease conferences and training days, which should be circulated on Trust and University websites with individuals supported to attend where possible. Dedicated training events for specific rare diseases / rare disease groups with input from international experts were welcomed – but online training is preferred. Dedicated rare disease training for local professionals, using a mixed approach of interactive sessions, seminars, training courses, printed material, and online training modules. Ideally such events would be posted on a centralised rare disease 'events' website *(could be hosted within ranked priority recommendation 2; online information hub).*

1) Many training sessions and conferences held locally have secured CPD accreditation for attendees.

Registry focus

H: Extend newborn screening
I: Rare cancer factsheet

Summary

Recommendation	Progress to date
(H) Extend newborn screening *(To encompass more conditions where early intervention can significantly improve patient outcomes. Newborn screening approaches, such as the standard heel-prick test, identify babies at risk of developing rare conditions, often facilitating diagnosis before symptoms develop. This is particularly important for rare conditions where early treatment may prevent the onset or minimise irreversible decline for patients. Across the UK, nine conditions are screened for in newborns[16] in England, Scotland and Wales (sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT), phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1), homocystinuria (HCU)), with only five screened in Northern Ireland (PKU, CHT, CF, MCADD, and SCD), which is behind the best-practice in Europe. Recent calls have been made to widen the Irish Newborn Screening Program from the present 8 to the EU standard where screening occurs for more than 20 conditions routinely)*	From 1st March 2020, babies in NI will be offered newborn screening for PKU, CHT, CF, MCADD, SCD + MSUD, IVA, GA1, HCU in line with the rest of the UK.
(I) Rare cancer factsheet *(Summarising the incidence and prevalence of rare cancers in Northern Ireland)*	Scoping work is underway with researchers within the Centre for Public Health (QUB) and NICR staff to produce an initial review of rare cancers. Development of an infographic and rare disease factsheet is planned. **NOTE:** Ongoing support is required by the NICR team to routinely deliver annual reports of rare disease statistics within Northern Ireland

[16] https://www.gov.uk/guidance/newborn-blood-spot-screening-programme-overview#conditions-screened-for

Full background

H: Extend newborn screening

Recommendation H: That consideration be given to expanding the newborn blood spot screening in Northern Ireland to encompass more conditions where early intervention can significantly improve patient outcomes. Newborn screening approaches, such as the standard heel-prick test, identify babies at risk of developing rare conditions, often facilitating diagnosis before symptoms develop. This is particularly important for rare conditions where early treatment may prevent the onset or minimise irreversible decline for patients. Across the UK, nine conditions are screened for in newborns[17] in England, Scotland and Wales (sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT), phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1), homocystinuria (HCU)), with only five screened in Northern Ireland (PKU, CHT, CF, MCADD, and SCD), which is behind the best-practice in Europe. Recent calls have been made to widen the Irish Newborn Screening Program from the present 8 to the EU standard where screening occurs for more than 20 conditions routinely.

I: Rare cancer factsheet

Recommendation I: That an **annual factsheet should be produced by the NI cancer registry** summarising the incidence and prevalence of rare cancers in Northern Ireland.

1) Scoping work is underway with researchers within the Centre for Public Health (QUB) and NICR staff to produce an initial review of rare cancers. Development of an infographic and rare disease factsheet is planned.

2) Ongoing support is required by the NICR team to routinely deliver annual reports of rare disease statistics within Northern Ireland

[17] https://www.gov.uk/guidance/newborn-blood-spot-screening-programme-overview#conditions-screened-for

Short-term priorities (within 12 months to end of August 2021)

Education and training focus

J: CPD working group
K: RD online training module

Summary

Recommendation	Comments
(J) Continuous Professional Development (CDP) working group *(That meets quarterly with input from the HSC education and training team, training and education leads from the NI genomic medicine centre, relevant Royal Colleges, the Northern Ireland Medical and Dental Training Agency, Queen's University Belfast and Ulster University leads delivering genomic medicine, to conduct annual training needs 'update' analysis for rare diseases and identify synergies with potential to adapt existing training material in this rapidly evolving field. Patients with a rare disease and their carers should be involved in the development of such training programs)*	
(K) Rare disease online training module *(More information is required for healthcare professionals that is easily accessible – GPs in particular emphasised the need for more information to better support their patients who have rare diseases. A rare disease online training module for rare disease in NI should be developed to highlight rare disease facts in Northern Ireland and help raise awareness of relevant rare disease resources (such as Orphanet) for healthcare professionals. Improved signposting for healthcare professionals is required from pre-diagnosis to the development of a care plan for rare diseases; for example, many doctors were unaware of the process in NI to consider orphan drugs or extra contractual referrals for their rare disease patients so links to these resources could be included in the training module)*	Working with hospital consultants and local GPs, a "quick reference guide for rare disease" has been published as a 'red flag' toolkit to help GPs better identify and manage patients with a rare disease[18]. **NOTE:** Should explore developing an online generic rare disease training module focused on NI resources as a priority.

[18] A quick reference guide for rare disease: supporting rare disease management in general practice. Ashleen Crowe, Helen McAneney, Patrick J Morrison, Margaret E Cupples and Amy Jayne McKnight. British Journal of General Practice 2020; 70 (694): 260-261.

Full background

J: CPD working group

Recommendation J: To establish **a three-year continuing professional development working group** that meets quarterly with input from the HSC education and training team, training and education leads from the NI genomic medicine centre, relevant Royal Colleges, the Northern Ireland Medical and Dental Training Agency, Queen's University Belfast and Ulster University leads delivering genomic medicine, to conduct annual training needs 'update' analysis for rare diseases and identify synergies with potential to adapt existing training material in this rapidly evolving field. Patients with a rare disease and their carers should be involved in the development of such training programs.

K: RD online training module

Recommendation K: More information is required for healthcare professionals that is easily accessible – GPs in particular emphasised the need for more information to better support their patients who have rare diseases. **A rare disease online training module for rare disease in NI** should be developed to highlight rare disease facts in Northern Ireland and help raise awareness of relevant rare disease resources (such as Orphanet) for healthcare professionals. Improved signposting for healthcare professionals is required from pre-diagnosis to the development of a care plan for rare diseases; for example, many doctors were unaware of the process in NI to consider orphan drugs or extra contractual referrals for their rare disease patients so links to these resources could be included in the training module.

5) Working with hospital consultants and local GPs, a "quick reference guide for rare disease" has been published as a 'red flag' toolkit to help GPs better identify and manage patients with a rare disease[19].

6) Should explore developing an online generic rare disease training module as a priority.

Registry focus

L: RD basic statistics
M: Support existing registries
N: RD exemplar registry projects supported

Summary

Recommendation	Comments
(L) Generate RD basic statistics *(Initial prevalence data should be derived via HSC Business Services Organisation and the regional genetics clinical information system using ICD codes and free text for the 150 rare diseases that are believed to contribute the largest population burden to rare diseases. Local participants contributing to national programs such as the 100,000 genomes project already have data cleaned and verified for inclusion in a NI rare disease registry)*	

[19] A quick reference guide for rare disease: supporting rare disease management in general practice. Ashleen Crowe, Helen McAneney, Patrick J Morrison, Margaret E Cupples and Amy Jayne McKnight. British Journal of General Practice 2020; 70 (694): 260-261.

(M) Support existing registries *(To continue until a standardised infrastructure is in place, which would enable them to be future-proofed and integrated with national IT systems using a unique personal identifier (such as the Health and Care Number) to enhance interoperability and integration. Such registries should be supported to incorporate Orphacode nomenclature and EU data standards to align local rare disease registration with international standards and facilitate engagement with international registries and clinical trials. A Registry infrastructure should be developed to provide administrative support, and technical help for more detailed disease-focused databases, registers, resources under an inclusive 'rare disease registry and bioresource' ethics and governance approval)*	
(N) RD exemplar registry projects supported *(NI rare disease registry researchers should continue to work with NCARDRS (England), CARIS (Wales), CADRISS (Scotland), Orphanet, RD-CONNECT, and the Irish National Rare Disease Office to ensure a NI rare disease and congenital anomaly register is aligned, where possible, to national and international standards and outcomes. NI should work collaboratively with ongoing UK-wide initiatives within rare disease registries for exemplar projects)*	

Full background

L: Generate RD basic statistics

Recommendation L: Initial prevalence data should be derived via HSC Business Services Organisation and the regional genetics clinical information system using ICD codes and free text for the 150 rare diseases that are believed to contribute the largest population burden to rare diseases. Local participants contributing to national programs such as the 100,000 genomes project already have data cleaned and verified for inclusion in a NI rare disease registry.

M: Support existing registries

Recommendation M: That **existing and developing rare disease specific registries should be supported** to continue until a standardised infrastructure is in place, which would enable them to be future-proofed and integrated with national IT systems using a unique personal identifier (such as the Health and Care Number[20]) to enhance interoperability and integration. Such registries should be supported to incorporate Orphacode nomenclature[21] and EU data standards to align local rare disease registration with international standards and facilitate engagement with international registries and clinical trials. A Registry infrastructure should be developed to provide administrative support, and

[20] http://www.hscbusiness.hscni.net/services/2748.htm

[21] http://www.orphadata.org/cgi-bin/index.php

technical help for more detailed disease-focused databases, registers, resources under an inclusive 'rare disease registry and bioresource' ethics and governance approval.

N: RD exemplar registry projects supported

Recommendation N: NI rare disease registry researchers should continue to work with NCARDRS (England), CARIS (Wales), CADRISS (Scotland), Orphanet, RD-CONNECT, and the Irish National Rare Disease Office to ensure a NI rare disease and congenital anomaly register is aligned, where possible, to national and international standards and outcomes. NI should work collaboratively with ongoing UK-wide initiatives within **rare disease registries for exemplar projects**.

Information and communication focus

O: Improve linkages & include RD question in national surveys
P: Host annual RD conference

Summary

Recommendation	Comments
(O) Improve linkages & include RD question in national surveys *(including the 10,000 Voices project. A question on rare disease should be incorporated to the minimum dataset for demography in national surveys to facilitate sub-group analyses, e.g. "Do you have a rare disease or Syndrome Without A Name (SWAN)")*	
(P) Host annual RD conference *(To share best practice, update on rare disease progress, introduce new initiatives, and provide networking opportunities)*	

Full background

O: Improve linkages & include RD question in national surveys

Recommendation O: To improve linkages with ongoing projects such as national surveys and the 10,000 Voices team. A question on rare disease should be incorporated to the minimum dataset for demography in national surveys to facilitate sub-group analyses, e.g. "Do you have a rare disease or Syndrome Without A Name (SWAN)".

P: Host annual RD conference

Recommendation P: To host an annual rare disease conference to share best practice, update on rare disease progress, introduce new initiatives, and provide networking opportunities.

Medium-term priorities (within 1-4 years)

Education and training focus

Q: Develop non-specialist 'omics' training tools
R: Promote RD placements & supported clinical time
S: Improved coordination of care
T: Appoint a training and education coordinator

Summary

Recommendation	Comments
(Q) Develop non-specialist 'omics' training tools *(Further training of mainstream (non-genetic clinical specialists) would enhance the efficiency of the 'genomic medicine' process in terms of enhancing the information returned from Genomics England and conveyed to recruiting clinicians, initial clinician review of variants, and in-depth analysis of variants. A non-specialist 'omics' training tool should be developed highlighting best practice for interpreting and returning results from WGS locally as part of mainstreaming genomic medicine)*	
(**R) Promote RD placements & supported clinical time** *(Rare disease 'Placement' opportunities and protected clinical time should be available to support medical, nursing and associated healthcare professionals to gain practical experience in specialist clinics)*	
(S) Improved coordination of care *(Would minimise duplication of effort and ensure appropriate expertise is available to provide improved care – more rare disease training where healthcare professionals listen to patients and are aware of multidisciplinary treatments would improve outcomes)*	
(T) Appoint a training and education coordinator *(That a training and education coordinator be appointed for rare diseases to act as a primary point of contact for:* *a) Signposting and connecting individuals living and working with rare diseases seeking to undertake formal / informal training.* *b) Maintaining an updated list of training opportunities and liaising with speciality leads to identify and coordinate ongoing training and education needs*	

c) *Supporting charities organising education and training events in NI in terms of appropriate accreditation, signposting, and disseminating information.* d) *Connecting Royal Colleges for accredited training courses and promoting CPD opportunities for rare disease to relevant health and social care staff.* e) *Working with Royal Colleges and local health and social care professionals to develop local training resources to meet education and training needs.* f) *Working with the Centres for Medical Education, Biomedical Sciences, Public Health, and Nursing at Queen's University Belfast, alongside the School of Nursing and planned medical school at Ulster University to develop new teaching tools and further embed rare disease teaching across Northern Ireland.* g) *Working with the Encompass teams to develop an online rare disease training tool to support appropriately coded entry of data and rare disease information retrieval.* h) *Develop a rare disease online training tool for GPs and other healthcare professionals – adapted from that existing elsewhere to support people working in NI's health and social care system and legislative environment.* i) *Developing and / or maintaining an online training tool / website links for accessing orphan drugs and extra contractual referrals with a clear pathway to access described* j) *Developing an online genomic medicine training tool – MSc / PGCert*	

Full background

Q: Develop non-specialist 'omics' training tools

Recommendation Q: Further training of mainstream (non-genetic clinical specialists) would enhance the efficiency of the 'genomic medicine' process in terms of enhancing the information returned from Genomics England and conveyed to recruiting clinicians, initial clinician review of variants, and in-depth analysis of variants. **A non-specialist 'omics' training tool should be developed** highlighting best practice for interpreting and returning results from WGS locally as part of mainstreaming genomic medicine.

R: Promote RD placements & supported clinical time

Recommendation R: Rare disease 'Placement' opportunities and protected clinical time should be available to support medical, nursing and associated healthcare professionals to gain practical experience in specialist clinics.

S: Improved coordination of care

Recommendation S: Improved coordination of care would minimise duplication of effort and ensure appropriate expertise is available to provide improved care – more rare disease training where healthcare professionals listen to patients and are aware of multidisciplinary treatments would improve outcomes.

T: Appoint a training and education coordinator

Recommendation T: That a **training and education coordinator** be appointed for rare diseases to act as a primary point of contact for:

a) Signposting and connecting individuals living and working with rare diseases seeking to undertake formal / informal training.
b) Maintaining an updated list of training opportunities and liaising with speciality leads to identify and coordinate ongoing training and education needs
c) Supporting charities organising education and training events in NI in terms of appropriate accreditation, signposting, and disseminating information.
d) Connecting Royal Colleges for accredited training courses and promoting CPD opportunities for rare disease to relevant health and social care staff.
e) Working with Royal Colleges and local health and social care professionals to develop local training resources to meet education and training needs.
f) Working with the Centres for Medical Education, Biomedical Sciences, Public Health, and Nursing at Queen's University Belfast, alongside the School of Nursing and planned medical school at Ulster University to develop new teaching tools and further embed rare disease teaching across Northern Ireland.
g) Working with the Encompass teams to develop an online rare disease training tool to support appropriately coded entry of data and rare disease information retrieval.
h) Develop a rare disease online training tool for GPs and other healthcare professionals – adapted from that existing elsewhere to support people working in NI's health and social care system and legislative environment.
i) Developing and / or maintaining an online training tool / website links for accessing orphan drugs and extra contractual referrals with a clear pathway to access described
j) Developing an online genomic medicine training tool – MSc / PGCert

Registry focus

U: Appoint a registry lead and information support officers

Summary

Recommendation	Comments
(U) Appoint a registry and information support officers *(Should be employed to work with a register stakeholder team that meets quarterly including clinical specialists for validation of diagnoses)*	

Full background

U: appoint a registry lead and information support officers

Recommendation: U registry lead and at least two information support officers should be employed to work with a register stakeholder team that meets quarterly including clinical specialists for validation of diagnoses.

Information and communication focus

V: Develop patient portal
W: Enhance mental wellbeing; counselling support
X: Accrediting information
Y: Raise awareness of RD across communities
Z: Facilitate peer support and advocacy

Summary

Recommendation	Comments
(V) Develop patient portal *(To provide patients, and primary carers if necessary, with access to their electronic healthcare record through a patient portal embedded in ENCOMPASS. To facilitate patient access to their medical records to help them coordinate their care between medical specialties and / or across different locations)*	
(W) Enhance mental wellbeing; counselling support *(To provide resilience training and enhance the mental wellbeing of patients and carers through access to relevant services. Links to existing services should be signposted from the NI Information Hub. Counselling support should routinely be made available to individuals (patients, carers, other family members) struggling with the impact of a rare disease)*	
(X) Accrediting information *(To consider 'accrediting' alert cards, flyers, and / or patient passports that patients could carry with information about their condition in case of emergency. Many such cards and passport style documents exist, but without clinical 'accreditation' or an 'NHS stamp', they are often dismissed)*	
(Y) Raise awareness of RD across communities *(As to the complex nature of rare diseases, which often necessitates multiple hospital appointments at short notice. Flexible working for individuals with disabilities and those with significant caring responsibilities should be promoted throughout the workforce)*	
(Z) Facilitate peer support and advocacy *(To facilitate peer-support and training of people living with rare diseases to act as peer support, peer mentors, and advocate in relevant health and community panels)*	

Full background

V: Develop patient portal

Recommendation V: To provide patients, and primary carers if necessary, with access to their electronic healthcare record through a **patient portal** embedded in ENCOMPASS. To facilitate patient access to their medical records to help them coordinate their care between medical specialties and / or across different locations.

W: Enhance mental wellbeing; counselling support

Recommendation W: To provide resilience training and **enhance the mental wellbeing** of patients and carers through access to relevant services. Links to existing services should be signposted from the NI Information Hub. Counselling support should routinely be made available to individuals (patients, carers, other family members) struggling with the impact of a rare disease.

X: Accrediting information

Recommendation X: To consider 'accrediting' alert cards, flyers, and / or patient passports that patients could carry with information about their condition in case of emergency. Many such cards and passport style documents exist, but without clinical 'accreditation' or an 'NHS stamp', they are often dismissed.

Y: Raise awareness of RD across communities

Recommendation Y: To raise awareness with schools and employers as to the complex nature of rare diseases, which often necessitates multiple hospital appointments at short notice. Flexible working for individuals with disabilities and those with significant caring responsibilities should be promoted throughout the workforce.

Z: Facilitate peer support and advocacy

Recommendation Z: To facilitate peer-support and training of people living with rare diseases to act as peer support, peer mentors, and advocate in relevant health and community panels.

Longer-term priorities (4+ years)

Multiple foci

AA: Develop an expert centre for RD

Summary

Recommendation	Comments
(**AA) Develop an expert centre for RD** *(This should include:* • *Development and maintenance of a network of expert support to help effectively manage many rare diseases. Virtual reference networks enable healthcare professionals to connect to experts on particular rare disease issues and facilitate patients benefiting from expertise at external centres of excellence for rare diseases.* ▪ *development and coordination of a rare disease registry (with room for 'placements' supported by external funding to develop more focused (e.g. disease specific resources) that enhance the rare disease registry infrastructure)* ▪ *a record of clinical nodes of expertise for specific conditions that are recorded on relevant sites such as Orphanet and support NI to participate in European research networks* ▪ *support to facilitate NI participating in more clinical trials* ▪ *a rare disease care coordinator to help develop models of care coordination, maintain an up-to-date listing of clinical expertise (local and centres of excellence)* ▪ *an education and training coordinator to help develop a NI Rare Disease Information Hub with relevant, accurate information and signposting to relevant resources where relevant* ▪ *protected clinical time (and administrative support) to enable clinical-academic research grants and engagement with UK-wide rare disease research initiatives.* ▪ *This would provide the systemic 'scaffolding' for an effective, cohesive, multidisciplinary Rare Disease Network across NI)*	

Full background

AA: Develop an expert centre for RD

Recommendation AA: That a **dedicated rare disease centre in Northern Ireland** should be supported with sustainable funding. This should include:

- Development and maintenance of a network of expert support to help effectively manage many rare diseases. Virtual reference networks enable healthcare professionals to connect to experts on particular rare disease issues and facilitate patients benefiting from expertise at external centres of excellence for rare diseases.
- development and coordination of a rare disease registry (with room for 'placements' supported by external funding to develop more focused (e.g. disease specific resources) that enhance the rare disease registry infrastructure)
- a record of clinical nodes of expertise for specific conditions that are recorded on relevant sites such as Orphanet and support NI to participate in European research networks
- support to facilitate NI participating in more clinical trials
- a rare disease care coordinator to help develop models of care coordination, maintain an up-to-date listing of clinical expertise (local and centres of excellence)
- an education and training coordinator to help develop a NI Rare Disease Information Hub with relevant, accurate information and signposting to relevant resources where relevant
- protected clinical time (and administrative support) to enable clinical-academic research grants and engagement with UK-wide rare disease research initiatives.
- This would provide the systemic 'scaffolding' for an effective, cohesive, multidisciplinary Rare Disease Network across NI.

Information and communication focus

AB: Appoint a team of RD information coordinators
AC: Develop an accessible technology resource
AD: Develop care pathways / models of care
AE: Improve RD clinical trial capacity
AF: Develop Computer Assisted Diagnosis (CAD) for RD

Summary

Recommendation	Comments
(AB) Appoint a team of RD information coordinators *(Should be appointed to facilitate access to information, both in person and through a dedicated telephone help/information line. This Team should also act as Northern Ireland's Orphanet representative, ensuring that local details are accurately maintained. "Contact points" should be established and maintained to facilitate information sharing and care coordination. There is a need to move away from the purely medical model to a more holistic social model, with a "whole systems approach" connecting patients, carers, allied health professionals, medical staff, and essential services such as*	

education, employment, housing and transport. Coordinated by the lead rare disease information coordinator (ranked priority recommendation 3), one part-time person for each regional Hospital Trust would ensure the effective delivery of the direct patient support and liaison as piloted in the Stronger Together project)	
(AC) Develop an accessible technology resource *(To collate details of supportive / assistive technologies available in Northern Ireland and develop a resource where such details are maintained and accessible. At a minimum this should be provided for occupational therapy and educational resources, and ideally link to/from the (ranked priority 2) online Rare Disease Information Hub)*	
(AD) Develop care pathways / models of care *(Leading from pre-diagnosis through diagnosis to condition specific, person-centered care plans, led by the paediatric and adult rare disease clinical leads. Transparent models of care are required for individuals with a rare disease. These must be flexible, and include streamlined processes for accessing multidisciplinary care from experts who may be geographically distant where required)*	
(AE) Improve RD clinical trial capacity *(To improve capacity and infrastructure to enable individuals with rare diseases to participate in research and clinical trials)*	
(AF) Develop Computer Assisted Diagnosis (CAD) for RD *(To consider computer assisted diagnosis of rare diseases within / complementing ENCOMPASS)*	

AB: Appoint a team of RD information coordinators

Recommendation AB: A team of **Northern Ireland Rare Disease Information Coordinators** should be appointed to facilitate access to information, both in person and through a dedicated telephone help/information line. This Team should also act as Northern Ireland's Orphanet representative, ensuring that local details are accurately maintained. "Contact points" should be established and maintained to facilitate information sharing and care coordination. There is a need to move away from the purely medical model to a more holistic social model, with a "whole systems approach" connecting patients, carers, allied health professionals, medical staff, and essential services such as education, employment, housing and transport. Coordinated by the lead rare disease information coordinator (ranked priority recommendation 3), one part-time person for each regional Hospital Trust would ensure the effective delivery of the direct patient support and liaison as piloted in the Stronger Together project.

AC: Develop an accessible technology resource

Recommendation AC: To collate details of **supportive / assistive technologies** available in Northern Ireland and develop a resource where such details are maintained and accessible. At a minimum this should be provided for occupational therapy and educational resources, and ideally link to/from the (ranked priority 2) online Rare Disease Information Hub.

AD: Develop care pathways / models of care

Recommendation AD: To **establish a care pathway / models of care** across the life course leading from pre-diagnosis through diagnosis to condition specific, person-centered care plans, led by the paediatric and adult rare disease clinical leads. Transparent models of care are required for individuals with a rare disease. These must be flexible and include streamlined processes for accessing multidisciplinary care from experts who may be geographically distant where required.

AE: Improve RD clinical trial capacity

Recommendation AE: To improve capacity and infrastructure to enable individuals with rare diseases to participate in research and clinical trials.

AF: Develop CAD for RD

Recommendation AF: To consider computer assisted diagnosis of rare diseases within / complementing ENCOMPASS.

Workshop

Due to the COVID-19 pandemic, this workshop was held remotely using Microsoft Team on 28th May 2020. The meeting was hosted by Dr AJ McKnight, facilitated by Grace Henry, and funded by an ESRC impact acceleration award. Together with hard copy material and electronic copies of files, Microsoft Whiteboard was used to facilitate discussions on the day.

In advance of this meeting everyone received a participant pack electronically and in hard copy where requested. This participant pack contained:

- An introduction to Microsoft Teams (Appendix 1: An introduction to Teams…)
- A participant information pack with lots of information relevant for the day, including original recommendations (Appendix 2: Original recommendations from the three reports).
- A summary of the recommendations to be prioritised in a quick reference workbook style (*document also attached; Confidential_Final_participant_worksheets_AJ_220520.pdf*)
- Individual A6 cards with recommendations (Appendix 3: Selected participant materials, including colour-coordinated A6 cards with recommendations). These are colour coded so:
    - the top three ranked priorities are ivory (peach online)
    - priorities where ongoing work is being delivered are green
    - short-term priorities are yellow
    - medium-term priorities are blue
    - longer-term priorities are purple.
- Blank paper and coloured pens

Participants were requested to advise AJ or Grace of any ongoing activities that were not mentioned in these documents so they could be included in the work going forwards. No updates were received.

Top-ranked priorities based on survey of participants prior to discussion.

Several recommendations from the January reports are already ongoing as noted below. Also agreed were the consistently top-ranked priorities for action in Northern Ireland (Figure 1).

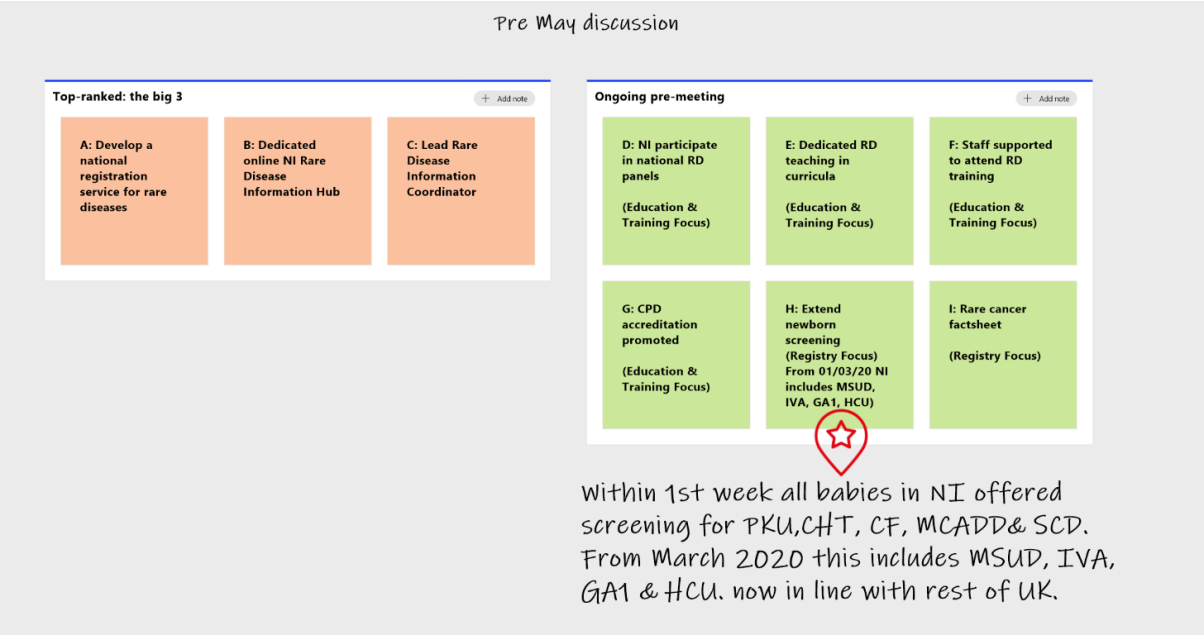


Figure 1: Three consistently top-ranked priorities and those with activities ongoing (May 2020)

In advance of the meeting all participants were requested to complete a short survey to rank their priorities prior to discussion at the meeting (Appendix 4: Cards for survey pre-meeting: ranking of short, medium and longer-term priorities). Accepting that the top three priorities were considered essential, and that several recommendations are being actively considered, participants were asked to rank the recommendations listed in figure 2 for priority action.

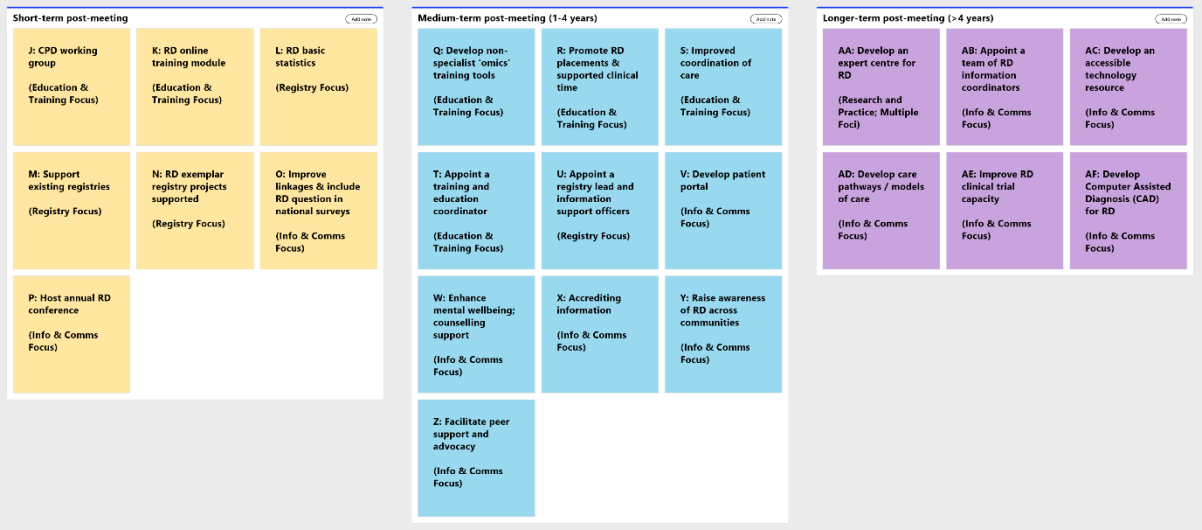


Figure 2: Recommendations for priority action pre-discussion

This survey was intended to provide a snapshot of the breadth of views where people placed the recommendations prior to discussion with stakeholders. There were some different priority rankings, but broad consensus was noted (Figure 3).

Based on this survey, the top three short-term priorities were:

i. **To generate rare disease basic statistics.** This includes for example, retrieving initial prevalence data, where available, for the 150 rare diseases that are believed to contribute the largest population burden to rare diseases.
ii. **To develop a rare disease online training module customised for NI.** More information is required for healthcare professionals that is accurate, easily accessible and targeted to NI resources.
iii. **To support rare disease exemplar registry projects.** Working collaboratively with colleagues across the UK, this will share best practice and ensure a NI rare disease and congenital anomaly register is aligned to national and international standards.

The top three medium-term priorities were:

i. **To improve coordination of care** to minimise duplication of effort and ensure appropriate expertise is available to provide improved care – more rare disease training where healthcare professionals listen to patients and are aware of multidisciplinary treatments would improve outcomes.
ii. **To appoint a registry and information support officers** who will work with a rare disease registry stakeholder team that meets quarterly including clinical specialists for validation of diagnoses.
iii. **To develop a patient portal** that will provide patients, and primary carers if necessary, with access to their electronic healthcare record through a patient portal embedded in

ENCOMPASS. This will also facilitate patient access to their medical records to help them coordinate their care between medical specialties and / or across different locations.

The top three longer-term priorities were:

- **To support a dedicated rare disease centre in NI** with sustainable funding. This should include development and maintenance of a (virtual) network of expert support to help effectively manage many rare diseases; development and coordination of a rare disease registry; a record of clinical nodes of expertise for specific conditions that are recorded on relevant sites such as Orphanet and support NI to participate in European research networks; support to facilitate NI participating in more clinical trials; a rare disease care coordinator to help develop models of care coordination, maintain an up-to-date listing of clinical expertise (local and centres of excellence); an education and training coordinator to help develop a NI Rare Disease Information Hub with relevant, accurate information and signposting to relevant resources where relevant; and protected clinical time (and administrative support) to enable clinical-academic research grants and engagement with UK-wide rare disease research initiatives. This would provide the systemic 'scaffolding' for an effective, cohesive, multidisciplinary Rare Disease Network across NI.

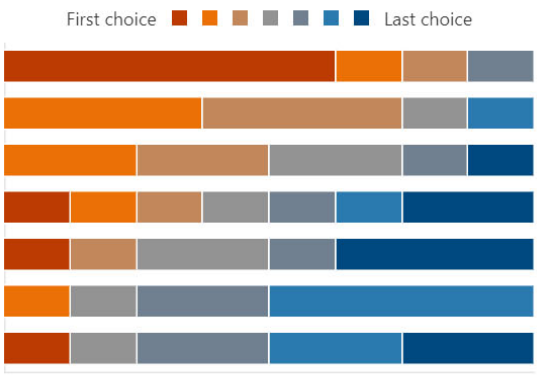
- **To appoint a team of RD information coordinators** who can facilitate access to information, both in person and through a dedicated telephone help/information line. This Team should also act as Northern Ireland's Orphanet representative, ensuring that local details are accurately maintained. "Contact points" should be established and maintained to facilitate information sharing and care coordination. There is a need to move away from the purely medical model to a more holistic social model, with a "whole systems approach" connecting patients, carers, allied health professionals, medical staff, and essential services such as education, employment, housing and transport. Coordinated by the lead rare disease information coordinator (ranked priority recommendation 3), one part-time person for each regional Hospital Trust would ensure the effective delivery of the direct patient support and liaison as piloted in the Stronger Together project)

- **To develop care pathways / models of care across the lifecourse.** Leading from pre-diagnosis through diagnosis to condition specific, person-centered care plans, led by the paediatric and adult rare disease clinical leads. Transparent models of care are required for individuals with a rare disease. These must be flexible, and include streamlined processes for accessing multidisciplinary care from experts who may be geographically distant where required)

1. Please rank the following short-term (actioned before end August 2021) priorities

More Details

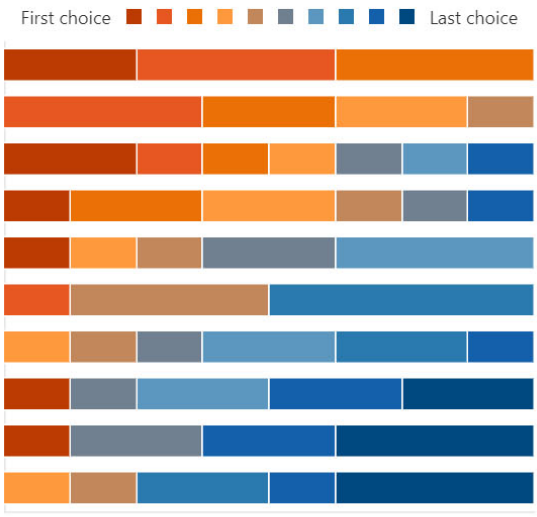
Rank	Options
1	L: RD basic statistics
2	K: RD online training module
3	N: RD exemplar registry proje...
4	M: Support existing registries
5	P: Host annual RD conference
6	O: Improve linkages & include...
7	J: CPD working group



2. Please rank the following medium-term (1-4 years) priorities

More Details

Rank	Options
1	S: Improved coordination of c...
2	U: Appoint a registry lead and ...
3	V: Develop patient portal
4	T: Appoint a training and educ...
5	Q: Develop non-specialist 'om...
6	R: Promote RD placements & ...
7	W: Enhance mental wellbeing;...
8	Y: Raise awareness of RD acro...
9	X: Accrediting information
10	Z: Facilitate peer support and ...



3. Please rank the following longer-term (4+ years) priorities

More Details

Rank	Options
1	AA: Develop an expert centre ...
2	AB: Appoint a team of RD info...
3	AD: Develop care pathways / ...
4	AC: Develop an accessible tec...
5	AE: Improve RD clinical trial ca...
6	AF: Develop Computer Assiste...

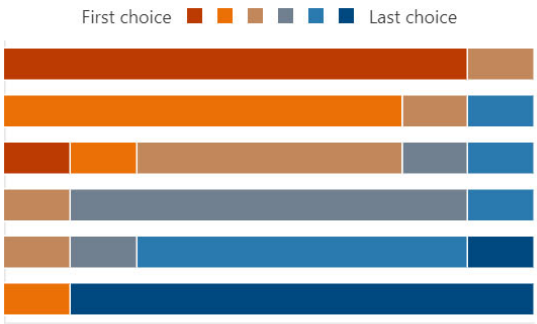


Figure 3: Summary of participant's individual ranking of priorities before the workshop took place.

Remote workshop discussions

Grace Henry hosted the workshop for 16 multidisciplinary participants who are active in the NI rare disease community. The workshop had the following objectives by the end of the session.

- To achieve a good understanding of the recommendations within the 3 reports.
- To review the three consistently top-ranked priorities from across the reports.
- To review all other recommendations and prioritise these within short, medium and longer term priority delivery timeframes.

And if time is permitting:

- Discussion on mechanisms of delivery and resources required.

Grace and AJ led the workshop with a discussion of the original three reports and workshop materials, which had been reviewed in advance by all participants. Multiple recommendations were overlapping between the three individual reports, so these were combined into a single recommendation where feasible; there was no additional weighting placed on such recommendations that were common to multiple reports. Participants considered if the current short, medium and longer term groupings were appropriate or if any should be changed. No ongoing work was identified that was not already recorded in the paperwork for this workshop. It was agreed that recommendations would need to be converted to SMART objectives at a later date, but that was outside the scope of this workshop. All participants agreed that we should consider the top-ranked priorities from across all reports and previous research as essential to rare disease progress in Northern Ireland.

Top-ranked (top 3) priorities for action:

1. To develop a national registration service for rare diseases in NI - NIRADCAR**:** Northern Ireland RAre Diseases & Congenital Abnormalities Registry. A collaborative steering committee with representation from universities, clinical and public health professionals, patient representatives, existing national registry leads, industry representatives, and encompass representatives should be created within six months to develop a dedicated NI rare disease registry model, with a formal rare disease registry plan launched within 12 months.

2. To develop a dedicated online NI rare disease information hub. This hub would link to ongoing programs and provide access to reliable rare disease clinical and non-clinical information. It was agreed as primarily signposting service that also facilities the development of strong interprofessional networks.

3. To develop a role for a lead rare disease information coordinator. This is foreseen as a non-medical role to complement the information hub, with the coordinator advising on rare disease accessible health and social care resources, research opportunities, events, developing rare disease networks and contact points, and acting as an Orphanet representative for Northern Ireland.

The pre-meeting survey results were discussed and it was agreed that the ongoing activities noted represented only a start to meeting individual recommendations; all other recommendations should be considered during the rest of the workshop. There was considerable discussion as to the best way

to prioritise recommendation – based on the needs for our rare disease community; resources (including funding) likely to be required to deliver the recommendations; short, medium, and longer-term timeframes, or by individual themes. Initially, recommendations were grouped by focused themes and colour coded into short-, medium- and longer-term goals, with participants agreeing that prioritising based on resource needs was not the goal or responsibility of this workshop (Figures 1 & 2).

Registry theme discussions. There is very poor recording of rare disease data across all care settings in Northern Ireland. All participants agreed that developing a national rare disease and congenital anomalies registry for NI (NIRADCAR) was essential, with the short-term priority to create a collaborative steering committee and 'launch' a registry plan within 12-24 months. Recommendations under the registry focus include incorporating newborn screening results, generating rare disease basic statistics including rare cancer data and factsheets in collaboration with the NI cancer registry, supporting existing registries, supporting exemplar projects with UK rare disease registries, and appointing a registry lead supporting information officers.

In addition to the research presented in the Jan 2020 registry report[22], there was considerable discussion about the logistics of these registry elements, including:

- Defining where NIRADCAR would be best placed for maximal benefit. Several options were discussed – are resources best placed to site this within the NI Public Health Agency (PHA), Business Services Organisation, Belfast Health and Social Care Trust, the proposed NI rare disease centre, or the Centre for Public Health within Queen's University Belfast. It was noted that multiple registries currently sit within QUB, including the national NI Cancer Registry (NICR) and associated subgroups such as the Barrett's Oesophagus register, a stroke register, and the Northern Ireland Cerebral Palsy Register (NICPR). The NICR receives core funding from the PHA, while the NICPR is commissioned by the PHA; both sit within QUB with required ethics and HSCT governance approvals to share data, alongside essential data security measures. Noted was the significant implications working within required legal and governance parameters, for example shorter-term ethics cycles with data access agreements needing approved every two years. Ultimately it was decided this location recommendation was outside the scope of the workshop and should be decided by the collaborative committee.

- We do not yet have an approved information and control of data processing bill for NI – and section 251 of the NHS Act 2006 does not apply here. The collaborative committee for NIRADCAR should have policy and practice guidance on NI's specific legislative requirements.

- While NIRADCAR is envisaged as the national resource for NI with an agreed minimum dataset (including facilitating incidence and prevalence annual reports) to inform rare disease health and social care services, it is understood that individual rare disease groups may wish to have more detailed information (e.g. natural history pathways) held in local registers, which would be externally funded. Multiple local registers/databases/lists of carefully curated rare disease information in NI is in danger of being lost due to retiring / moving staff or lack of resources

[22] McMullan, J., Crowe, A., Kerr, K., Bailie, C.... McKnight, A. J. (2020, Jan 31). Perspectives on a NI Rare Disease Registry. DOI 10.17605/OSF.IO/XU7P

to continuing updating required governance documents; it is essential to secure funding to ensure consistent, longevity and support sustaining existing registry data and rare disease biobanks.

- NIRADCAR should incorporate results from the newborn bloodspot screening program and the regional genetics service in the first instance.

- Privacy and consent for participation in the registry was discussed but agreed this was covered adequately in the original registry report, including opt-in vs opt-out models (extract below). 2017 research demonstrated that 84.2% (of 717 respondents in NI) agreed their anonymised personal data should be used to benefit 'non-commercial' rare disease research without explicit consent, where it is necessary to share data with commercial providers with specific expertise e.g. to develop clinical decision-making tools, participate in international rare disease registries, or smartphone apps, provided there is robust academic or NHS oversight. GDPR was discussed for information sharing – it is essential to share information internationally for rare disease progress. The NI rare disease registry team should continue to work with NCARDRS (England), CARIS (Wales), CADRISS (Scotland), Orphanet, RD-CONNECT, and the Irish National Rare Disease Office to ensure a NI rare disease and congenital anomaly register is aligned, where possible, to national and international standards and outcomes. NI should work collaboratively with ongoing UK-wide initiatives within rare disease registries for exemplar projects. It is recommended that a new workshop focused on privacy, consent and confidentiality specific to a rare disease registry in NI should be held to inform the NIRADCAR committee decisions.

  ***"Key theme 10:** The Registry must be capable of sharing information (subject to information governance requirements) with Registries elsewhere in the UK, in the Republic of Ireland, and internationally. Data sharing is critical for rare disease progress. It is essential that a core information dataset is recorded for rare disease patients in NI and harmonised to national / international registries to facilitate anonymised data sharing subject to local governance approvals. A data access committee should be established to consider requests for sharing identifiable data – sharing identifiable data for research purposes should be based on a fully consented opt-in model in order to comply with NI's current legislative requirements. Clarity is required for implementation of the HSC control of data processing legislation in terms of a fully consented / opt-out model for patient information."*

- Prioritising setting up the working group for this registry theme is essential – both to protect existing resources and to create an effective rare disease registry model. It was questioned if we could use COVID-19 as exceptional circumstances / special exemption where staff have been heavily committed to managing the pandemic and may not have been able to keep up with all governance requirements. ORECNI, Trust governance, QUB information compliance, and the Musaketeers' Memorandum were discussed.

- It was noted that correct coding is essential for a rare disease registry - there is a major opportunity to get it right with the development of this registry, online training materials for rare disease, and a focus on training coding input for encompass. Earlier preliminary research using the Child Health System for cerebral palsy or BSO's honest broker service for Bechet's Disease was suboptimal to capture cases. Existing disease-specific registries should adopt ORPHAcode nomenclature and work towards implementation of the common European data set to help standardise rare disease registration; including ORPHAcodes in encompass would

facilitate granular identification and retrieval of rare disease data. Consensus patient centred outcome assessments for existing registries should be developed in collaboration with patient representatives.

- Ideally NIRADCAR would link to education, social services and disability etc, but this is challenging. The first priority should be incidence and prevalence data with a view to obtaining longitudinal data for trajectory of care / received care linked to patient outcomes seen more as a research resource. Patient records and routine data collected across different health and social care providers (primary, secondary and community care) should be linked to provide the greatest opportunity to collect and validate rare disease data, quantify healthcare utilisation and track patient outcomes on a population basis. Patient-input data options should also be considered. A harmonised approach is essential with open access framework to facilitate transparent data sharing where required. Consideration of data linkages should be part of the NIRADCAR working group's processes.

- Ideally NIRADCAR would derive information primarily from encompass, with a 'yes/no' field included in the NI unique patient record (i.e. health and care number) asking if each individual has a rare disease. In the short-term, initial prevalence data may be derived via HSC Business Services Organisation and the regional genetics clinical information system using ICD codes and free text for the 150 rare diseases that are believed to contribute the largest population burden to rare diseases.

- It was emphasised that a question on rare disease should be incorporated to the minimum dataset for demography in national surveys to facilitate sub-group analyses, e.g. "Do you have a rare disease or Syndrome Without A Name (SWAN)".

- Sensitivities were discussed around the potential to use NIRADCAR for audit purposes, but it was recognised that this is useful tool to identify health services good practice and need for improvements. For example, the NICR contributes to regular local audits and international cancer datasets, while the data in the NICPR contributed to the 'Each and Every Need' report[23] for children and young people with complex medical needs.

Information and communication theme discussions. In addition to the research presented in the Jan 2020 information and communication report[24], there was considerable discussion about the logistics of these elements, including:

- A dedicated online NI rare disease information hub with complementary information coordinators acting as 'contact points' for patients, caregivers, health and social care professionals, promoting a "whole systems approach" connecting patients, carers, allied health professionals, medical staff, and essential services such as education, employment, housing and transport. This RDI hub should provide access (hosted, or signposted where possible) to reliable rare disease specific and generic clinical and non-clinical information, linking with ongoing programmes such as NI Direct, Digital Innovation Hubs through HDRUK, The UK Clinical Trials Gateway through NIHR, and Orphanet.

[23] The National Confidential Enquiry into Patient Outcome and Death. Each and Every Need. 2018. London
[24] McKnight, A. J., McMullan, J., Walker, R., & Collins, C. (2020, Jan 31). Communications and Information Review: NI Report for Rare Diseases. DOI 10.17605/OSF.IO/PUH5B

- NI is rebuilding the health agenda beyond transforming your care and in light of the COVID-19 pandemic. It was noted that COVID-19 has captured new ways of working that should be continued, such as telephone / video meetings with healthcare professionals where appropriate. However, COVID-19 has also raised challenges where many services have stalled and patients with rare diseases may be shielding for 12+ months.

- There is a need to liaise closely with the encompass team for info and comms elements – a working group should be set-up for the information hub and patient portal that would complement the NIRADCAR committee discussions.

- It would be ideal to work towards launching a registry plan and patient portal plan. People need to feel they are being kept in the loop and regularly updated on rare disease progress. Perhaps the information hub would have updates on the registry and portal progress? There is a need to be careful not to raise too many expectations- the plans should be strategically organised so they are deliverable and SMART.

- A strategy for a five year plan is improved coordination of care – how best to improve the quality of care? Care pathways may be seen as an interface between NIRADCAR and encompass strands. For example, some rare diseases have best practice guidelines published and awareness of these should be raised across NI – can a link be added to best practice guidelines (NICE / internationally published) associated with a disease diagnosis on a patient record?

- New 'general' rare disease care models have been described in RoI. Care pathways may fit within the information stream where registry audits can be used to improve quality of care for example. Raising awareness of established care guidelines may be a driver for improved care / service improvements and strategic funding considerations…being mindful of rare diseases where no published guidelines exist. *"To establish a care pathway leading from pre-diagnosis through diagnosis to condition specific, person-centered care plans, led by the paediatric and adult rare disease clinical leads. Transparent models of care are required for individuals with a rare disease. These must be flexible, and include streamlined processes for accessing multidisciplinary care from experts who may be geographically distant where required"*[25]. It was noted that participants were unaware of the names of any current paediatric or adult clinical leads for rare disease.

Education and training theme discussions. In addition to the research presented in the Jan 2020 education and training report[25], there was considerable discussion about the logistics of these elements, including:

- There is a peer program for nursing – Sandra Campbell highlighted she has been working with nursing educators at Ulster University (UU) to get rare disease teaching embedded into their curriculum. UU have a new medical school with rare disease lecturers keen to help integrate rare disease into the curricula. The need to harmonise rare disease teaching across NI (and beyond) was emphasised by participants – all agreed with discussions proposed to continue

[25] McMullan, J., Moore, K., & McKnight, A. J. (2020, Jan 31). A rapid scoping review of rare disease education needs for NI. DOI 10.17605/OSF.IO/UJ9KR

offline on how to progress this topic. Stephanie, Bill, Sandra and AJ to meet within two – four weeks to discuss options for collaboration and coordination of teaching.

- It was questioned if we need a continued professional working group if there is a training and education lead appointed. No consensus was reached on this discussion point. The need for protected clinical time for training and research was emphasised by participants.

Optimal approach. Recommendations were grouped by theme and according to revised delivery times (Figure 4). The registry theme had seven recommendations, the information and communications theme had 14 recommendations, the education and training theme had 11 recommendations while a cross-cutting theme was introduced for two recommendations – (i) developing an expert centre for rare diseases and (ii) improved coordination of care. However, this does not help deliver rare disease priority recommendations across all themes. Options as to how these should be best prioritised with a view towards recommended order for delivery were discussed. Taking a process-oriented approach, participants agreed an NI expert centre for rare disease was the overarching priority, which would enable and encompass all other elements (Figures 5). While it was recognised that developing a centre for rare disease would be a longer-term goal, it was agreed that this should be the overall aim with various elements that contribute to such a centre being feasible to introduce in the shorter term. ***Recommendations were consolidated under the proposed NIRADCAR and information hub.***

To establish an **effective rare disease registry**, the group agreed a collaborative steering committee should be established within six months and meet regularly. This committee, including legislative data expertise for NI, should be tasked with publishing a report within 12-18 months describing available data sources using a data model that can evolve over time as resources and infrastructure permit, barriers to data acquisition and data sharing, registry hosting, standardisation of coding, data linkages, required outcome measures, funding requirements, and a phased implementation plan. This committee should develop job descriptions to appoint required staff to lead and deliver an effective NI registry. Such staff should also liaise with the training and information coordinators to develop care pathways / models of care for rare diseases, providing important statistics that facilitate access to services. Longer term the committee should consider extending screening services and incorporating patient-derived data.

The second priority is to generate basic incidence data for rare diseases across NI in parallel with the steering committee discussions. The NICR should generate a factsheet summarising the incidence and prevalence of rare cancers in NI. NICR already collects data on cancers across Northern Ireland and shares some information on rare cancers internationally. However, the system is not ideally set-up to generate information on rare cancers so will require some staff time for coding and statistical analysis. Non-cancer data should be obtained from the newborn screening and regional genetics results in the first instance. Prevalence data should also be scoped from electronic healthcare records using ICD codes and free text for the 150 rare disease that contribute the largest population burden of rare diseases to inform the steering committee's report.

The third priority is to support exemplar projects for the NI rare disease registry, particularly those conducted in collaboration with UK-wide initiatives. Fourthly, existing and extended registries should be supported with standard infrastructure in a cost-effective manner. The recommendation to improve clinical trial capacity for rare diseases was shifted to the registry theme as it requires

knowledge of what data is collected for rare diseases and the number of individuals having particular diagnoses for patient participation and industry engagement.

Beyond the registry theme, a collaborative working group should be established within six months to oversee the information hub; this group would also facilitate generation of a report on rare disease model(s) of care for Northern Ireland within two years. A coordinator should be appointed as a training and education lead, as well as a complementary information coordinator – it was recognised coordinators may not be full time posts, but participants were keen to ensure there was a named person leading each of these important aspects of our rare disease delivery / action plan. Support staff helping to deliver the information recommendations would be beneficial delivering an effective service. It was recognised that improved diagnosis of rare diseases, improved coordination of care, and improved quality of life are all supported by training opportunities and access to accurate, readily accessible information.

Technology driven improvements to improve access to information may be facilitated by encompass for example developing the patient portal, exploring computer assisted diagnosis for rare diseases and accrediting information.

Training and education recommendations include the appointment of a training and education coordinator to oversee delivery of the recommendations. The need for a CPD working group was viewed as unnecessary provided a training and education coordinator was appointed who can liaise with HSC, Royal Colleges, University rare disease leads, ensuring NI training is aligned to GB and up to date *etc*. Short-term priorities included the development of online training modules for rare disease including a generic rare disease module targeted to NI resources, a module supporting appropriately coded data entry and retrieval for encompass, a module supporting understanding and returning results from whole genome sequencing, and a multi-omic medicine training module; these may be adapted from existing material elsewhere to have a NI focus. Dedicated teaching sessions should continue developing within University curricula, including supporting patients to have a voice in rare disease teaching. HSC staff should be supported to attend accredited training, avail of placement opportunities in specialist clinics, and CPD accreditation for events should be promoted where possible. Longer-term, NI representatives should be facilitated to participate in national panels with protected clinical time for supporting rare disease training, service development, and UK-wide rare disease research.

Information coordinator led recommendations overlap with that of the training and education coordinator, including improved signposting for healthcare professionals from pre-diagnosis to the development of a care plan for rare diseases; e.g. many doctors were unaware of the process in NI to consider orphan drugs or extra contractual referrals for their rare disease patients; many GPs struggle to identify and manage rare diseases. The information coordinator would provide signposting to relevant health and care services, as well as access to a network of expert professional support to manage many rare diseases more effectively. Improved coordination of care and access to expertise at external centres of excellence for rare diseases would minimise duplication of effort and ensure appropriate expertise is available to provide improved care. Working with the rare disease clinical leads for adult and paediatric care, as well as the registry lead and training leads, the information coordinator should facilitate developing care pathways / models of care for rare diseases, resulting in a report for publication.

Short-term priorities include improving signposting, data linkages and promoting inclusion of a rare disease 'question' (e.g. "Do you have a rare disease or Syndrome Without A Name (SWAN)") as part of the minimum dataset for demography in national surveys. Facilitation of an annual rare disease conference in collaboration with academic, industry, and third sector partners is the second short-term priority.

Longer-term, the information hub (an online resource and associated staff) should help raise awareness of rare disease across Northern Ireland by acting as the central knowledgebase for local rare disease information. It should host a map of key services, provide important signposting, host a diary of rare disease events, facilitate peer support (professional and third sector links), provide signposting to mental health and counselling support services, and host the training modules.

To maintain public confidence in rare disease progress, 'intention to deliver' plans should be shared with the rare disease community. Participants suggested that the registry plan and information hub should be launched in the short term, with later information releases including details of the patient portal, and a report for developing rare disease model(s) of care. Training modules should be launched as and when they become available. This structured approach will provide the framework for rare disease progress across Northern Ireland.

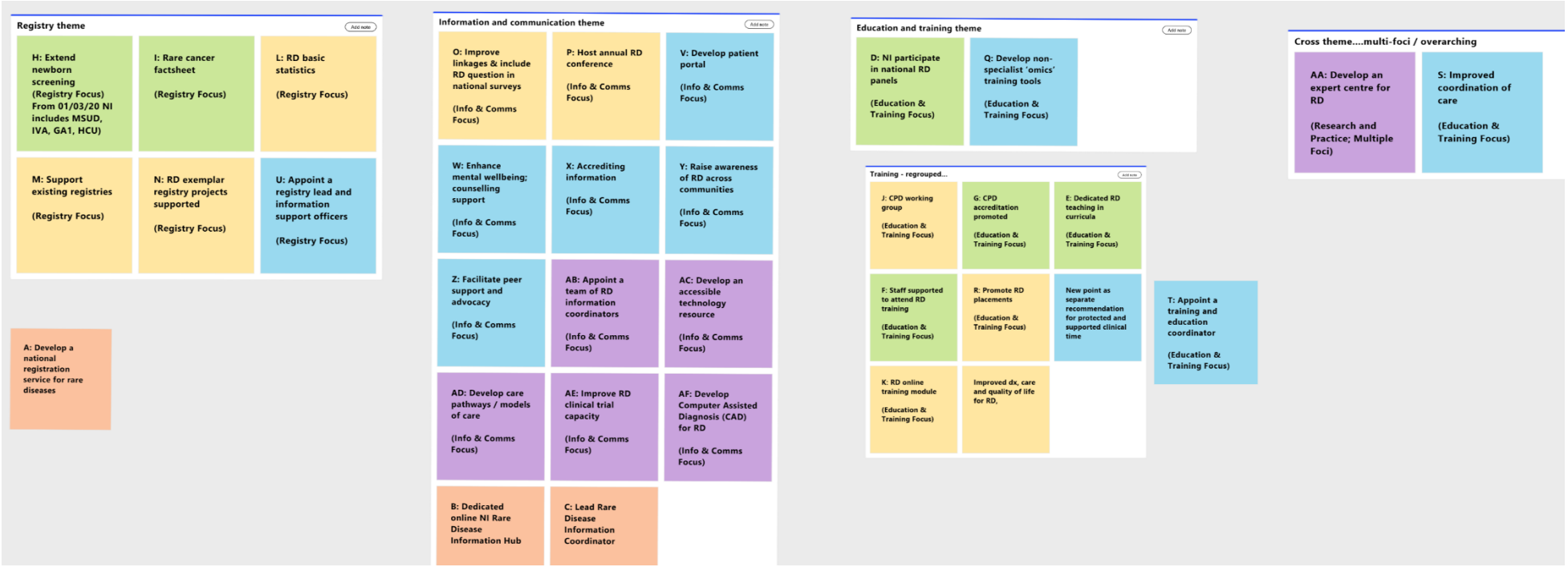


Figure 4: Priorities grouped under key themes during discussions, with several changed from short-, medium-, and longer-term priorities and a cross-cutting theme identified. The top-three priorities are peach, those where ongoing work is being delivered are green, short-term priorities are yellow, while medium-term are blue and longer-term are purple.

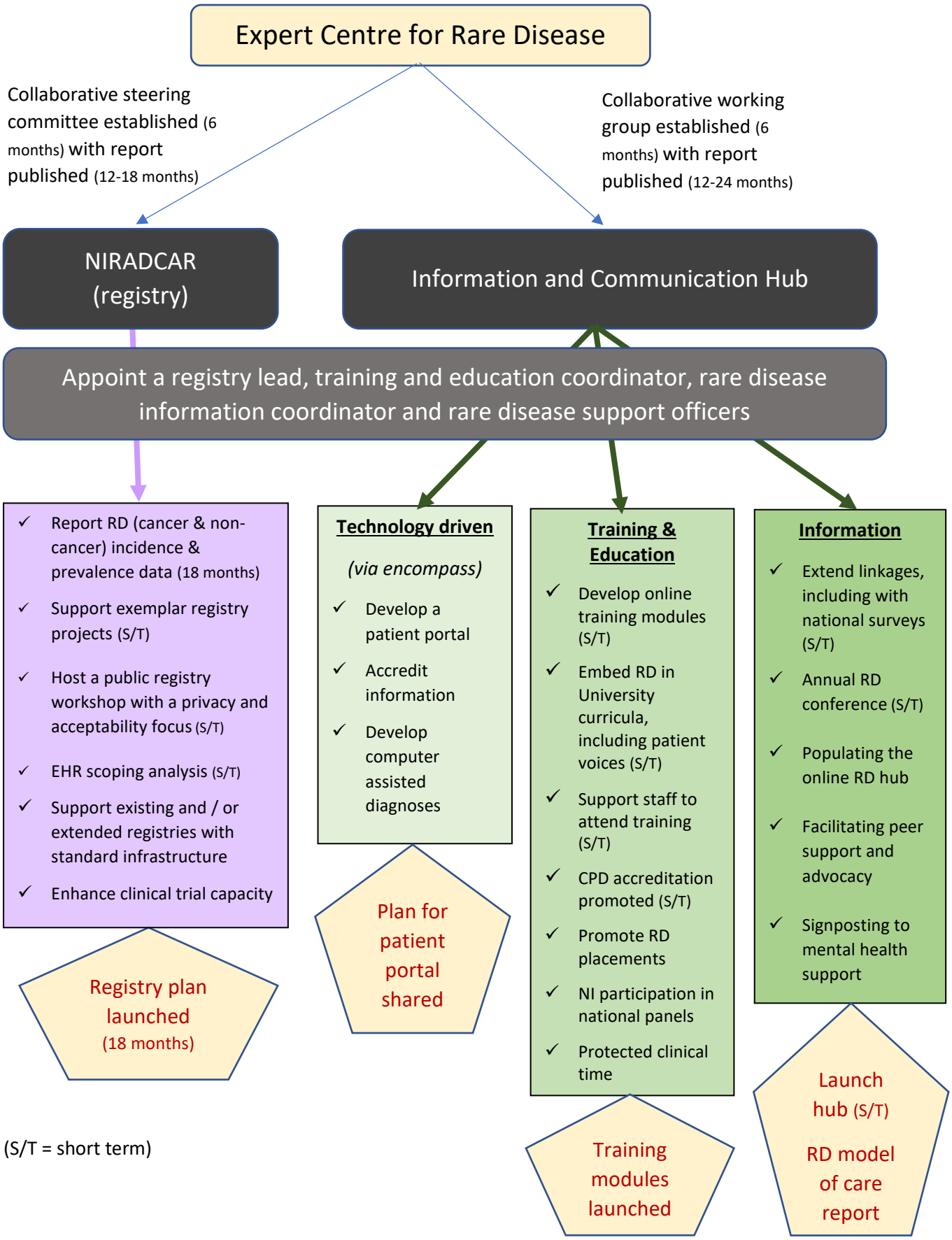


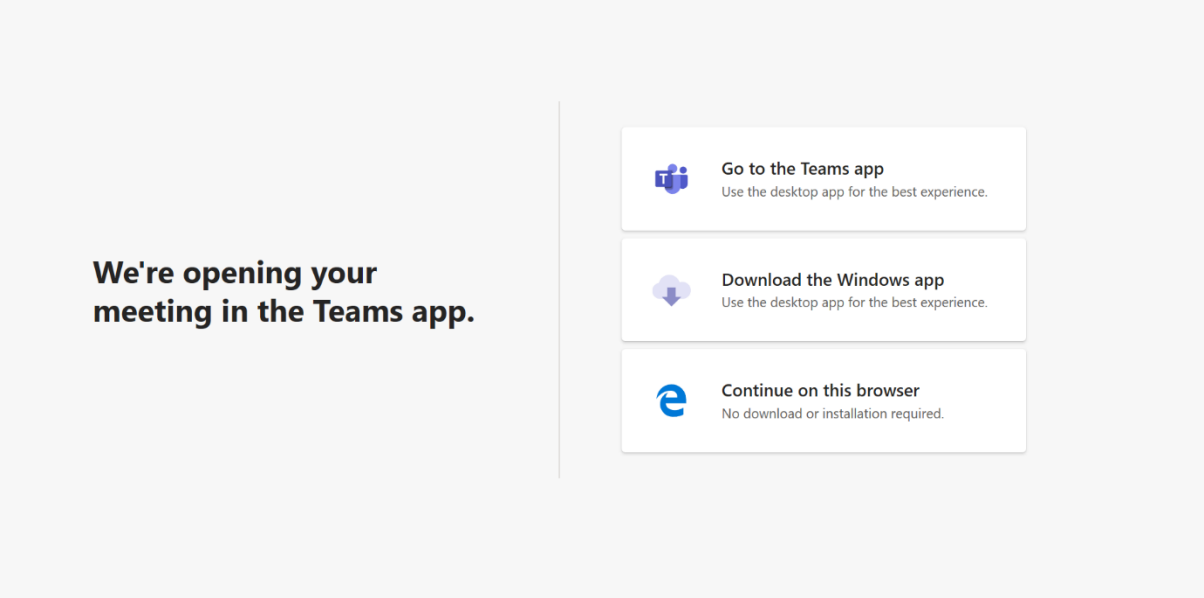
Figure 5: Workshop streamlined, prioritised recommendations for rare disease progress in NI

Appendix 1: An introduction to Teams…

https://www.microsoft.com/en-gb/microsoft-365/microsoft-teams/group-chat-software

Microsoft Teams is a program that facilitates 1:1 meetings, live events with up to 10,000 people, instant messaging, virtual conferences, and online 'team' meetings. It offers similar facilities to many other remote meeting options (such as sharing content, files, and screen sharing), but has the advantage of excellent security and data compliance measures alongside the typically high-quality audio, video, and sharing tools.

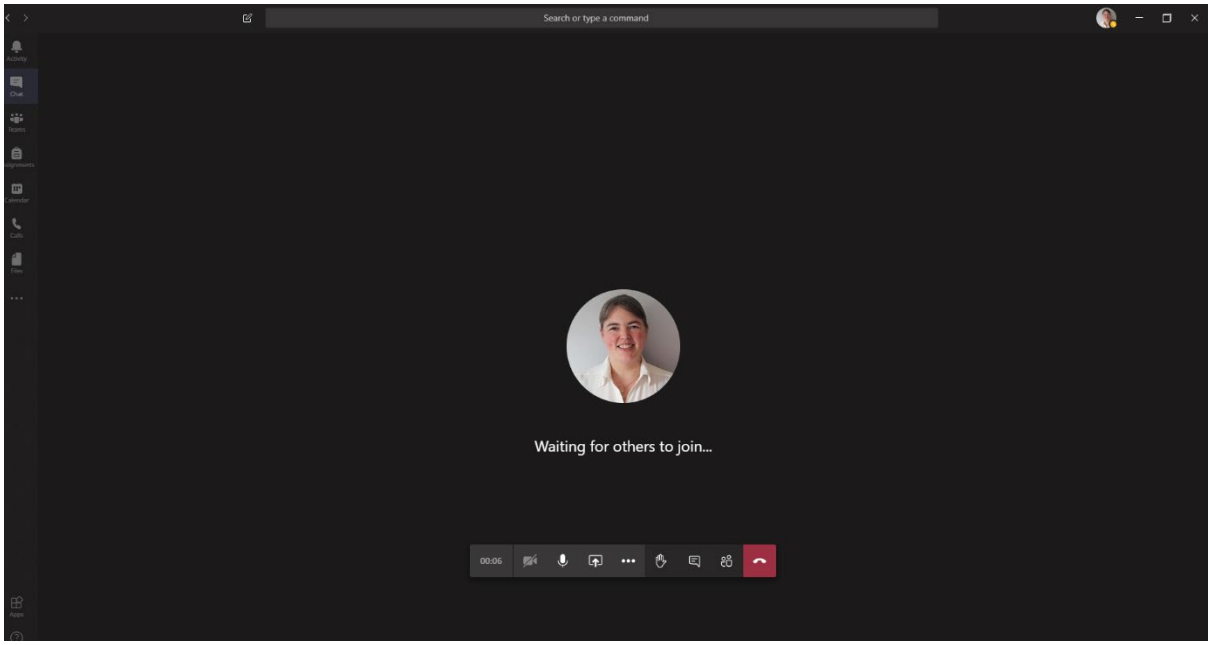
To join the conversation for this scheduled meeting, you can follow the link in your calendar invitation or in your usual Microsoft Teams account. It is not essential to have used or installed Microsoft teams before this meeting. You can alternatively follow this link to our meeting: Join Microsoft Teams Meeting, which may directly link to the meeting (depending on your own computer or telephone's settings) or present you with something similar to the following screen:



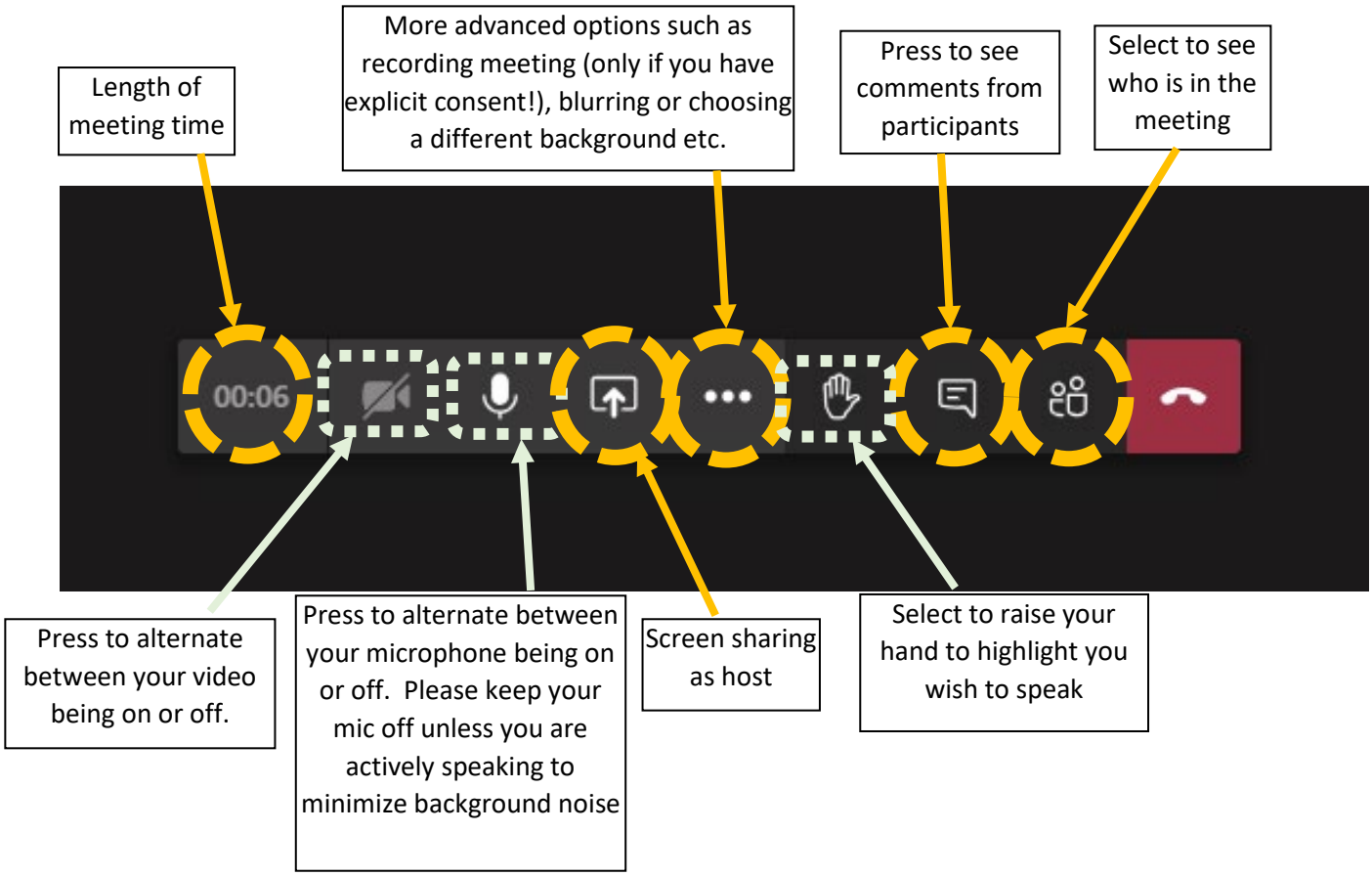
Any option will work and you may join with video, computer audio, or telephone audio as you prefer. *For those not subscribed to Office 365, it is usually easiest to continue on this browser / join via the web.*

Please note, if you already use Teams, then the email address you use to log in with should be the one that was used to send your invitation to this meeting. If you prefer to use a different email address, please let me know and I'll invite you using that new email address. If you have any trouble joining the meeting please do send me an email (a.j.mcknight@qub.ac.uk) or text message (07801 705514).

When you join the meeting you should see an image similar to that below. If participants have their video on then you see their faces. If there is no video then you will see their initials or a picture to indicate their presence. Remote meetings such as this work best if everyone's video is turned on, but if you are having 'bandwidth' issues (i.e. the video is slow or the connection is poor) then please feel free to turn your video off to improve the audio connection.



When in the meeting, you should see the bar with option buttons below. A short summary of their roles is below. **Please keep your microphone muted if you are not actively speaking.**



Appendix 2: Original recommendations from the three reports

Registry report

Key theme 1: NI should support the unanimous request from patients, as identified from public consultation, voluntary groups and healthcare providers, to develop a national registration service for rare diseases. A centralised NI rare disease resource (NIRADCAR: Northern Ireland RAre Diseases & Congenital Abnormalities Registry proposed) should strive to collect high quality data on every child identified with a congenital anomaly and every person living with a rare disease. There should be a formal 'launch' of a rare disease registry *plan*.

Key theme 2: That an annual factsheet should be produced by the NI cancer registry summarising the incidence and prevalence of rare cancers in Northern Ireland.

Key theme 3: The NI rare disease registry should incorporate results from the Newborn Bloodspot Screening Program.

Key theme 4: That existing and developing rare disease specific registries should be supported to continue until a standardised infrastructure is in place, which would enable them to be future-proofed and integrated with national IT systems using a unique personal identifier (such as the Health and Care Number[26]) to enhance interoperability and integration. Such registries should be supported to incorporate Orphacode nomenclature[27] and EU data standards to align local rare disease registration with international standards and facilitate engagement with international registries and clinical trials. A Registry infrastructure should be developed to provide administrative support, and technical help for more detailed disease-focused databases, registers, resources under an inclusive 'rare disease registry and bioresource' ethics and governance approval.

Key theme 5: Correct coding and accurate data input is essential for a successful registry. Rare disease diagnoses should be aligned to Orphanet codes and a working group created with the ENCOMPASS team and the registry lead to discuss how this can be integrated with the digital integrated care records within Encompass[28], aligned with other coding systems such as ICD and SNOWMED CT. Healthcare professionals should be trained in appropriate coding for rare diseases to improve data accuracy and recording.

Key theme 6: There is a need for robust mechanisms to be developed to ensure developmental milestones are established and met with relevant stakeholder involvement initiated and maintained through an inclusive steering committee. A collaborative steering committee with representation from universities, clinical and public health professionals, patient representatives, existing national registry leads, industry representatives, and encompass representatives should be created within six months. The registry must align with, and link to, the developing NI patient and service user information infrastructure. Patient representatives are essential members of the registry stakeholder team. An annual report should be generated reporting outcomes and progress with the registry.

[26] http://www.hscbusiness.hscni.net/services/2748.htm
[27] http://www.orphadata.org/cgi-bin/index.php
[28] http://www.hscboard.hscni.net/encompass/

Key theme 7: The NI rare disease registry team should continue to work with NCARDRS (England), CARIS (Wales), CADRISS (Scotland), Orphanet, RD-CONNECT, and the Irish National Rare Disease Office to ensure a NI rare disease and congenital anomaly register is aligned, where possible, to national and international standards and outcomes. NI should work collaboratively with ongoing UK-wide initiatives within rare disease registries for exemplar projects.

Key theme 8: A multi-source approach should be employed including secure access to routine HSC data feeds with a model developed for accurate case ascertainment. There are multiple diverse information sources that capture local information on rare diseases, but at present these do not have an adequate coding system recording relevant information for a comprehensive congenital abnormality and rare disease registry. A registry lead and at least two information support officers should be employed to work with a register stakeholder team that meets quarterly including clinical specialists for validation of diagnoses. The NI rare disease registry should be developed to derive information primarily from encompass, with a 'yes/no' field included in the patient record asking if each individual has a rare disease. In the short-term, initial prevalence data should be derived via HSC Business Services Organisation and the regional genetics clinical information system using ICD codes and free text for the 150 rare diseases that are believed to contribute the largest population burden to rare diseases. Local participants contributing to national programs such as the 100,000 genomes project already have data cleaned and verified for inclusion in a NI rare disease registry.

Key theme 9: A combined rare disease registry model should be created with the core, 'quality assured' dataset collected for incidence, prevalence, service planning, and potential clinical trial participation. The Registry should have scope for staff 'placements' supported by external funding to develop more focused (e.g. disease specific resources) that enhance the rare disease registry infrastructure. Data linkage *via* the Honest Broker Service may be used to develop a more extensive dataset for research and health service use with appropriate approvals. More details such as linking with apps, wearable devices, care of particular rare diseases could be supported by relevant charities through partnership working, but note that not all rare diseases will have a charity or funds to support such a study, which disadvantages patients without a large 'charity' voice.

Key theme 10: The Registry must be capable of sharing information (subject to information governance requirements) with Registries elsewhere in the UK, in the Republic of Ireland, and internationally. Data sharing is critical for rare disease progress. It is essential that a core information dataset is recorded for rare disease patients in NI and harmonised to national / international registries to facilitate anonymised data sharing subject to local governance approvals. A data access committee should be established to consider requests for sharing identifiable data – sharing identifiable data for research purposes should be based on a fully consented opt-in model in order to comply with NI's current legislative requirements. Clarity is required for implementation of the HSC control of data processing legislation in terms of a fully consented / opt-out model for patient information.

Key theme 11: An open source framework should be used so that data is not limited to a specific vendor and can be extensively reused.

Key theme 12: Longer-term, consideration should be given to complementary direct patient input to research registries with possibility of case confirmation by interested clinicians and potential for long-term data collection, support forums and groups, and information resource linked to a NI rare disease information hub. A working group to evaluate this option should include utility, validation, and information governance issues.

Key theme 13: Taking advantage of planned computational developments across Northern Ireland a reliable, flexible 'Clinical Trial Ready' Rare Disease Registry that provides accurate disease data would enable complementary innovative analytics to be developed that could be embedded into routine care and used to inform clinical decision-making tools and health services planning, thus positioning the NI rare disease registry as a 'go-to' local resource for rare disease research, pending regulatory approvals.

Information and communication report

Priority recommendations:

Priority 1. To develop a dedicated online NI Rare Disease Information Hub that provides access (hosted, or signposted where possible) to reliable rare disease specific and generic clinical and non-clinical information. This resource should link with ongoing programmes such as NI Direct, Digital Innovation Hubs through HDRUK, The UK Clinical Trials Gateway through NIHR, and Orphanet.

The following components were repeatedly requested within an online Rare Disease Hub:

13) Disease specific information for individuals living and working with rare diseases, including signposting to expert standards of care guidelines
14) Access to referral resources e.g. physio, genetics, counselling, treatment options, and emergency telephone numbers (e.g. to resolve wheelchair, hoist, malfunctions)
15) A diary of events, conferences, workshops and experts invited to NI
16) Access to work, educational support, and benefit related information
17) A list of local and national experts and charities / support groups for many rare diseases
18) Map of key services e.g. accessible hydrotherapy and accessible leisure facilities
19) List of research and a map of local rare disease research projects / developments
20) Regular email updates / newsletter with updates from the Hub
21) Discussion forum / networking facility
22) Online training modules; generic and specific
23) Specialised rare disease training for professionals

Priority 2: To develop a centralised NI registry (NIRADCAR: Northern Ireland RAre Diseases & Congenital Abnormalities Registry) / registries for rare disease linked to Encompass.

Priority 3: A team of Northern Ireland Rare Disease Information Coordinators should be appointed to facilitate access to information, both in person and through a dedicated telephone help/information line. This Team should also act as Northern Ireland's Orphanet representative, ensuring that local details are accurately maintained. "Contact points" should be established and maintained to facilitate information sharing and care coordination. There is a need to move away from the purely medical model to a more

holistic social model, with a "whole systems approach" connecting patients, carers, allied health professionals, medical staff, and essential services such as education, employment, housing and transport.

Note: in the list of priorities identified by colleagues in the Republic of Ireland, our NI Priority 3 does not appear, however they have established their National Rare Disease Office, with associated website, and National Clinical Programme for Rare Diseases. Urgent consideration should be given to appointing a Lead Rare Disease Information Coordinator to fulfil the critical role of*, 'A person who maintains a list of information and advises on accessible health and social care resources. This rare disease information coordinator should have a non-medical role directing / sign-posting individuals to appropriate health and social care resources. This includes keeping a record of ongoing clinical trials, research opportunities, rare disease events in NI'.* This is a demanding role with a substantial commitment that should include advising on content within the online information hub, developing networks and contact points within each 'system' to efficiently advise patients, carers, professional colleagues, and importantly acting as the Orphanet representative for Northern Ireland. Coordinated by this senior appointment, one part-time person for each Hospital Trust area at a lower grade would ensure the effective deliver of the direct patient support and liaison piloted in the Stronger Together project.

Priority 4: To continue developing dedicated rare disease training for local professionals, using a mixed approach of interactive sessions, seminars, training courses, printed material, and online training modules. Building on that developed in England and the Republic of Ireland, there should be an online rare disease presentation developed to highlight rare disease facts in Northern Ireland and signpost individuals to relevant support sites for local professionals.

Priority 5: To establish a care pathway leading from pre-diagnosis through diagnosis to condition specific, person-centered care plans, led by the paediatric and adult rare disease clinical leads. Transparent models of care are required for individuals with a rare disease. These must be flexible, and include streamlined processes for accessing multidisciplinary care from experts who may be geographically distant where required.

Note: a substantial proportion of participants defined a 'rare disease coordinator' as a healthcare professional who coordinates complex clinical care needs, or as a personal care coordinator to help individuals with rare diseases manage their health and social care needs.

These are two distinct roles. The first sits clearly within the medical teams, where some current provision exists but would benefit from increased clarity and improved pathways. The second is focussed on providing information and support for the individual in their interactions with their medical and social environment.

Priority 6: To provide patients, and primary carers if necessary, with access to their electronic healthcare record through a patient portal embedded in ENCOMPASS.

Priority 7: To provide resilience training and enhance the mental wellbeing of patients and carers through access to relevant services. Links to existing services should be signposted from the NI Information Hub. Counselling support should routinely be made available to individuals (patients, carers, other family members) struggling with the impact of a rare disease.

Priority 8: That a question on rare disease should be incorporated to the minimum dataset for demography in national surveys to facilitate sub-group analyses, e.g. "Do you have a rare disease or Syndrome Without A Name (SWAN)".

Priority 9: To improve capacity and infrastructure to enable individuals with rare diseases to participate in research and clinical trials.

Priority 10: To host an annual rare disease conference to share best practice, update on rare disease progress, introduce new initiatives, and provide networking opportunities.

Additional recommendations include:

- To facilitate patient access to their medical records to help them coordinate their care between medical specialties and / or across different locations.
- To consider 'accrediting' alert cards, flyers, and / or patient passports that patients could carry with information about their condition in case of emergency. Many such cards and passport style documents exist, but without clinical 'accreditation' or an 'NHS stamp', they are often dismissed.
- To consider computer assisted diagnosis of rare diseases within / complementing ENCOMPASS.
- To facilitate improved communication and the development of strong inter-professional networks (MDTs, ERNs, Project ECHO, etc.)
- To improve linkages with ongoing projects including national surveys and the 10,000 Voices team.
- To raise awareness with schools and employers as to the complex nature of rare diseases, which often necessitates multiple hospital appointments at short notice. Flexible working for individuals with disabilities and those with significant caring responsibilities should be promoted throughout the workforce.
- To facilitate peer-support and training of people living with rare diseases to act as peer support, peer mentors, and advocate in relevant health and community panels.
- To collate details of supportive / assistive technologies available in Northern Ireland and develop a resource where such details are maintained and accessible. At a minimum this should be provided for occupational therapy and educational resources, and ideally link to/from the online Rare Disease Information Hub.
- That consideration be given to expanding the newborn blood spot screening in Northern Ireland (currently five conditions) to encompass more conditions where early intervention can significantly improve patient outcomes.
- That a dedicated rare disease centre in Northern Ireland should be supported with sustainable funding. This should include:
  - development and coordination of a rare disease registry (with room for 'placements' supported by external funding to develop more focused (e.g. disease specific resources) that enhance the rare disease registry infrastructure)
  - a record of clinical nodes of expertise for specific conditions that are recorded on relevant sites such as Orphanet and support NI to participate in European research networks
  - support to facilitate NI participating in more clinical trials
  - a rare disease care coordinator to help develop models of care coordination, maintain an up-to-date listing of clinical expertise (local and centres of excellence)
  - an education and training coordinator to help develop a NI Rare Disease Information Hub with relevant, accurate information and signposting to relevant resources where relevant

- protected clinical time (and administrative support) to enable clinical-academic research grants and engagement with UK-wide rare disease research initiatives.
- This would provide the systemic 'scaffolding' for an effective, cohesive, multidisciplinary Rare Disease Network across NI.

Training and education report

Recommendations for a NI training action plan:

Recommendation 1: To establish a three-year working group that meets quarterly with input from the HSC education and training team, training and education leads from the NI genomic medicine centre, relevant Royal Colleges, and University leads delivering genomic medicine, to conduct annual training needs 'update' analysis for rare diseases and identify synergies with potential to adapt existing training material in this rapidly evolving field.

Recommendation 2: NI should maintain updated knowledge to ensure NI training is aligned to that in GB. HEE is working with a range of stakeholders to ensure genomics is embedded in training curricula and revalidation requirements to ensure sufficient genomics understanding by all staff involved in clinical decisions; NI representatives should be supported to participate on relevant national panels.

Recommendation 3: Dedicated rare disease teaching sessions should continue to be incorporated to undergraduate medical, nursing, and biomedical university curricula, including providing signposting to useful rare disease resources such as Orphanet. Teaching methods and clinical techniques for genomic medicine should be formally incorporated to taught curricula.

Recommendation 4: To continue developing dedicated rare disease training for local professionals, using a mixed approach of interactive sessions, seminars, training courses, printed material, and online training modules. An online training module for rare disease in NI should be developed to highlight rare disease facts in Northern Ireland and help raise awareness of relevant rare disease resources (such as Orphanet) for healthcare professionals. Collaborative work should be undertaken with the Northern Ireland Medical and Dental Training Agency, Queen's University Belfast, Ulster University and Royal Colleges to develop accredited training tools. Patients with a rare disease and their carers should be involved in the development of such training programs. More information is required for healthcare professionals that is easily accessible – GPs in particular emphasised the need for more information to better manage their rare disease patients.

Recommendation 5: Further training of mainstream (non-genetic clinical specialists) would enhance the efficiency of the 'genomic medicine' process in terms of enhancing the information returned from Genomics England and conveyed to recruiting clinicians, initial clinician review of variants, and in-depth analysis of variants. A training tool should be developed highlighting best practice for interpreting and returning results from WGS locally as part of mainstreaming genomic medicine.

Recommendation 6: Consideration should be given to supporting individuals from the existing workforce in NI to attend postgraduate training courses in genomic medicine. Due to the number of individuals needing training locally consideration should be given to developing an accredited MSc compatible with the UK scientist training program (STP). HSC staff should continue to be supported to undertake clinically relevant rare disease PhD research, for example through HSC or ICAT schemes.

Recommendation 7: CPD accreditation should be sought for rare disease conferences and training days, which should be circulated on Trust and University websites with individuals supported to attend where possible. Dedicated training events for specific rare diseases / rare disease groups with input

from international experts were welcomed – but online training is preferred. Ideally such events would be posted on a centralised rare disease 'events' website.

Recommendation 8: Rare disease 'Placement' opportunities and protected clinical time should be available to support medical, nursing and associated healthcare professionals to gain practical experience in specialist clinics.
Recommendation 9: Patients should be further supported to be involved in the development and delivery of rare disease training programs. Supporting patients to become educators, 'patient voices' videos should be developed where rare disease patients may not be able to attend scheduled teaching sessions due to the nature of their condition. Patients are ideally placed to communicate their lived experience as a fundamental message to health and social care professionals at undergraduate and postgraduate levels.

Recommendation 10: An online Information Hub should include a directory of accredited educational resources for rare diseases, including those identified by this review.

Recommendation 11: Improved signposting for healthcare professionals is required from pre-diagnosis to the development of a care plan for rare diseases. For example, many doctors were unaware of the process in NI to consider orphan drugs or extra contractual referrals for their rare disease patients. Improved coordination of care would minimise duplication of effort and ensure appropriate expertise is available to provide improved care – more rare disease training where healthcare professionals listen to patients and are aware of multidisciplinary treatments would improve outcomes.

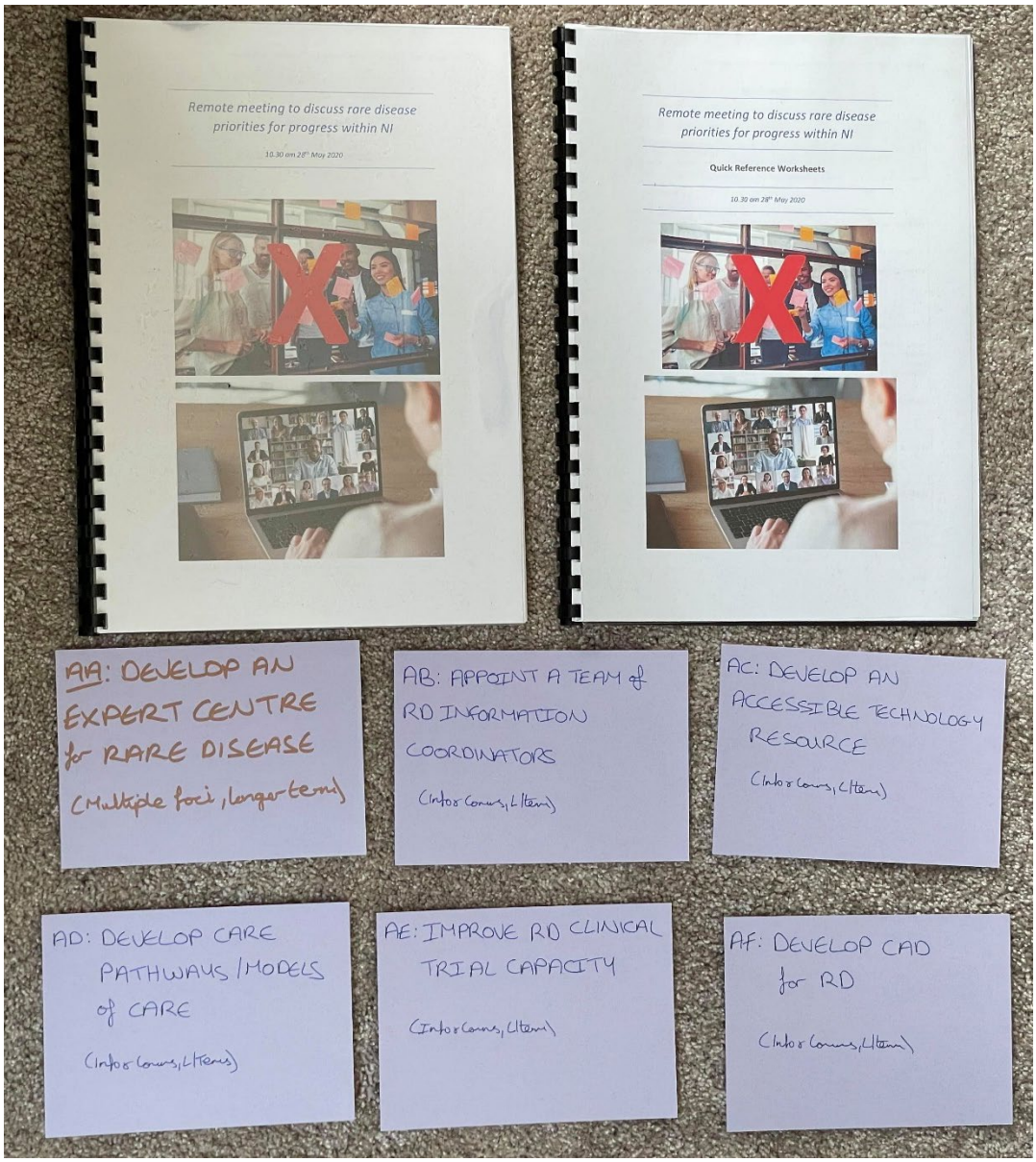
Recommendation 12: A network of expert support is required to effectively manage many rare diseases – consideration should be given to developing an expert centre(s) for rare disease where appropriate, and supporting clinicians to participate in UK and International Reference Networks such as European Reference Networks and Clinical Interpretation Partnerships. Virtual reference networks enable healthcare professionals to connect to experts on particular rare disease issues and facilitate patients benefiting from expertise at external centres of excellence for rare diseases.

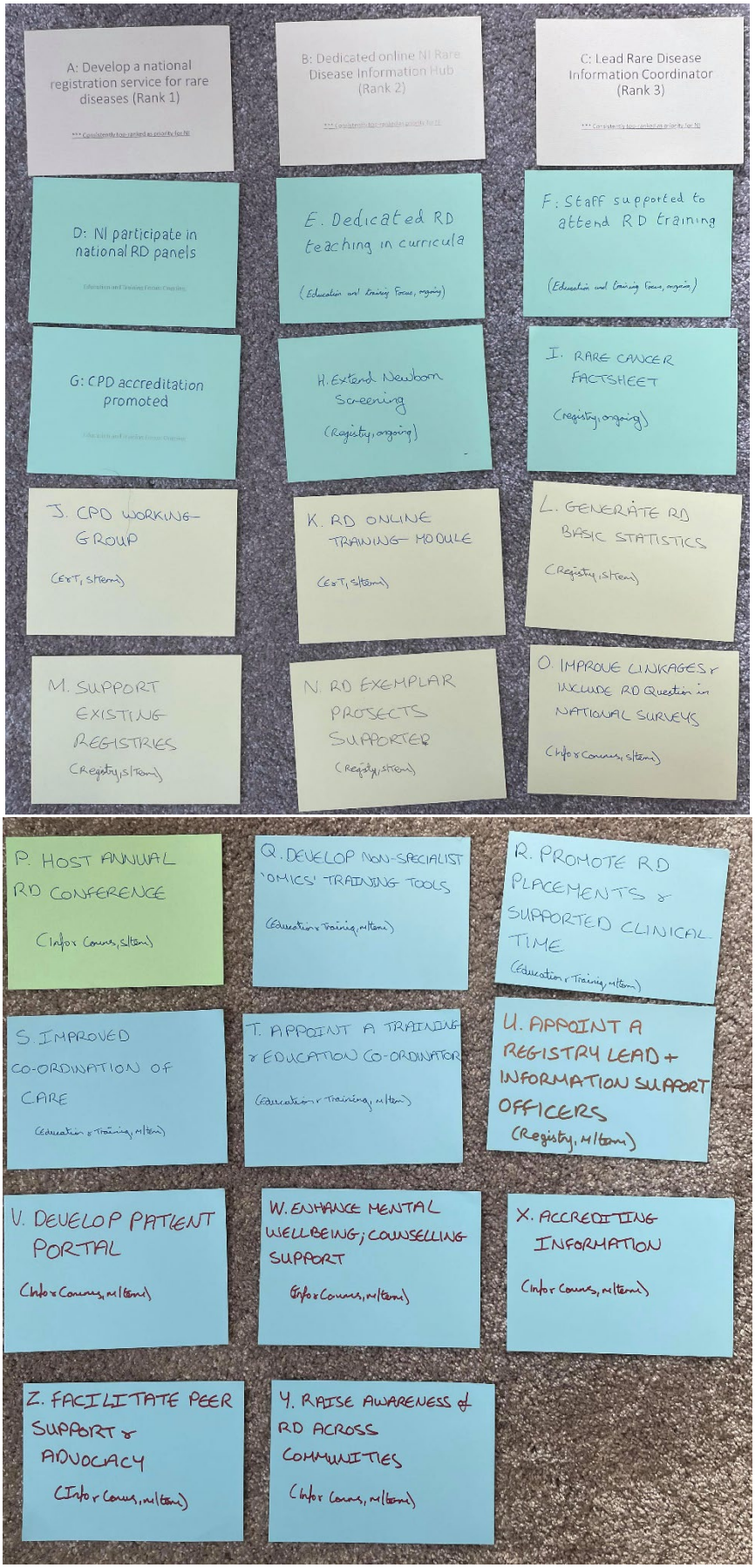
Recommendation 13: That a training and education development coordinator be appointed for rare diseases to act as a primary point of contact for:

a) Signposting and connecting individuals living and working with rare diseases seeking to undertake formal / informal training.
b) Maintaining an updated list of training opportunities and liaising with speciality leads to identify and coordinate ongoing training and education needs
c) Supporting charities organising education and training events in NI in terms of appropriate accreditation, signposting, and disseminating information.
d) Connecting Royal Colleges for accredited training courses and promoting CPD opportunities for rare disease to relevant health and social care staff.
e) Working with Royal Colleges and local health and social care professionals to develop local training resources to meet education and training needs.
f) Working with the Centres for Medical Education, Biomedical Sciences, Public Health, and Nursing at Queen's University Belfast, alongside the School of Nursing and planned medical school at Ulster University to develop new teaching tools and further embed rare disease teaching across Northern Ireland.
g) Working with the Encompass teams to develop an online rare disease training tool to support appropriately coded entry of data and rare disease information retrieval.

h) Develop a rare disease online training tool for GPs and other healthcare professionals – adapted from that existing elsewhere to support people working in NI's health and social care system and legislative environment.
i) Developing an online genomic medicine training tool – MSc / PGC
j) Developing an online training tool / website links for accessing orphan drugs and extra contractual referrals with a clear pathway to access described

Appendix 3: Selected participant materials, including colour-coordinated A6 cards with recommendations




A: Develop a national registration service for rare diseases (Rank 1)
B: Dedicated online NI Rare Disease Information Hub (Rank 2)
C: Lead Rare Disease Information Coordinator (Rank 3)
D: NI participate in national RD panels
E. Dedicated RD teaching in curricula
(Education and training Focus, ongoing)
F: Staff supported to attend RD training
(Education and training focus, ongoing)
G: CPD accreditation promoted
H. Extend Newborn Screening
(Registry, ongoing)
I. RARE CANCER FACTSHEET
(registry, ongoing)
J. CPD WORKING-GROUP
(CEoT, s/term)
K. RD ONLINE TRAINING- MODULE
(CEoT, s/term)
L. GENERATE RD BASIC STATISTICS
(Registry, s/term)
M. SUPPORT EXISTING REGISTRIES
(Registry, s/term)
N. RD EXEMPLAR PROJECTS SUPPORTED
(Registry, s/term)
O. IMPROVE LINKAGES & INCLUDE RD Question in NATIONAL SURVEYS
(Info & Comms, s/term)
P. HOST ANNUAL RD CONFERENCE
(Info & Comms, s/term)
Q. DEVELOP NON-SPECIALIST 'OMICS' TRAINING TOOLS
(Education & Training, m/term)
R. PROMOTE RD PLACEMENTS & SUPPORTED CLINICAL TIME
(Education & Training, m/term)
S. IMPROVED CO-ORDINATION OF CARE
(Education & Training, m/term)
T. APPOINT A TRAINING & EDUCATION CO-ORDINATOR
(Education & Training, m/term)
U. APPOINT A REGISTRY LEAD + INFORMATION SUPPORT OFFICERS
(Registry, m/term)
V. DEVELOP PATIENT PORTAL
(Info & Comms, m/term)
W. ENHANCE MENTAL WELLBEING; COUNSELLING SUPPORT
(Info & Comms, m/term)
X. ACCREDITING INFORMATION
(Info & Comms, m/term)
Z. FACILITATE PEER SUPPORT & ADVOCACY
(Info & Comms, m/term)
Y. RAISE AWARENESS of RD ACROSS COMMUNITIES
(Info & Comms, m/term)

Appendix 4: Cards for survey pre-meeting: ranking of short, medium and longer-term priorities