Healthier Scotland
Scottish Government

Rare Disease
Action Plan

Contents

Ministerial Foreword, Maree Todd MSP, Minister for Public Health, Women's Health and Sport

Rare diseases may be individually rare, but they are collectively common: it is estimated that around 412,080 people in Scotland have a rare disease.

The Scottish Government is absolutely committed to ensuring that all those people living with a rare disease are able to access the best possible care and support, and benefit from healthcare services that are safe, effective, and person-centred.

This is a significant challenge for our healthcare services: although rare diseases are collectively widespread, there are thousands of different rare diseases and each one can require highly specialised treatment.

This means that people living with rare diseases can face the prospect of a long 'diagnostic odyssey' before they are accurately diagnosed and able to access treatment. The various aspects of their treatment and care also often lack coordination.

It is vital then that we continue to strive for improvement and ensure that all those living with a rare disease get the right diagnosis faster and can access coordinated care and specialist treatment.

We have worked closely with the UK Government and other devolved nations to produce a new UK-wide Rare Disease Framework in response to the concerns raised by the rare disease community in the National Conversation on Rare Diseases survey.

This Action Plan has been developed through significant engagement with the rare disease community and I thank Genetic Alliance UK for their facilitation of this. Through our combined efforts we can ensure that we are putting those living with a rare disease at the heart of policymaking, ensuring this meets the needs of the rare disease community in Scotland whilst reaping the benefits of a four-nations approach.

In addition to this, the work of the Rare Disease Implementation Board (RDIB) for Scotland continues to lead the way in our efforts to support the rare disease community. I would like to thank Dr Jonathan Berg for his chairing of RDIB since its inception and am pleased that our Genomics work will continue to benefit from his expertise. RDIB members and their new chairperson will continue to steer our direction of travel over the lifespan of the UK Framework.

While we made good progress through the UK Rare Disease Strategy in 2013 under our previous implementation plan - *It's Not Rare to Have a Rare Disease*[1] - we recognise that there is continued room for improvement, and that there are always new challenges to address.

This plan will feel different to our previous documents. We want this plan to evolve over time and be reflective of the changing rare disease landscape. Our plan will be reviewed regularly with actions refreshed and renewed, taking advantage of the innovations we have seen as a result of our recovery from the Covid-19 pandemic amongst other advances in diagnostic therapies. We will continue to engage with the rare disease community throughout this process and beyond to hold us to account on our actions, ensuring we are ambitious in our delivery for people with a rare disease.

I am confident that the actions set out here will ensure we continue to make progress in how rare diseases are researched, diagnosed and treated here in Scotland, and will make a real difference for people in Scotland living with a rare disease.

Maree Todd MSP

1 It's Not Rare to Have a Rare Disease - gov.scot (www.gov.scot)

Dr Jonathan Berg, Senior Lecturer and Honorary Consultant in Clinical Genetics, University of Dundee

Outgoing Chair of the Scottish Rare Disease Implementation Board (2021-2022)

The UK Rare Disease Framework has provided us with a platform for real change in Scotland. With this, and with the flexibility of our own Action Plan we are able to take a different approach than perhaps what we have seen in the past.

Scotland's Rare Disease Implementation Board (RDIB) was convened to respond to the priorities of the Framework, and work towards Scotland's Action Plan for Rare Disease, the first iteration of which I am delighted to see published. Over the course of RDIB meetings to date, we have explored in 'deep dives' how to get the best out of our forthcoming registry, the ways in which we can support the Awareness Raising priority, and how to encourage participation in research and unlock barriers to rare disease research.

To enhance our understanding of action required to shorten the diagnostic odyssey and improve care coordination, we created working groups within RDIB which were tasked with investigating these issues. From this work we already have gleaned valuable insight into the challenges faced by healthcare professionals, and also models of good practice to progress our response to the Care Coordination priority. Now that we are moving to the first stage of implementing our Action Plan, we have stood up further working groups with RDIB that will bring together the required expertise and interests to act on the priorities herein.

Along with the crucial lived experience that our Patient Voices Advisory Group has fed in, all of this work has informed the first iteration of Scotland's Action Plan for Rare Disease. As we move now to publication and implementation, RDIB and our wider governance structure will continue to support improving the lives of people living with rare diseases in Scotland.

I would like to thank all RDIB members for offering their time, expertise, and experience to our deliberations during my time as Chair. I know that RDIB will continue to take advantage of developments in genomics, care pathways and research, ensuring an approach that is flexible and future-focused.

1. Introduction

A rare disease is defined as a condition which affects less than 1 in 2,000 people. However, approximately 412,080 people out of a population of 5,151,000 in Scotland have a rare disease, and it is estimated that there are over 7,000 different rare diseases.

Rare diseases are often life-threatening or chronically debilitating, and can have a wide-ranging effect on someone's life, impacting on education, financial stability, mobility and mental health.

Rare diseases can be present from birth or develop soon after, requiring substantially more care and support for children with rare diseases, or can have a later onset. They can also be difficult to identify and can require highly specialised treatments throughout a person's lifetime.

Governments therefore recognise that we must work hard to ensure that our healthcare services offer quick and accurate diagnosis as well as coordinated care and support. The Scottish Government worked closely with the UK Government, along with the other devolved nations, to produce a new UK Rare Diseases Framework. The Framework builds on progress across the 51 commitments of the 2013[2] UK Strategy for Rare Diseases, whilst recognising new challenges and continued room for improvement.

The 2013 Strategy was a collaborative vision – between the four nations of the UK as well as between government, the NHS, research and industry. The new Framework, in the same way, forms the basis of how we will work together to improve the lives of people living with a rare disease and those who care for them. It has identified the key government priorities for rare diseases and created a vision for the future shared by all four UK nations.

This Action Plan sets out how we will implement the priorities of the Framework for the rare disease community here in Scotland. Our actions here are in response to the Scotland-specific needs identified through the Scottish Government's continued engagement with the rare disease community. The section on our approach to engagement has more information on how this was carried out.

This approach ensures that our Action Plan has the rare disease community at its heart. Throughout this Plan, we have included examples of "what matters to you": this is the first principle in supporting and promoting person-centred care, which needs to be central to all that we want to achieve.

The themes which emerged from the National Conversation on Rare Diseases have also been integral in developing both the UK Framework and Scotland's Action Plan. This survey was carried out UK-wide by the Department for Health and Social Care. Over 6,200 responses were received from people living with a rare disease, clinicians, researchers and industry. Over 400 were received specifically from Scotland.

Scotland's Rare Disease Implementation Board (RDIB) have considered this evidence base – which includes patient group representatives as well as industry, research and healthcare professionals – to identify where the Scottish Government can build on the progress made under our 2014 Implementation Plan, *It's Not Rare to Have a Rare Disease.*[3]

2 UK_Strategy_for_Rare_Diseases.pdf (publishing.service.gov.uk)

3 It's Not Rare to Have a Rare Disease - gov.scot (www.gov.scot)

Engaging with the rare disease community through our Patient Voices Advisory Group and other events has also been crucial to our work. Community engagement, and the voices of lived experience, will continue to support the development of future iterations of our Action Plan and the ways in which we implement actions. This will be key to ensuring that our policies take into account “what matters to you”, and enable people living with a rare disease to get the most out of the priorities for their care and their lives.

This Action Plan, and the further iterations that will follow, will set out the measures the Scottish Government will take to put into action the priorities of the UK Rare Diseases Framework. We will achieve this by working with partners across NHS Scotland, third sector organisations and beyond, and continue to strive to ensure that all people living with a rare disease are able to access the best possible care and support.

2. Strategic Context

UK Rare Diseases Framework

The UK Rare Diseases Framework was published on 9 January 2021.[4] Co-produced by the UK Government and devolved nations, the Framework set out the key priorities for the next five years in improving the lives of people living with rare diseases. Each nation committed to producing their own Action Plan based on how these key priorities will be implemented.

The key priorities identified in the Framework – faster diagnosis; awareness raising; care coordination and improving access to specialist care and treatment – came from a programme of engagement with the rare disease community. This included the National Conversation on Rare Diseases Survey, which ran for six weeks from October 2019 and received over six thousand responses. The findings were then discussed with the UK Rare Diseases Policy Board, the UK Rare Diseases Framework Editorial Board, and with NHS England's Rare Diseases Advisory Group. A summary of the findings can be found within the published Framework.

Further engagement with the rare disease community took place via a UK-wide round table, and clinicians, industry, regulatory and research bodies were also consulted. This programme of engagement ensured that the identified priorities were reflective of the lived experience of people with rare diseases.

Governance and Oversight Groups

Once those four priorities had been identified through engagement, and needed to be turned into action, groups were put in place that would govern and enable the development of our response to the Framework.

At the UK-wide level, these groups are the UK Rare Disease Framework Board, which oversees our response to the Framework, and the UK Rare Disease Forum, which is more focused on engagement. Representatives from all four nations have been attending these meetings to ensure that we continue to collaborate as four nations wherever appropriate, and to oversee the development of our four Action Plans. We introduced a 'deep dive' format to the Forum meetings, so that all the representatives could tackle a particular key priority in depth and hear from various spheres of expertise on what resources are available and what improvements could be made.

To oversee Scotland's specific response to the Framework, our Rare Disease Implementation Board (RDIB) was formed, bringing together those with clinical expertise, representatives of patient advocacy groups and other NHS Scotland and third sector representatives to work on the development of our Action Plan.

4 UK Rare Diseases Framework - GOV.UK (www.gov.uk)

Action Plan Structure

As mentioned in the introduction, the previous Strategy for Rare Diseases had 51 commitments in its final publication. The problem that can arise when such a list of commitments is published is that they can quickly go out of date or are not as responsive as we would like them to be to new developments.

This is why we have designed Scotland's Action Plan for Rare Disease as an iterative process, meaning that this document is by no means our final Action Plan. This publication sets out the short to medium term actions that we will take over the next 18 -24 months. After this, we will be able to take stock of our progress so far and publish the next iteration of our Action Plan. This flexibility will make us more agile in responding to the constantly changing health and social care landscape, and new developments in treatment, care and research studies. We will continue to review our Action Plan over the next five years in this way.

Implementation

As health and social care is devolved across the UK, each Action Plan will be implemented in a different healthcare system. We will work with NHS Scotland Boards, and also with NHS Scotland's National Services Division (NSD) to ensure that the actions required to improve the lives of people living with rare diseases can be embedded into standard practice across Scotland. NSD are particularly important here as they deal with national and specialist services for small patient groups, which is often the case for the care and treatment of rare diseases. The "Once for Scotland" strategy that NSD apply to their services will also help with better coordination of care.

NHS Education for Scotland (NES) are a national Board within NHS Scotland, and the education and training body for NHS Scotland. This is important when we consider the Awareness Raising priority from the Framework: where do health and social care professionals go for training and information resources, and how can we tap into these resources to increase the information available for rare diseases? We have established a working relationship with NES, who are also represented on RDIB, and we will continue to work with NES on strengthening and signposting rare disease information resources, as described in the Awareness Raising actions.

NHS Inform is Scotland's national health information service, which aims to provide the people of Scotland with accurate and relevant information to help them make informed decisions about their own health and the health of the people they care for. NHS Inform hosts information about a variety of health conditions, but until now very little regarding rarer conditions. We recognise that with over 7,000 rare diseases it is not feasible to have information about them all, but we want to have some information about rare diseases featured on NHS Inform, and we will draw on the expertise of our RDIB membership to explore what this should be.

Third sector partners will also be key to implementing the actions from our Plan. Organisations such as Genetic Alliance UK and Medics 4 Rare Diseases, as well as the Office for Rare Conditions (a University of Glasgow-led project, with some charitable funding) all have experience in developing information resources, and we will continue to draw on their expertise and consider ways in which their work can be better signposted for healthcare professionals.

Covid-19 Pandemic

We know that although the Covid-19 pandemic has affected everyone's lives, some have been more affected than others. For people living with a rare disease, the pandemic has often meant difficulties in accessing routine appointments and treatment. Many people with rare diseases have been at higher risk of severe illness from Covid-19, and shielding has led to increased isolation, as well as difficulties in accessing routine appointments and important treatment or care.

We acknowledge that the last two years have added to the challenges faced by people living with rare diseases, and we must ensure that our recovery from the pandemic includes the needs of the rare disease community. As part of our recovery from the pandemic, we are reassessing all our priorities to take a systematic, person-centred approach to planning and delivering health and social care services for the people of Scotland.

As we go through this process of realignment, we are taking time to ensure all our strategies are designed around the people using and delivering health and social care services and working together to improve health outcomes. Over time, our action plan will become more aligned with this work to allow us to get this right and ensure maximum impact for the people who need it. This will ensure that we create a stronger, interlinked Action Plan that has a real impact for people living with rare diseases.

We also want to learn as much as possible from the pandemic, such as the ways in which remote consultations and multi-disciplinary team working have been successful and can be made more commonplace. This can help with care coordination across different specialisms and NHS Board areas.

3. Our approach to engagement

During the development of this Action Plan, we have carried out more engagement than ever before, and rightly so. We have worked closely with Genetic Alliance UK to facilitate a number of virtual events to enable the rare disease community to influence and shape our policies. Our engagement during this period has been conducted in virtual spaces by necessity due to the Covid-19 pandemic. Following the publication of our Action Plan, we will consider how best to safely hold further engagement events in ways that will increase the diversity of patient voices that we can reach.

Our engagement took a "What matters to you?" approach. This approach is about listening and understanding what matters to a patient within the larger context of their life. Fundamentally it means asking not "What's the matter *with* you?", but: "What matters to you?". This model supports NHS Scotland staff to understand what's important to the people they are caring for and supporting and establishing a caring compassionate connection. When patients are fully engaged with their health care decisions and the development of pathways, it can greatly improve their outcomes. At the beginning of each of the following sections, a graphic sets out a snapshot of the feedback we have received from the rare disease community through this approach.

Engagement events with the Rare Disease Community

Our patient engagement programme in the development of this Action Plan was twofold. One strand of this was to create a formal advisory group, which became known as our Patient Voices Advisory Group. The membership was drawn from both representatives from patient advocacy groups, and from people with lived experience (and/or familial experience) of a rare disease. The group first met in December 2021. The meetings so far have been extremely helpful in identifying what matters to the rare disease community, what challenges they face and what their priorities are in their treatment and care. We recognise the difficulties and strain on individuals and small organisations in engaging on many platforms. As a result, our Voices Group hasn't perhaps had the turn out we would expect. However, we are reviewing our Patient Voices Advisory Group with Genetic Alliance UK to ensure it works and our methods for engagement do not create an unnecessary burden.

The other main strand of our engagement with the rare disease community has been through a more informal series of online events. Like the chairing of our Advisory Group, this has been led by led by Genetic Alliance UK. These events have provided an open platform for discussion and sharing of information and lived experience, as well as a chance for Scottish Government officials to share information about this Action Plan during its development. Patient feedback from these events, and from the meetings of the Advisory Group, has directly informed the structure and priorities of this Action Plan, and will continue to do so in future publications and the actions we take forward. At the beginning of each chapter of the plan, a summary of the feedback is represented in a graphic. We're keen to demonstrate the feedback we received and ensure our action plan is reflective of what we heard.

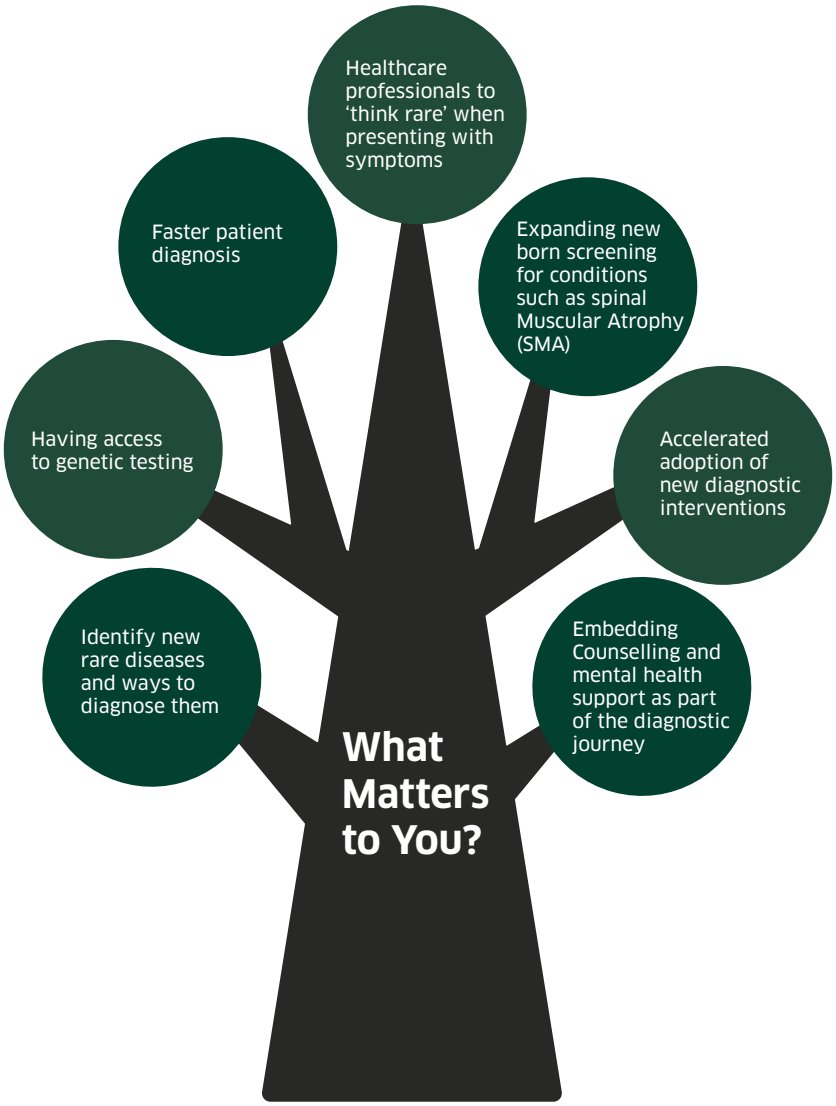
Timescales for action

The Rare Disease Framework has a lifespan of five years, and as we have learned from previous strategies, actions set out in year one can quickly expire or change due to innovation or the evolvement of other policies that replace the original action. With that in mind, this document sets out the specifics of our short term to medium term actions (2–3 years) and our ambitions for the long term (3+ years). Over the course of the next five years, we hope to have many iterations of this Action Plan building our medium- and long-term ambitions into actions.

4. Implementing the UK Rare Disease Framework

The following sections set out a number of actions that will support the delivery of the four priorities of the UK Rare Disease Framework. Each section will contain both actions specific to Scotland and actions that we will seek to accomplish in collaboration across the four UK nations and beyond.

4.1 Ensuring patients get the right diagnosis faster



Our vision is for people living with a rare disease across the UK to get a final diagnosis faster and for research into previously unrecognised conditions to identify new rare diseases and provide new diagnoses.

We know that for those living with a rare disease, getting a diagnosis is crucial in understanding how they move forward with their lives, their treatment and prognosis. Receiving a diagnosis can also open up a wealth of support from the rare disease community, as well as opportunities to take part in research to contribute to a better understanding of rare diseases. Without a diagnosis this is very difficult.

For many people, that diagnostic odyssey can take time. Time which can be taken up by multiple diagnostic interventions that can be invasive and have both a physical and mental health impact on the individual. Appointments with the wrong consultant or speciality can result in wasted time.

Ending the diagnostic odyssey with a faster and accurate diagnosis provides the person (or parents, if a child is diagnosed effectively) with the best chance of being able to seek treatment before the onset of symptoms.

The diagnostic odyssey for genetic and non-genetic conditions is equally challenging, even though advances in the use of genetic medicine have given many people the crucial answers they need. We are committed to improving the diagnostic odyssey for both genetic and non-genetic rare conditions, and over the course of this Action Plan we will seek to work with both genetic and general diagnostic specialties. We recognise that non-genetic conditions can pose greater challenges to early diagnosis. For non-genetic conditions, much of the work to reduce the diagnostic odyssey will involve awareness raising, to ensure healthcare professionals know what to look for. This is covered in greater detail in the following chapter.

We know that for many people the Covid-19 pandemic has extended that diagnostic odyssey further. Our work to recover our NHS allows us to consider how we can deliver for patients in different and better ways, and throughout the lifespan of the Framework we will work with the rare disease community to ensure learning from the pandemic shapes our future.

The following sections set out our initial actions on how we will support this priority. These actions will evolve over time, and we will add further actions as we evaluate and update our Action Plan in future iterations.

Action 1: Implementation of Genome UK

Back in September 2020, the Scottish Government signalled support for Genome UK (published that month).[5] This strategy sets out how the genomics community will work together to harness the latest advances in genetic and genomic science, research, and technology for the benefit of patients. This community includes patients, families, charities and officials across the four nations, NHS bodies, academia, and industry amongst others. With all four nations signalling support for the strategy, this provides a blueprint for expanding our genomics offering across the UK.[6]

Since the publication of Genome UK, the four nations have worked closely to develop a series of shared commitments: Genome UK: Shared Commitments for UK-wide implementation 2022 to 2025 (March 2022). This publication signals a commitment to work together with our delivery partners across the UK in order to realise the potential of genomic technologies for the benefit of patients at home and abroad, including supporting people living with a rare disease.

In Scotland specifically, we have recently established the Scottish Strategic Network for Genomic Medicine, as a 'front door' for engagement and strategy development. The Network will advise and make recommendations on genetic testing availability as well as supporting the planning for future capacity into areas such as Whole Genome Sequencing or expanding our Whole Exome Sequencing services, which we recognise are of huge importance to the rare disease community. The Network will have a crucial horizon-scanning function which will work with the Scottish Medicines Consortium, Scottish Health Technology Group, researchers, academia and our Innovation Pipeline to ensure people with a rare disease can access new genomic technologies and testing.

5 Genome UK: the future of healthcare - GOV.UK (www.gov.uk)

6 Genome UK: shared commitments for UK-wide implementation 2022 to 2025 - GOV.UK (www.gov.uk)

Soon after the publication of this Action Plan, we will publish our first ever genomics publication, which will set out our intention for developing Scotland's genomics medicine services over the next five years. This will be following by Scotland's first ever genomics strategy in the course of 2023, which will build on work to date, including the Bridge to a Scottish Genomics Strategy, which focused on rare disease genomics, but also explore how we further propel our genomics offering, benefitting those who need it most.

Most importantly, our strategy will have a real focus on rare and inherited conditions with the support of the appointed Network Clinical Lead for Rare Disease, Dr Jonathan Berg.

The Scottish Government will support this strategy through significant investment, with £5 million committed for 2022/23 alone, previous investment of £8 million since 2017. This is in addition to almost £20 million of funding allocated to the four genetic laboratories annually in Scotland by NHS Boards through NHS National Services Division (NSD) commissioning arrangements.

We also recognise that we must have capacity within our genetic laboratories to deliver an equitable service for rare disease patients. For this reason, we will be implementing the recommendations from the Major Service Review of Scottish Genetic Laboratories. By acting on these recommendations, we will ensure that our genetic medicine service is fit for the future and can flex to meet increasing demand and advances in the field to benefit a range of conditions, covering both cancer and rare diseases.

Action 1: What does this mean for people living with a rare disease?

The needs of those with a rare disease will be a key consideration on how we expand our genomic medicine services in Scotland. The Rare Disease Implementation Board for Scotland will work closely with the new structures, becoming advocates for the rare disease community. In addition, we will ensure relevant rare disease clinicians and patients' representatives can feed directly into our implementation plan by being part of the new structures. The rare disease clinical lead for the Genomics Network will be key to this.

Action 2: Newborn Screening

Screening has a vital role in allowing early diagnosis of some rare diseases and the initiation of early treatment to reduce complications. The Scottish Government is represented on the UK National Screening Committee (UK NSC) which makes its recommendations to all four Health Departments across the UK.

The UK NSC advises Ministers and the Health Service in all four UK nations on all aspects of screening. Using research evidence, pilot programmes and economic evaluation, the Committee assesses the evidence for national screening programmes against a set of internationally recognised criteria, taking a range of distinct factors into account. At present, nine rare conditions are screened in new-born babies through the NHS Newborn Blood Spot Screening Programme.

The UK NSC has recently undergone significant changes to both its remit and membership and will now consider recommendations for targeted as well as national screening programmes. For our year one action, we commit to continuing to participate in the UK NSC; to following guidance to ensure appropriate use of screening tools in line with UK National Screening Committee recommendation, and to work with the Committee to embed its new remit.

Through the Rare Disease Implementation Group, we will engage in any considerations on new-born screening, ensuring the voice of those with a rare disease are part of any future screening considerations.

We will also continue to engage across the four nations on any new UK-wide screening research pilots. Furthermore, we will consider Scotland's participation in the UK National Newborn Screening Pilot which aims to carefully evaluate the benefits and risks of implementing new-born genomic screening to accelerate diagnosis and enable earlier access to treatments for rare genetic conditions.

In addition to this, through our engagement with the rare disease community, we realise the need to improve the understanding of screening policies particularly for rare diseases.

Over the next 12 months, we will work with the rare disease community to improve the understanding of screening decision-making processes. We will look to embed this information into existing materials, ensuring it is readily available and understandable by those impacted by such policies.

Action 2: What does this mean for people living with a rare disease?

Those living with a rare disease will have access to key information on screening in Scotland. In particular, we will equip the rare disease community with information on how screening policies are agreed and the processes on expanding screening programmes. Information will be made available to those living with a rare disease, so that so they understand how screening impacts on them and their families.

Action 3: Expanding the functionality of the Congenital Conditions and Rare Diseases Registration and Information Service for Scotland (CARDRISS).

Congenital conditions are important in their own right, and also comprise a key subgroup of all rare diseases. Initial funding to establish CARDRISS was provided by the Scottish Government to NHS NSS Information Services Division (ISD) and latterly to Public Health Scotland (PHS), the current home of CARDRISS, over the three-year period October 2018 to March 2022.

In the first instance, CARDRISS will register babies affected by a major structural or chromosomal anomaly or recognised syndrome. This is in line with the standards recommended by the European Registry of Congenital Anomalies and Twins (EUROCAT), a European network of congenital anomalies registers. Live-born babies diagnosed within the first year of life; spontaneous stillbirths occurring at ≥24 weeks gestation; spontaneous late foetal losses occurring at 20–23 weeks gestation, and pregnancies terminated at any gestation due to an included anomaly will all be registered.

In due course, when registration of major anomalies is securely established, the plan will then be to widen the remit of CARDRISS to include registration of other rare diseases. This will help inform the planning of services for individuals and families affected by congenital anomalies and rare diseases. It will also allow NHS Scotland to support the prevention of anomalies where possible, understand the impact of antenatal screening and support research into these conditions.

At the beginning of the project the plan had been that CARDRISS would prospectively register affected pregnancies, ending in 2021 onwards. Work on the CARDRISS IT system then took longer than expected, due to the Covid-19 pandemic. The registration of affected pregnancies was therefore delayed until the summer of 2022.

In recognition of the quality of the linked database, Scotland has been accepted as an associate member of the European network of population-based registries for the epidemiological surveillance of congenital anomalies (EUROCAT). A data-sharing agreement with EUROCAT is in place, and aggregate data covering pregnancies ending 2005-2019 will be transferred to the central European database on congenital conditions for publication on the EUROCAT website, inclusion in country level-monitoring of trends and clusters of conditions, and in international studies.[7]

Over the medium term, we will work with PHS to build on the success of CARDRISS and explore how the functionality of the CARDRISS IT system can further support data capture for rare diseases. This will include exploring options for establishing new national data returns. New national data returns would also bring wider benefits, in particular supporting monitoring of the pregnancy and new-born screening programmes. PHS will also explore how the registration of babies can be extended to focus on rare diseases covered by the pregnancy and new-born screening programme. Specifically, this could include the inherited metabolic disorders (IMDs) currently covered by the new-born screening programme.

We will work with PHS to understand the options to expand CARDRISS. Extensions are likely to require financial investment by the Scottish Government, which we will consider as part of our future budgetary considerations.

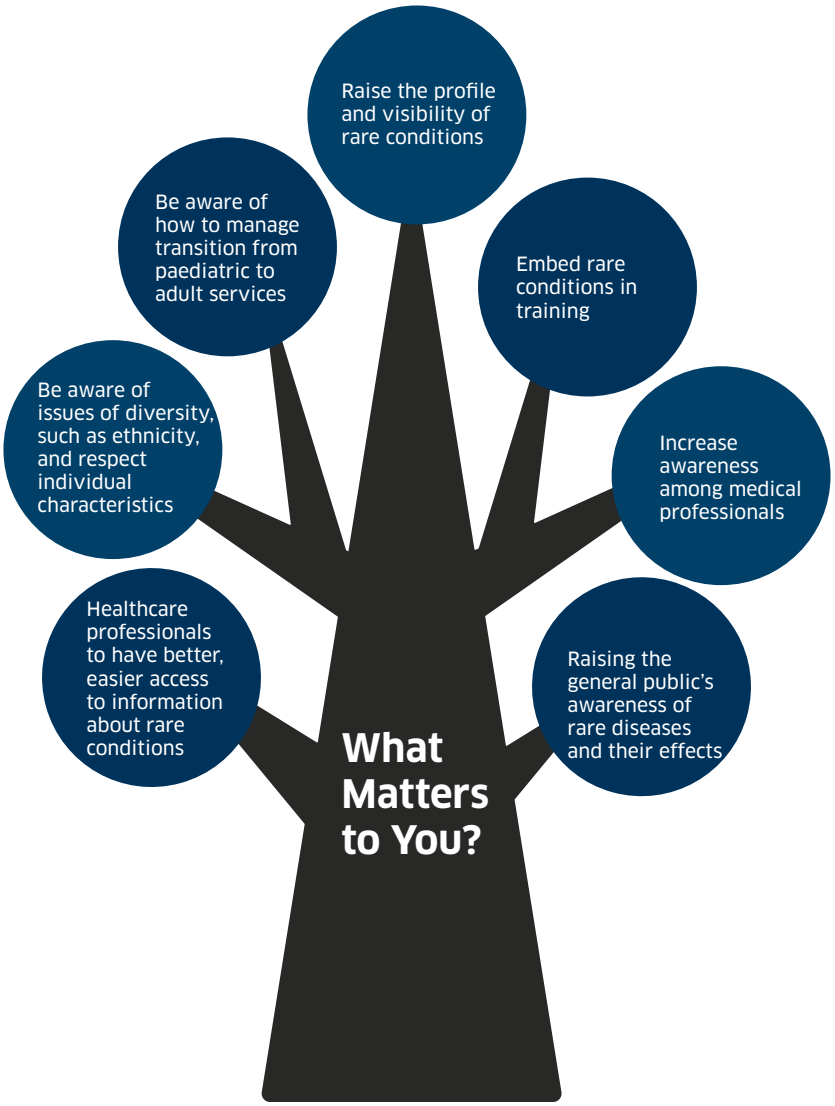
This action is linked to action 18.

7 Prevalence charts and tables | EU RD Platform (europa.eu)

Action 3: What does this mean for people living with a rare disease?

Having access to better data on rare disease can help clinicians make better informed decision on care, prevent disease and also allow better access to research and clinical trials. While those living with a rare disease won't necessarily have access to CARDRISS, they will see benefit through their care as well as support for future diagnoses.

4.2 Increasing awareness of rare diseases among healthcare professionals



Our vision is for healthcare professionals to have an increased awareness of rare diseases and use of genomic testing and digital tools to support quicker diagnosis and better patient care.

Increasing awareness of rare disease amongst health care professionals is key to the success of the Framework, and of our Action Plan. Healthcare professionals who have a foundational awareness of rare diseases will support better diagnosis, coordination of care and access to specialist treatment and drugs.

As there are over 7,000 rare diseases it is not possible for healthcare professionals to receive comprehensive training on every condition. It is therefore important that they are aware of rare diseases more broadly and are alert to considering them.

While awareness is important, even more so is the embedding of more formal education about rare diseases during training. This means including rare disease education as part of the curriculum for those progressing through medical school or other educational route leading to a position as a healthcare professional. It is also important to ensure that that there can be Continuing Professional Development (CPD) linked to rare disease available throughout the career of a healthcare professional.

We will treat raising awareness as the highest of our priorities.

This section sets out our initial actions to support this priority. Beyond these actions, throughout the life span of the Framework we will seek constant opportunities to inform and present to health care professionals, policy makers and wider society to improve rare disease awareness more generally. We will do this in partnership with third sector organisations like Genetic Alliance UK, and the University of Glasgow's Office for Rare Conditions.

Action 4: Improving information about Rare Disease on NHS Scotland platforms

The importance of having access to accurate and timely information has been clearly seen and highly valued by us all in the last two years. A key platform for hosting NHS information in Scotland is NHS Inform.

NHS Inform has been at the forefront of providing crucial information to people across Scotland throughout the Covid-19 pandemic. It has become the first point of call for many seeking advice and information relating to health services.

Over the next 12–18 months we will work with NHS Inform to improve information about rare diseases online. We recognise that due to the volume of rare diseases we will be unable to have bespoke information for each rare disease. However, as part of their patient journey, we want to ensure people have access to relevant information and are signposted to places for support while living with a rare disease or waiting to receive a diagnosis of their condition.

In doing so, we will work with third sector organisations to develop supporting materials which for hosting via NHS Inform. We will look to use existing work such as the Genetic Alliance UK Rare Resources for Scotland Toolkit, but we will also work with our Patient Voices Advisory Group to further understand what information they would expect to see on a national platform about rare diseases.

Furthermore, we will seek further opportunities to improve the information about rare diseases on NHS Scotland digital platforms, including those used by healthcare professionals themselves.

Action 4: What does this mean for people living with a rare disease?

Those newly diagnosed with a rare disease will be able to seek initial information on NHS Inform, and signposting towards further resources. This could be the difference between a person dealing with their diagnosis alone or accessing support groups and information which we know are crucial to individuals. The information may also highlight additional support spaces that those already living with a rare disease may not have been aware of.

Action 5: Optimising Rare Disease Day

Rare Disease Day is an internationally recognised day. On the last day of February every year the rare disease community comes together to raise awareness of rare diseases and their impact.

The day is an opportunity to truly maximise our awareness-raising powers for rare diseases. In the past, the Scottish Government has supported Rare Disease Day through Parliamentary events, lighting up buildings in homage to the day as well as publishing an open letter to the rare disease community.

Rare disease day provides an important platform to raise awareness of rare diseases, and we will seek to optimise this platform to support the delivery of this priority. We will work with stakeholders to develop a suite of events which engage a broad range of people including clinicians, senior leadership in NHS Boards and those working within primary care.

We recognise that raising awareness is about reaching those who aren't already engaged or have not yet gained some awareness of rare diseases. Those are who we will target through this action.

Action 5: What does this mean for people living with a rare disease?

Using the internationally recognised Rare Disease Day can can provide more opportunities provide more opportunities to raise awareness of rare disease. We want to ensure we take full advantage of this so that raising awareness continues to move forward.

Action 6: Working with NHS Education for Scotland

NHS Education for Scotland (NES) is the national health board with statutory responsibilities to effect sustainable change through workforce development, education and training across the health and social care system in Scotland, while working at UK level with partner organisations.

NES's purpose is to drive change and improve the quality of care experienced by citizens across Scotland by ensuring that the health and social care system has the right staff, with the right skills, in the right place, at the right time. NES is integral to improving outcomes for people and in ensuring a skilled and capable workforce underpins the design and delivery of services.

NES will be a key partner in our efforts to increase awareness of rare diseases among healthcare professionals and over the course of this plan we will establish partnership working to enable this.

Over the next 12 months, we will work with NES to understand the existing rare disease training and education material already developed by a range of organisations like those developed by Genetic Alliance UK, Medics 4 Rare Disease and in other nations. We will consider where these training and education materials can be hosted to ensure optimal recognition and pick up across Health Care Professionals.

One key platform is Turas. Turas is a modern and accessible digital platform developed by NHS Education for Scotland to support health and care professionals working in the public sector. We will work with NES and in partnership with these third sector organisations to understand how we utilise national platforms like Turas to continually raise awareness of rare diseases.

We know the importance of linking training with Continuing Professional Development (CPD) points. Linking rare disease training with CPD points will ensure maximum uptake by health care professionals, and over the medium term (2–3 years) we will explore the appetite for this and how to enable it.

Over the longer term (3–5 years), we will seek to work with NES and a range of other educational partners to consider how we embed basic training on rare diseases into the curriculum for undergraduate and post-graduate students. This will be challenging, but we recognise that to significantly improve awareness of rare diseases it is imperative that we do so as early as possible in the education pathway for health care professionals, particularly in nursing and GP training. Exploring where links can be established in existing curricula will be a useful starting point in such considerations.

Action 6: What does this mean for people living with a rare disease?

Healthcare professionals having access to key information on rare disease will support the full patient journey. Importantly, building in rare disease training and education to the curriculum for health care professionals will ensure that those who work on our frontline services can do so with a basic understanding and awareness of rare disease.

Action 7: Enabling opportunities for third sector organisations to raise awareness of rare diseases

We recognise the crucial role that organisations like Genetic Alliance UK, the Office for Rare Conditions and Medics 4 Rare Diseases play in increasing awareness of rare diseases. Through partnership working, we will seek to work with rare disease organisations to enable opportunities for awareness raising. We will facilitate introductions and connections wherever possible between these organisations and their valuable expertise and NHS Scotland bodies like NES and NHS Inform to ensure that their work can reach the widest audience.

We will seek to utilise existing platforms such as NHS Scotland events and conferences as well as facilitating opportunities to bring rare disease awareness raising to the door of primary care, hospitals and community settings.

We recognise that the power in awareness raising lies in amplifying the voices of those most affected. We will therefore ensure that any opportunities to engage with health care professionals include the voices of the rare disease community. Sharing the lived experience of rare diseases with those working with patients on a daily basis will have the most powerful impact.

Through working with charities and other third sector organisations we will look to better understand the needs of healthcare professionals when it comes to rare disease. It is important that we are providing materials and information which are of use to healthcare professionals.

We will also look to work with Medics 4 Rare Diseases in a useful piece of work to assess the current levels of rare disease awareness and understanding among medical students. From the results of this survey, we will be able to see where there are gaps and what further action we should be taking to address them.

Action 7: What does this mean for people living with a rare disease?

We recognise we can't deliver this plan alone. Working with organisations who have direct links to the rare disease community will be crucial to the success of this plan. By doing so, we will be able to strengthen our engagement and make more progress which will ultimately have a positive impact for those with a rare disease.

Action 8: Understanding and acting on the information needs of Health Care Professionals to raise awareness of rare diseases

To further our understanding of what healthcare professionals need to improve their awareness of rare diseases, we will work with Genetic Alliance UK and the Office for Rare Conditions.

Genetic Alliance UK's research team will develop a survey to understand healthcare professional's views of rare conditions, assess level of awareness and to identify gaps in information needs and how to address them. This will be distributed through Genetic Alliance UK's communications team, Office for Rare Conditions communications team and wider NHS Scotland networks. This will inform the development and distribution of information materials.

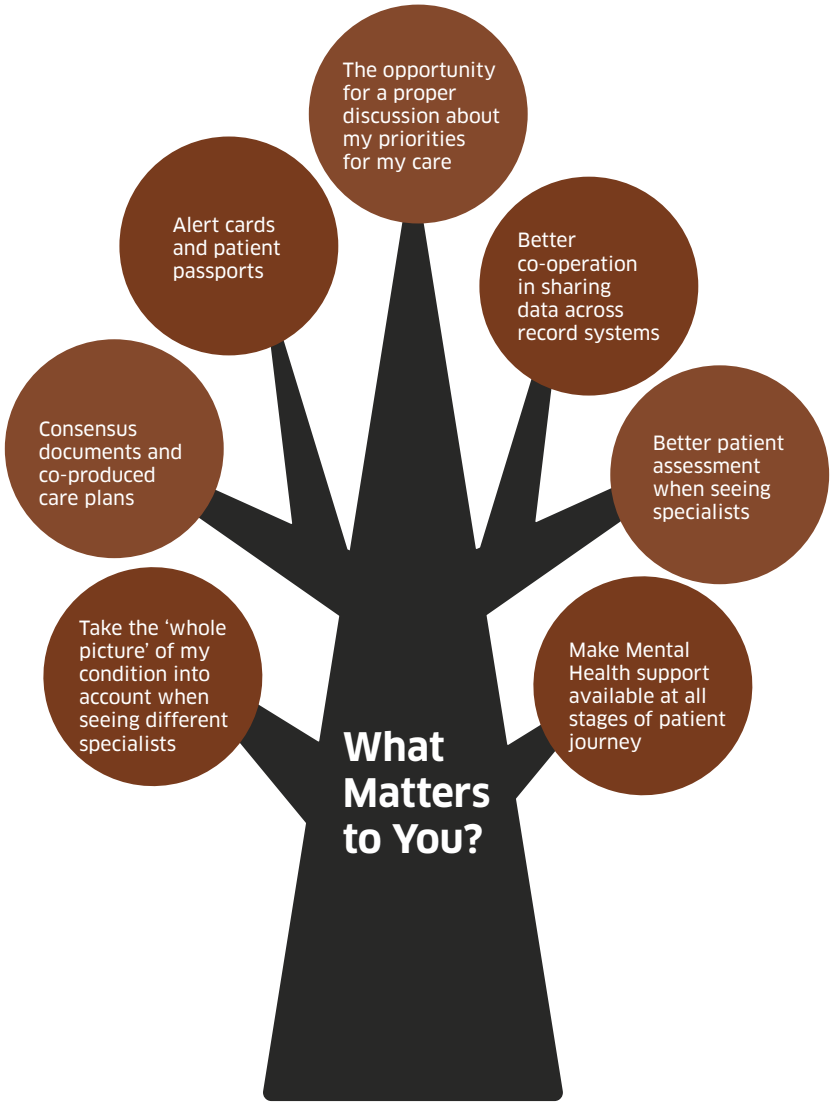
Following the results of the survey Genetic Alliance UK and the Office for Rare Conditions will re-develop the Rare Resources Professionals Guide, which focuses on signposting healthcare professionals to reliable sources of information, to ensure it meets the needs of healthcare professionals in Scotland. We will also develop promotional materials such as posters and business cards for distribution in appropriate healthcare settings.

Furthermore, we will work with Genetic Alliance UK and the Office for Rare Conditions to support the development of a programme of events and activities to coincide with Rare Disease Day 2023 to improve awareness of rare conditions with NHS Scotland staff.

Action 8: What does this means for people living with a rare disease?

Having more tailored resources available to medical students and healthcare professionals will help to raise the profile of rare diseases and empower them to 'think rare'. Getting the awareness raising priority right means that all the others will follow – diagnostic odysseys will be reduced, and people living with rare diseases will have better and quicker access to the treatment and care that care that they require.

4.3 Better coordination of care



Our vision is for people living with a rare disease to experience better coordination of care throughout the patient journey.

With so many rare diseases, very few have dedicated services – and even those that do can have poorly-coordinated care. The chronic and varied nature of rare diseases can lead to those living with a rare condition requiring input from a range of specialties across health and social care. This means many appointments or diagnostic interventions across many specialities at various times and/or places.

In many cases responsibility for coordinating appointments and services falls to individuals affected by the condition or a family member or carer, which can result in a significant care burden.

We recognise that care coordination should be flexible, keeping the person at the heart of decision making. It is important to recognise that our NHS is a health service for everyone, and it would be ineffective to have dedicated, discrete services for every patient cohort, particularly given the breadth of rare diseases. This doesn't mean we can't deliver high-quality person-centred care, with care coordination a key part of how care is delivered.

We want to embed the needs of those living with rare diseases and their carers to have the support they need to navigate the healthcare system and should be empowered to do so if they wish.

Improving coordination of care is essential to ensure care is effectively managed, the burden on patients and carers is minimised, and that healthcare professionals are working together to provide the right care at the right time.

Action 9: Consider a future Care Coordination Service

Identifying a suitable coordination of care model for use in Scotland is a process that we have already started to consider.

We also began from the recommendation in the Framework that coordination of care should be improved, and also had recent reports to consider:

- The Scottish Parliament Cross Party Group on Rare, Genetic and Undiagnosed Conditions, via Genetic Alliance UK (the CPG secretariat) published the report *Improving Care for Rare Conditions in Scotland* in March 2021. The report recommended that a short-life group should be established to explore a coordination of care model for Scotland, and that the Scottish Government should commit to a pilot project for a care coordination service.[8]
- The *COordiNated Care of Rare Diseases* (CONCORD) study began in 2018 and was published in March 2022.[9] This UK-wide study found that poorly coordinated care can be detrimental to patients and families, and suggested classifications and flowcharts for effective care coordination.

We have also heard from our programme of patient engagement that people living with rare diseases often end up coordinating their own care (or their child's care), as there can be so many specialists and appointments to deal with and organisational coordination is lacking. This takes up lots of time, money and energy and can become a full-time role.

We stood up a Care Coordination Working Group in 2021 as part of our Rare Disease Implementation Board. The group began by looking at models of good practice and considering how one of these might be adopted or adapted for use in Scotland.

This information-gathering exercise provided useful insights into care coordination models that have been successfully implemented. One such model is the East of Scotland Clinical Genetic Service that has been developed in NHS Tayside.

8 CPG Report: Improving Care for Rare Conditions in Scotland | Genetic Alliance UK

9 Co-ordinated care for people affected by rare diseases: the CONCORD mixed-methods study (nihr.ac.uk)

The East of Scotland Clinical Genetic Service employs two full-time genetic nurses. They provide a specialist service, the aim of which is to ensure that patients with rare diseases are supported on a long-term basis and that they are provided with the best information about their condition and its management. They both also act as a point of contact for patients and family members.

The Band 7 genetic specialist nurse role is aimed at those with complex (and sometimes multisystem) disorders such as the muscular dystrophies, ataxias and neurofibromatosis providing nurse-led clinics which are likened to a yearly 'MOT'. Each patient and other involved professionals (health and social care) will receive either a care plan or clinic summary after each encounter.

Care coordination is a vital part of this role for those who need regular screening investigations but don't necessarily require specialist clinical input (this may include cardiac, respiratory or blood tests). This role also provides support during the transition journey from child to adult services to ensure the process is as smooth and supportive as possible. Another essential part of this role is multidisciplinary working involving preparing, facilitating and disseminating information for multidisciplinary team meetings and ensuring patients are always provided with feedback about their condition and their management plan.

The Band 6 genetic nurse coordinates care for those patients with either a genetic mutation (Lynch Syndrome, Familial Adenomatous Polyposis) or family history giving them an increased risk of bowel cancer. This involves organising screening and offering appointments at nurse led clinic to provide support, information, and lifestyle advice. It also involves working with other departments to ensure timely screening for the highest risk patients and to discuss results with Pathology, Endoscopist or Gastroenterology MDT when necessary.

Other patients seen at the nurse-led clinic include those with a moderate risk family history of bowel or breast cancer for risk assessment and referral for screening, alpha-1 antitrypsin deficiency, haemochromatosis and a follow up appointment for patients recently found to have Lynch Syndrome to give them the opportunity to clarify information and ask any additional questions. The nurse is also a member of the Hereditary Haemorrhagic Telangectasia (HHT) multi-disciplinary team (clinical genetics and ENT), coordinating clinics, maintaining the database, being a point of contact for patients and assessing patients pre-clinic regarding quality of life and epistaxis to determine specific issues to be addressed.

The genetic nurse role involves co-ordinating care for people with a wide range of conditions and is significantly different from that of other clinical nurse specialists whose role is usually single condition specific. The service provided by the two genetic nurses is unique in Scotland, but the model of care could provide the basis for the development of a Scottish wide care co-ordination service to meet the needs of people with rare diseases.

We will use the information gathered, such as the example above, as a starting point to consider a future care coordination service for people living with rare diseases in Scotland. This will include looking at the role of genetic nurses in supporting coordination of care and considering whether we can develop a model for Scotland.

Getting the right model for Scotland will take time and is likely to need investment, but this is worth implementing in the right way so that those who need coordinated care can benefit from joined-up expertise. We will continue to prioritise this area and take advantage of the expertise available in our stakeholder networks.

Action 9: What does this mean for people living with a rare disease?

Having a suitable model of care coordination implemented in Scotland will mean that people living with rare diseases will have fewer wasted appointments, benefit from the expertise of multi-disciplinary care, experience shorter diagnostic odysseys, and ultimately benefit from care that is better tailored to their needs.

Action 10: Improving the use of Anticipatory Care Plans

Anticipatory Care Planning is a person-centred, pro-active approach to care that involves conversations between individuals, their families, carers and health and social care professionals. It helps people communicate goals and preferences for their care.

Anticipatory Care Plans, or ACPs, are the way these conversations are recorded. They can be updated as required, for example if a person's condition changes, or if they develop additional needs. ACPs are sometimes associated with end-of-life care, but in this context, they are about living with a long-term condition and communicating what is important to the person who lives with that condition.

Information about Anticipatory Care Planning and ACPs is hosted on Healthcare Improvement Scotland's "iHub" website.[10] There is also information on Anticipatory Care Planning being used in the context of Covid-19, or for neurological conditions.[11, 12]

ACPs can contain valuable information about the complexities of living with a rare disease. For example, if emergency care were required, and a person's condition meant that they had specific needs, allergies or could not be intubated, an ACP can make this clear to the responding medics.

We know that there are useful information resources relating to Anticipatory Care Planning and the use of ACPs, such as the aforementioned pages hosted by Healthcare Improvement Scotland. We want to encourage take-up of ACPs among healthcare professionals and signpost this information to clinicians and teams who care for people living with rare diseases.

Over the next 12–18 months, we will work with Healthcare Improvement Scotland and third sector organisations on ACP information and resources that relate to rare diseases, along the same lines as those for neurological conditions. We will look for opportunities to promote the use of Anticipatory Care Planning when we are considering the training and education aspects of our Action Plan. We will then publish updates on progress in this area.

10 Anticipatory Care Planning Toolkit - ihub | Health and social care improvement in Scotland - Anticipatory Care Planning oolkit

11 Anticipatory Care Planning in the context of COVID-19 - Anticipatory Care Planning Toolkit - ihub - Anticipatory Care Planning n the context of COVID-19

12 ACP and neurological conditions - ihub | Health and social care improvement in Scotland - Anticipatory Care Planning and Neurological Conditions

Action 10: What does this mean for people living with a rare disease?

The use of Anticipatory Care Planning by people living with a rare disease or condition can lead to better coordination of care. When "what matters to you" has been discussed and clearly noted on an ACP, it can be used by all the healthcare professionals involved in a person's care and treatment. The goals and wishes identified by the person living with the rare condition can be considered by multi-disciplinary teams when they are planning the right levels of care for that person.

Action 11: Digital Passports

Patient passports have been around for some time, and were in use in Scotland in the 1990s, first in paper form and then as digital documents, this development supported by the Scottish Government.[13] A digital patient passport is a simple to use document in the style of an e-book, which can hold information about a person's needs, priorities for their care and their life and family.

Similar to the way ACPs can be used, patient passports enable a person living with a long-term condition to record what matters to them. The ACP can then be shared among multiple healthcare professionals and specialisms to ensure they all have access to the same information. This is another way to improve care coordination. To explore how this could benefit people living with a rare disease, we will work with third sector organisations who have used patient passports to understand how they could be better promoted for use by people living with rare diseases, their families and healthcare professionals.

PAMIS Digital Passports

The PAMIS passport is a simple, easy to use, flick-through e-book that can be created and displayed on tablet devices, computers and phones. Each PAMIS passport contains information about one person and uses video, photography, sound and text to help that person express their needs. The passport, uniquely, is owned by the individual and is shared with those they choose to share it with. The initiation and development of the passport might come from family carers, paid carers, practitioners, and individuals themselves. The development process has often been used as a way of building a truly person led approach with people who need extra support. The passports are freely available to anyone who needs them.

Action 11: What does this mean for people living with a rare disease?

The wider availability and adoption of care passports will provide another way for people living with a rare disease to inform healthcare professionals of their priorities for their care. If different healthcare specialities all have access to this same information, patients won't have to spend as much time explaining the same things at appointments. More appointment time is then freed up for discussing current needs and priorities.

13 PAMIS Digital Passports | PAMIS

Action 12: Supporting the Implementation of: It's OK to Ask

In 2021, NHS Scotland launched the public campaign "It's OK to Ask".[14] Developed and piloted by NHS24, and support by the Scottish Government's Realistic Medicine Value Improvement Fund, the campaign encourages people to ask healthcare professionals four key questions:[15]

- What are the benefits of my treatment?
- What are the risks of my treatment?
- Are there any alternative treatments I can try?
- What if I do nothing?

The campaign states that knowing the right questions to ask at healthcare appointments can make all the difference. This enables people to make the right decisions about their treatment and care. The webpage (linked at footnote 16) includes a downloadable patient leaflet that can be taken to appointments.

We would like to encourage the take-up of "It's OK to Ask" as widely as possible. Wider adoption of "It's OK to Ask" would also assist healthcare professionals in knowing a person's wishes and priorities, leading to more productive discussions at appointments. Transitions from paediatric to adult care can also benefit from this approach.

Action 12: What does this mean for people living with a rare disease?

We know that people living with rare diseases, or the parents of children with rare diseases, will already come to appointments with a high level of knowledge about their condition (once it has been diagnosed). This could still be a helpful method of focusing appointment time so that people living with a rare disease are able to get the most out of every appointment and have more control over their priorities for their care.

14 It's OK to Ask | NHS inform

15 It's OK to Ask – Public messaging campaign from NHS Scotland – Realistic Medicine

Action 13: Mental Health Strategy

Our programme of rare disease community engagement has highlighted that people living with rare diseases place a high priority on mental health: improving resources, more tailored support and generally reducing the burden that living with a rare disease can have on their mental health. Mental health services can be required at any stage of the patient journey, from the diagnostic odyssey and the processing of a diagnosis, to ongoing support when accessing care and treatment.

The Scottish Government's Mental Health Strategy 2017–2027 was published on 30 March 2017.[16] The Strategy set out actions to be achieved with the guiding ambition that we must prevent and treat mental health problems with the same commitment, passion and drive as we do physical health problems.

The Scottish Government committed to reviewing and refreshing the Mental Health Strategy in 2022. As part of our considerations for a future strategy, we will consider the mental health needs of all patient groups, including people living with rare diseases. Throughout the development we will work to ensure that we reflect the voice of those living with a rare disease in the refreshed strategy.

Rareminds

Rareminds are a non-profit company who provide online counselling and wellbeing services for rare disease charities.[17] We spoke to Rareminds earlier this year to find out more about the support they provide, and what mental health challenges people living with rare diseases often face. They gave us some helpful pointers, such as the need for more tailored mental health support for people with rare diseases at all stages of their diagnostic odyssey.

We will keep engaging with Rareminds as we progress our actions in this area and look for ways to signpost their support where appropriate.

We will also look to work with Genetic Alliance UK and other organisations to consider developing downloadable Mental Health resources for those living with a rare condition.

Action 13: What does this mean for people living with a rare disease?

Considering the needs of people living with rare diseases when developing mental health policy and resources means that tailored resources can be developed. Promoting these more widely will mean that people's mental health needs can be integrated into their care.

16 Mental Health Strategy 2017-2027 - gov.scot (www.gov.scot)

17 About Us | Rareminds

Action 14: Continuing to Promote and Embed Person Centred Care

Practical improvements to person-centered care are promoted and supported through a framework of five key "Must Do With Me" principles:

- 1. "What matters to you?" Your personal goals and the things that are important to you have been discussed and form the basis of your care or treatment.
- 2. "Who matters to you?" We have asked you about the people that matter most in your life and we have given you the opportunity to involve them in the way that you choose.
- 3. "What information do you need?" We have provided you with understandable full information and supported you to make decisions that take account of your personal goals and the things that are important to you.
- 4. "Nothing about me without me." You will always be given the opportunity to be involved in discussions. All information exchanges and communication between professionals or between different services or supports are transparent and always provide you with the opportunity either to be present or to contribute to the process.
- 5. "Personalised contact." As much as possible, the timing and methods by which you contact and use services or supports are flexible and can be adapted to your personal needs.

Together, these five "Must Do With Me" areas will help to ensure that all the interactions between people using services and the staff delivering them are characterized by listening, dignity, compassion, and respect.

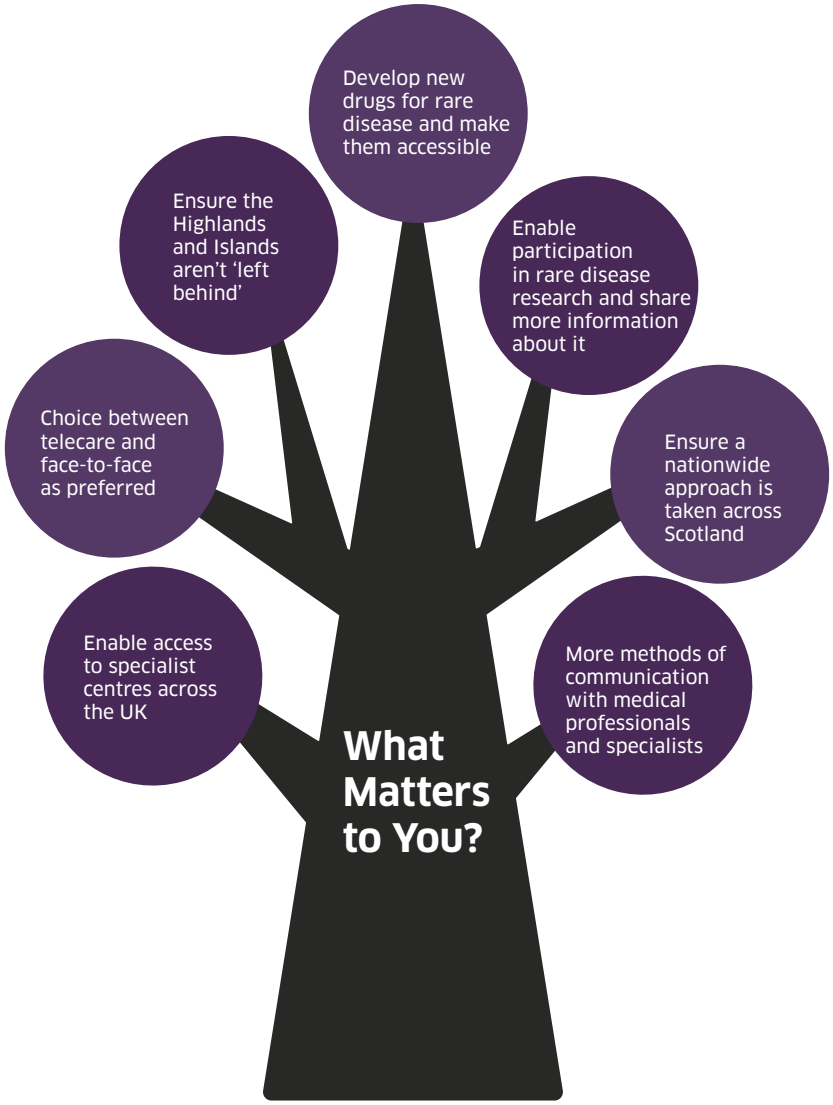
We will continue throughout the lifespan of this person-centred framework to promote and embed person centred care ensuring those with lived experience are at the heart of the services we are delivering.

We will continue to take a "what matters to you" approach to our engagement. We recognise the need to continue our engagement with the rare disease community from development through to implementation of our Action Plan.

Action 14: What does this mean for people living with a rare disease?

These five principles, once embedded in regular practice at appointments, will help people living with rare diseases to get the most out of every consultation, and have their wishes and priorities at the centre of the conversation.

4.4 Improving access to specialist care, treatment and drugs



Our vision is for people living with a rare disease to have improved access to specialist care, treatments and drugs.

Ensuring patients have access to the best possible high quality specialist care when they require it remains a key commitment for the Scottish Government.

Evidence from the UK National Conversation on Rare Diseases showed that those receiving treatment for their rare disease were reporting an increase in the quality of care provided compared to those that were not accessing specialist care. Many rare diseases do not have established treatments, making it difficult to establish specialist services. We recognise though that where they do exist, specialist services can be lifesaving and life-changing, significantly improving prognoses and quality of life.

Providing access to safe, high-quality specialist care and treatments can present challenges, with some patients needing to travel significant distances to access specialist centres. The small numbers of patients affected by individual rare conditions mean that the scale of clinical trials typically used to assess the safety and efficacy of medicines may not be possible. Assessment of, and access to, rare disease medicines can also require additional consideration from health technology assessment bodies, due to limited and uncertain data.

It is therefore imperative that we ensure we have the ability to commission and access these services, treatments and drugs for those who will benefit the most.

Action 15: Recognising the role of specialist services

Specialist services and national networks are commissioned through National Commissioning by NHS National Services Scotland on behalf of NHS Boards and the Scottish Government, to provide quality assurance for a range of specialist interventions for people living with rare diseases.

These national commissions help ensure patients across Scotland have the best possible access to high quality specialist care. NSS brings together a diverse team of programme management and commissioning experts including clinical and public health, all working within the governance of NHS Scotland. This team works working with a service provider to set a service specification, agree the activity profile, staffing, and resources required to commission a service that demonstrates positive clinical outcomes and patient experience.

These commissioned services are critical to coordination of care. NHS National Services Scotland will continue to support the development of services for rare diseases where possible, and engage with the clinical and rare disease community to ensure they are fully involved in service planning.

We recognise too that there are a wide variety of non-commissioned specialist services across Scotland, covering a range of rare conditions. These clinics or specialists are not always evident, and we will work to understand the landscape of care pathways across health and social care in Scotland, by initially focusing on an agreed set of rare conditions.

We will seek to understand the availability of clinics for these non-commissioned conditions and increase the awareness of these specialist services to ensure patients have appropriate access. It is important to highlight that referral outwith a patient's host NHS Board is at the discretion of the host and receiving Boards.

Action 15: What does this mean for people living with a rare disease?

If we ensure that we consider the needs of people living with rare diseases when planning future services, people who need these services will have a clear pathway for referral and treatment when clinically appropriate. This will improve access to the right care at the right time.

Action 16: Ultra Orphan Pathway

The Scottish Government recognises that for people living with ultra-orphan diseases (those affecting fewer than 1 in 50,000 people), there tends to be no or a very limited number of medicines available to treat their specific condition. Ultra-orphan medicines often face difficulties being approved through traditional health technology assessments, due to the low number of patients available to take part in clinical trials, and thus there can be higher uncertainty about their efficacy and cost effectiveness. The 2016 Montgomery Review of Access to New Medicines highlighted that there was a need to accelerate access and provide advice on medicines for ultra-orphan diseases.[18]

18 Review of Access to New Medicines - gov.scot (www.gov.scot)

In Scotland, the Scottish Medicines Consortium (SMC) appraises the clinical and cost-effectiveness of newly-licensed medicines.[19] The SMC carries out appraisals of new medicines, based on the clinical and cost-effectiveness of the medicine at a population level. The SMC then determines whether a medicine should be accepted for routine use within the NHS in Scotland, and publishes advice for Health Boards. The 2016 Montgomery Review recommended that an alternative assessment pathway be developed for ultra-orphan medicines that preserves the integrity of the SMC and its processes, yet achieves a level of access to these medicines that is comparable for orphan and end-of-life medicines.

In response to the review findings, the Ultra-Orphan Pathway was launched in 2018, to support people living with ultra-orphan diseases to receive faster access to new and innovative medicines.[20] The Ultra-Orphan Pathway supports pharmaceutical companies to make medicines available through the NHS in Scotland for an initial period of three years, to enable further data collection on the medicine's efficacy, prior to a further review by the SMC on its routine use in NHS Scotland at the end of the initial three year period on the pathway.

Since 2018, a number of medicines for rare diseases have become available through the Ultra-Orphan Pathway, such as nusinersen (Spinraza®) for the treatment of 5q spinal muscular atrophy, and burosumab (Crysvita®) for the treatment of X-linked hypophosphatemia in children and adolescents. The introduction of the Ultra-Orphan Pathway has and continues to support patients to receive faster access to new medicines for rare diseases.

Why Medicines Matter

The Why Medicines Matter project was conceived by Genetic Alliance UK to better understand the views of stakeholders on steps that can be taken to improve access to medicines for rare conditions in Scotland. An initial meeting of experts took place in May 2021 to understand the key issues and challenges to improving access to medicines for rare conditions. Emerging themes from this meeting were then workshopped and surveyed among key stakeholders.

A summary report, and long-form magazine-style report on the project's findings, were published on 19 July 2022.[21] Presenting views from clinicians, industry, third sector partners and those with lived experience, the report provided a wealth of information on how people living with rare conditions are impacted by access to medicines. The report set out 25 recommendations to assist discussions of how to address this important priority. These recommendations can now be taken up by RDIB and other groups to plan how we will consider the steps that ought to be taken to support wider access to medicines for rare conditions in Scotland.

Action 16: What does this mean for people living with a rare disease?

By taking on board feedback from the rare disease community and from healthcare professionals, we will ensure that our engagement with new medicines takes account of the community's priorities. Better access to new medicines will improve treatment and care for people who can benefit from these developments.

19 SMC | Scottish Medicines Consortium

20 Ultra-orphan medicines pathway: guidance - gov.scot (www.gov.scot)

21 Why Medicines Matter | Genetic Alliance UK

Action 17: Digital Front Door

This action impacts several priorities. We are using data across health and social care more efficiently and effectively than ever before, empowering patients to take better control of their health and care journey. The Covid-19 pandemic has increased our reliance on digital developments and accessible data, and we want to harness these advances to create a more seamless digital experience in healthcare for the future.

As referenced earlier, the Digital Health and Social Care Plan and subsequent delivery plan will be central to this underpinning theme. While not specific to rare diseases, the Digital Health and Social Care Plan sets out a number of actions which will benefit people living with a rare disease by improving access to care, reducing the burden of coordinating their care and improving access to the information they need to live their lives.

The Digital Front Door work will be a key enabler for people interacting with health and social care services in Scotland. This development will aim to allow anyone to book or rearrange appointments, order prescriptions, update their details and generally conduct routine 'transactions' online. This will support better coordination of care for people living with a rare disease and provide the ability for people to use digital products and services to manage their condition. This means being able to access health assessments, diagnosis, monitoring and treatments, making it an option for everyone with a long-term condition to use digital tools. This will be crucial to all of the priorities of this Framework in the short, medium and long term.

The response to the Covid-19 pandemic has accelerated the pace of change across health and social care, with services moving quickly and innovatively to provide better access to flexible and digitally enabled support. For many people, this has increased choice and flexibility. For services, it has eased pressures, freeing up time and capacity for services which cannot be delivered digitally. One of those advancements is the rapid expansion of Near Me, also known as Attend Anywhere. This is a safe and secure NHS video consulting service that enables people to attend appointments from home or wherever is convenient.

During the pandemic, the use of Near Me in Scotland rose from around 1,000 consultations per month to a peak of 90,000 per month. Current usage is around 40,000 consultations per month. A consultation completed by clinicians and members of the public highlighted that:[22]

- There was high acceptability for the use of video consulting with 87% of public respondents saying video appointments should be offered.
- Members of the public preferred the use of video over phone consulting.
- Near Me has a wide range of potential uses.

In March 2022, we reached 1.5 million Near Me appointments across Scotland, saving an estimated 49 million travel miles for patient, families and staff.

Our aim is to continue to provide safe, person-centred and sustainable care through video consulting. Where appropriate clinically and for the individual, everyone should have the choice of attending appointments via an easy and convenient Near Me video call.

22 Video consultations - public and clinician views: consultation summary - gov.scot (www.gov.scot)

We need to make sure that efforts to add more Digital elements into the health and social care system are proportionate to ensure that nobody is left behind, while meeting the expectations of those who want to interact in this way. Digital Front Door will be an iterative development with the first release expected in 2023/24. There will be a programme of engagement as part of the development process, and we will seek participation from the rare disease community to ensure that they are included in this process in the short and long term.

Action 17: What does this mean for people living with a rare disease?

The use of new digital elements in healthcare consulting can save on travel, and allow appointments to be conducted in the comfort of a person's own home. Remote consultations such as Near Me video calling should be offered as a choice if it is appropriate for the person and their condition, with the option of an in-person appointment if they prefer.

Action 18: Clinical Research for Rare Disease

Clinical research for rare diseases presents a number of challenges for Scotland, but we know how valuable this is to patients.

Such challenges include the distances involved between clinical research centres and the low population density, factors which make equitable access to clinical trials more difficult to achieve. At the same time, the Network of Clinical Research Centres in Scotland, and the development of the Congenital Anomaly Register (CARDRISS) both provide an opportunity to develop new approaches that can improve access to interventional and observational clinical studies for patients.

In parallel to improving access to research for people with rare disease, there has been rapid evolution in genomic technology that has the potential to make significant improvements to health of the Scottish population. The development of a strategy for genomics will be highly relevant for the considerable proportion of rare disease that has a genetic basis.

We will therefore focus on three areas:

Develop a Scottish Register for Rare Disease

As covered in Action 3, in the short term, we are committed to consolidating the registration of congenital conditions covered by EUROCAT through the national congenital and rare condition registry service, CARDRISS. In time, we are also committed to extending the scope of the CARDRISS register to cover additional congenital and rare conditions covered by the pregnancy and newborn screening programmes.

Over the medium term, we will work with CARDRISS to develop plans for further extension of the registry service to cover a wider range of rare conditions affecting children and adults. We will work with the registry services in England, Wales, and Northern Ireland to ensure alignment across the UK in this work where feasible and beneficial.

Development of new national data returns providing information on the results of germline genetic testing (summary results, not full DNA sequencing data) from clinical genetic laboratories to Public Health Scotland will be critical to both consolidating current registration of congenital conditions and allowing CARDRISS to cover other rare conditions. We will work with CARDRISS, the team procuring a new information system for labs across Scotland, and the SSNGM to develop these new genetic testing data returns.

Strengthening national data on congenital and rare conditions through CARDRISS will provide many benefits, improving our understanding of specific conditions, supporting policy and service development, and enabling research, including through identification of individuals who may benefit from inclusion in clinical studies. Fit-for-purpose ethical safeguards will need to be developed which protect patients, but facilitate access to data, within the existing governance structure. As noted under Action 3, development of new national data returns and extending the remit of CARDRISS will require investment, which we will consider as part of our future budgetary considerations.

Improve networking of research across Scotland

We have established a collaborative group with representation that includes clinical investigators, NHS research Scotland and industrial partners. This group will identify the barriers to a "One Scotland" approach to conducting clinical research. Suggested improvements are expected to include an improved governance structure that facilitates referral of patients to studies between regions and a hub and spoke model for the organisation of clinical trials. We will also explore the development of a Rare Disease Platform for observational research in rare disease that allows integration with the NIHR UK Rare Genetic Disease Research Consortium Agreement.

Develop Genomics research infrastructure

The most common clinical indication for whole exome or whole genome sequencing in Scotland is for the diagnosis of rare disease. The Rare Disease Implementation Board will, with the new Scottish Strategic Network for Genomics Medicine, seek to deliver safe storage of sequence data within a data system that includes research functionality, but also permits reanalysis of extant sequence data as knowledge improves, providing an increased diagnostic return on testing.

Action 18: What does this mean for people living with a rare disease?

Engaging with the wider research landscape in this way will enable access to more studies of benefit to people living with rare diseases. Increased research into rare diseases, and linking this with our genomics offering, will lead to faster diagnoses and help identify new treatments.

5. Next steps

As can be seen from this first iteration of Scotland's Action Plan for Rare Diseases, there is much to be done to ensure that the lives of people living with rare diseases continue to improve. There are examples of good practice in a number of places: we must ensure that these are made available to all who can benefit from them, whether that is a learning resource for a healthcare professional or a care/support resource for a person with a rare condition.

The actions that we have set out in this first Action Plan will be achieved at different stages. Some things can be put in place quickly and use existing resources in smarter, better-signposted ways; others will take more time and investment and be subject to available funding. In either case, we will always work at pace to ensure that people in Scotland living with rare diseases start to notice real change. We will keep agile in responding to new developments and the rapid pace of change in health and social care, and always keep the voice of those with lived experience at the heart of this.

We will continue to work with our counterparts in the other UK nations to discuss and work together on the implementation of our actions plans. This knowledge-sharing will help us all to maximise the impact of our actions, both in our own nations and collaboratively. We will continue to share our progress at the meetings of the UK Rare Diseases Framework Board and elsewhere.

Throughout the lifespan of the Framework and our Action Plan, we will continue to engage with people living with rare diseases. We know that your lived experience is the most valuable in informing our work on what challenges need to be addressed, and it will be crucial to keep the rare disease community informed on the progress being made. We will keep working with Genetic Alliance UK and other key partners to schedule further engagement opportunities at which we can discuss our progress with you and identify further priorities for the future.

Monitoring and Evaluation

We recognise that measuring outcomes for patients, carers and families will be difficult.

However, measuring the success of our actions in the short to medium term will be achieved in different ways, depending on what needs to be measured. Work such as better signposting of awareness-raising resources can be measured by assessing their take-up among healthcare professionals, and professional and patient reported improvements in the time taken to diagnose. We will look to develop metrics by which to measure the success of our various actions, such as patient-reported outcome measures.

Annex – Underpinning Themes

The priorities and actions set out in this plan are crucial to improving the lives of those living with a rare disease. We also recognise though that there are a range of activities and actions that will require implementation outwith the four main priorities. The Framework identified five underpinning themes in which work will continue to be progressed to support the priorities and contribute to improving the lives of people living with rare diseases.

The work on these themes will be embedded into our actions on the key priorities. In addition to doing so, the information below sets out a summary of our commitments to each underpinning theme below.

Patient Voice

People living with a rare disease are best placed to shape and influence our policies on rare diseases. We recognise this and have embedded the voice of those living with a rare disease into the government of our Action Plan. However we also recognise the need to identify the best method of engagement to ensure it doesn't add a burden on to patients and carers. We will work with rare disease organisations to achieve this, taking advantage of existing groups and platforms wherever possible.

National and international collaboration

The small numbers of patients with individual rare diseases make collaboration essential, both for the support of patient care and the delivery of robust research. We are committed to continuing collaboration with the rare disease community across the world, including patients, healthcare professionals, researchers, and industry, to share knowledge and ideas to improve outcomes.

We will continue to work closely with our counterparts in the UK Government and the other devolved administrations to ensure close alignment of the rare diseases Action Plans that each of the four nations has developed.

One key area of collaboration is on national registries for congenital anomalies.

Pioneering research

Participation in research, and promotion of opportunities for research, play vital roles in how our healthcare is delivered and improved. We have set out a clear action in this plan on how we might address this. This is at the centre of all we do. For people living with a rare disease, research can fulfil unanswered questions that may support a diagnosis, cause, symptoms and treatment for many rare diseases.

The Scottish Government, through the Chief Scientist Office (CSO), has established and maintained an active programme of co-funded research with third sector organisations, including research in rare diseases.

NHS Research Scotland is committed to actively involving patients, those who care for them and the public in all aspects of the research process. This includes shaping future research activity and there are currently supporting more than thirty genetics studies, about a third of which are led from Scotland.

We will continue to work with CSO and the NHS Research Scotland infrastructure to enable more opportunities for people with rare genetic diseases to participate in research, which in turn leads to improved care and the development of new treatments.

Digital, data and technology

Across health and social care, we are using data more efficiently and effectively than ever before, empowering patients to take better control of their health and care journey. The Covid-19 pandemic has increased our reliance on digital developments and accessible data, and we want to harness these advances to create a more seamless digital experience in healthcare moving forwards.

As referenced earlier in the plan, the Digital Health and Social Care Plan and subsequent delivery plan will be central to this underpinning theme. While not specific to rare diseases, the Digital Health and Social Care Plan sets out a number of actions which will benefit people living with a rare disease by improving access to care, reducing the burden of coordinating their care and improving access to the information they need to live their lives.

Wider policy alignment

We recognise that the impact of living with a rare disease is much wider than just a person's health. Due to the nature of their condition, many people living with a rare disease require housing adjustments, social care, financial aid, mental health support and special educational needs support. Wider policy development in these areas must reflect the needs of those with rare conditions.

Over the long term (3+ years) and linked with our patient voice priority we will work with policy makers across Scottish Government to put forward the needs of the rare disease community when developing policy. Given the importance of genomics in improving diagnosis for some rare diseases, and to support new research into potential treatments, our plans will align with our Genomics Implementation Plan, as well as other relevant strategies and policies.

We will work with policies across the Scottish Government to ensure those living with a rare disease are represented during the development of relevant policies.

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