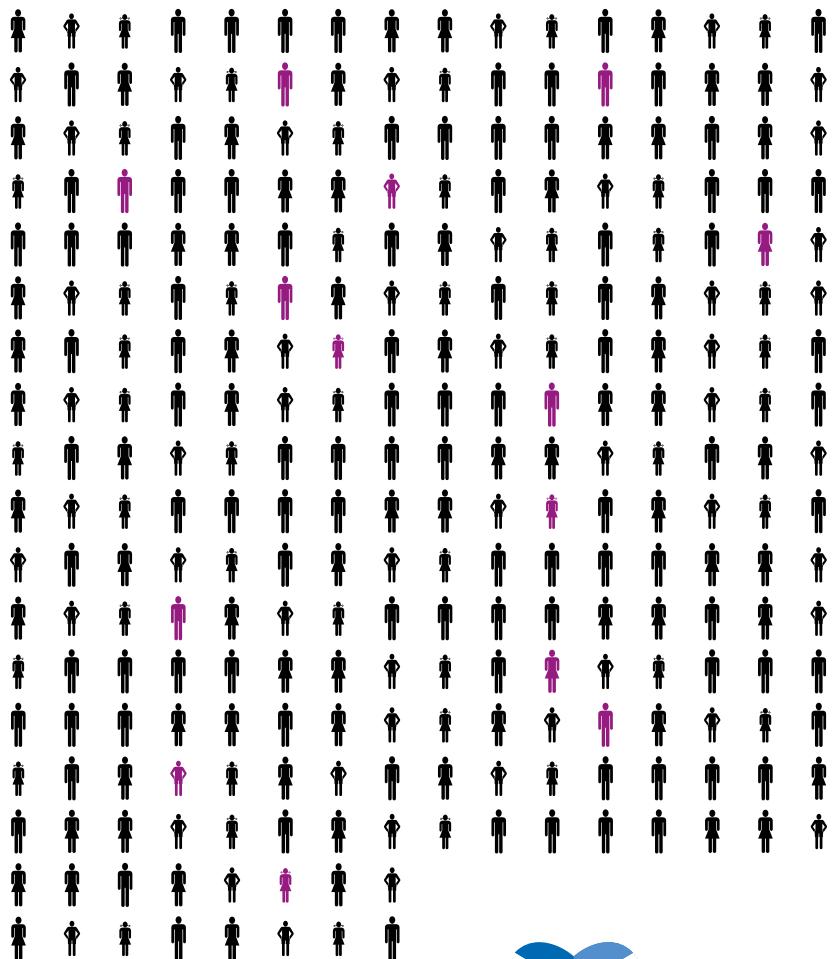


# RARE DISEASE SCOTLAND PROGRESS REPORT

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2016 - 2018

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Healthier  
Scotland

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# FOREWORD

The past two years have seen significant changes in the world of rare disease, especially the rapid advancement of genomics. Genomic medicine has the potential to revolutionise healthcare, especially in terms of diagnosis timescales and treatments for people with rare diseases. I am pleased that Scotland is participating in this exciting discipline, and look forward to seeing how this will develop over the next few years.

While genomics will change healthcare in the future, it is important that government and the National Health Service do not lose sight of the person at the centre of the care we provide. It is therefore very encouraging to see legislation, policies and frameworks that aim to improve health and social care now being drafted through listening to stakeholders, and most importantly people who access these services, to put the person at the very centre of their own healthcare. I am pleased to include my own Chief Medical Officer's report, 'Realising Realistic Medicine' in this.

I am also pleased at the progress that has been made against the 51 commitments set out in the UK Strategy for Rare Disease. Of course there is still a lot of work ahead, and challenges to face. I would encourage you to give any feedback you can, to ensure that placing the person at the centre continues to be at the heart of these commitments.



*Catherine J Calderwood*

**Dr Catherine Calderwood MA Cantab FRCOG MBChB FRCP,  
Chief Medical Officer for Scotland**

The design and quality of services for people is key to the future of the NHS. While a large number of people have a rare disease, as many as 1 in 17 people in Scotland, we continue to hear of difficulties from people who have experienced long journeys to receiving timely diagnosis, and not always receiving joined up specialist and social care that they need.

This report shows that a lot of good work is being done to improve the road to a faster diagnosis through the introduction of genomics and the policies and frameworks that seek to put the person at the centre of their own care. However, much more is needed to raise awareness of rare diseases and to promote a more integrated approach to services, which cuts across all disciplines and departments and to embed seamless care and support.

In the report, we seek to capture the progress made against the main themes of the UK Strategy for Rare Diseases which we believe will help to address some of these issues and deliver the highest quality services. However, there is still plenty of work to do before we reach the targets of 2020 and beyond.



*Tracey Gillies*

**Tracey Gillies, Chair, Rare Disease Implementation Oversight Group**

# INTRODUCTION

The Scottish Implementation Plan, *It's Not Rare to Have a Rare Disease*<sup>1</sup> was first published in 2014. The title came from the statistics that showed whilst rare diseases in themselves affect a small number of people (the definition states 5 people or fewer in 10,000) there are actually between 6,000 – 8,000, and these affect approximately 8% of the population. To put this in context, there are approximately 424,000 people out of a population of 5,295,000 in Scotland with a rare disease.

*It's Not Rare to Have a Rare Disease* was produced following the publication of the the UK Strategy for Rare Diseases<sup>2</sup> in November 2013. The Strategy includes a list of 51 commitments that cover five specific areas.

- Empowering those affected by rare diseases
- Identifying and preventing rare diseases
- Diagnosis and early intervention
- Co-ordination of care
- The role of research.

The Strategy forms the basis of work across the four countries of the UK to improve services, support patients and promote the role of research. Each country has then followed onto produce an implementation plan of how this will be done.

Part of the UK Strategy included a commitment to publish a biennial report on progress against each of the commitments. The first report, 'Delivering for patients with rare diseases: Implementing a strategy'<sup>3</sup> was published in 2016 and noted progress from the UK as a whole. The second report, will be published in February 2018. However, the Rare Disease Implementation Oversight Group felt that it would be beneficial to publish a report based on the progress made in Scotland. As a result, this report notes the progress that Scotland has made against each of the 51 commitments and the actions that need to be taken in order to meet all the Commitments by 2020.

There are a number of areas that may be affected by the UK exit from the European Union, however, at this point in time it is too early to say what this means fully. The Scottish Government will be working closely with colleagues in the rest of the UK to ensure that people living with a rare disease in the UK will still receive the best care and services that are available.

<sup>1</sup> <http://www.gov.scot/Publications/2014/07/4751>

<sup>2</sup> [https://www.gov.uk/government/uploads/system/uploads/attachment\\_data/file/260562/UK\\_Strategy\\_for\\_Rare\\_Diseases.pdf](https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf)

<sup>3</sup> <https://www.raredisease.org.uk/media/2364/ukrarediseaseforum-progress-report-2016.pdf>

# 1. EMPOWERING THOSE AFFECTED BY RARE DISEASES (1 – 8)

Commitment	Description
1	Strengthen the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research, recognising patient groups as key partners – including in the development of the four country plans to implement the Strategy.
2	Improve awareness amongst service providers and others of the effects that rare diseases can have on a person's education, family, social relationships and ability to work.
3	Encourage effective and timely liaison between the NHS and other public service providers, and encourage providers to consider the effects of rare disease on people's lives when they are developing and managing services.
4	Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.
5	Consider how to give all patients with rare disease clear and timely information about: their condition and its development; treatment and therapy options; practical support.
6	Improve access for patients (or where appropriate their parents or guardians) to their personal data.
7	Support patients to register on databases, where these exist.
8	Help patients to contribute to research and other activity related to rare disease.

The Scottish Government is fully committed to empowering people in terms of their health and social care. A number of plans, policies and recommendations have been made since the last biennial report that are being driven and delivered across Scotland. These will also directly impact on people who are affected by rare diseases. The plans and policies listed below give a brief explanation of the work that is progressing throughout Scotland.

## 1.1 The Rare Disease Implementation Oversight Group

The role of the Rare Disease Implementation Oversight Group is to monitor the implementation of the Scottish Rare Disease Plan 'It's Not Rare to Have a Rare Disease' and to ensure that the 51 Commitments in the UK Strategy for Rare Disease are being met.

The group is made up of clinicians, geneticists, biochemists, as well as representatives from NHS National Services Scotland, Patient groups, NHS National Education Scotland, the Farr Institute and the Scottish Government.

## **1.2 Health & Social Care Delivery Plan**

The Health and Social Care Delivery Plan<sup>4</sup>, published in December 2016, sets out the Scottish Government's plans for enhancing health and social care services by 2021. The plan aims to focus on care being provided to the highest standards of quality and safety, whatever the setting, with the person at the centre of all decisions. The Audit Scotland report 'NHS in Scotland 2016'<sup>5</sup>, underlined the importance of bringing together the different programmes of work to improve health and social care services.

### **Delivering the Plan**

The plan seeks to build on the excellence of NHSScotland, recognising the critical role that services beyond the health sector must play and ensuring that it is ultimately fit for the challenges facing us as a country. It focuses on three aims:

- Better care – “we will improve the quality of care for people by targeting investment at improving services, which will be organised and delivered to provide the best, most effective support for all”.
- Better health – “we will improve everyone’s health and wellbeing by promoting and supporting healthier lives from the earliest years, reducing health inequalities and adopting an approach based on anticipation, prevention and self-management”.
- Better value – “we will increase the value from, and financial sustainability of, care by making the most efficient and consistent delivery, ensuring that the balance of resource is spent where it achieves the most and focusing on prevention and early intervention.”

One of the main principles of the plan is that individuals, and where appropriate, their families should be at the centre of any decisions that affect them. They should be given more freedom, choice, dignity and control over their care. Care planning should anticipate the individual’s health and care needs – both by helping those with chronic and other complex conditions to manage their needs more proactively.

There are a number of actions set out within the plan with a view to achieving the aims, below are a sample of examples.

- In 2017, the Scottish Government will ensure that Health and Social Care Partnerships (NHS boards, local authorities and other care providers) make full use of their new powers and responsibilities to shift investment into community provision by reducing inappropriate use of hospital care and redesigning the shape of service provision across hospital, care home and community settings. This will be a key lever in shifting the focus of care across health and social care services.
- By 2018, the aim is to have increased health visitor numbers with a continued focus on early intervention for children through addressing needs identified through the Universal Health Visiting Pathway<sup>6</sup>, which started in 2016. As a result of this, every family will be offered a minimum of 11 home visits including three child health reviews by 2020, ensuring that children and their families are given the support they need.

<sup>4</sup> <http://www.gov.scot/Resource/0051/00511950.pdf>

<sup>5</sup> <http://www.audit-scotland.gov.uk/report/nhs-in-scotland-2016>

<sup>6</sup> <http://www.gov.scot/Resource/0048/00487884.pdf>

- By 2021, the aim is to ensure that everyone who needs palliative care will get hospice, palliative or end of life care. All who would benefit from a ‘Key Information Summary’<sup>7</sup> will receive one – these summaries bring together important information to support those with complex care needs or long-term conditions, such as future care plans and end of life preferences. More people will have the opportunity to develop their own personalised care and support plan. The availability of care options will be improved by doubling the palliative and end of life provision in the community, which will result in fewer people dying in a hospital setting.
- By 2021, the aim is to have strengthened the multi-disciplinary workforce across health services. We will have a refreshed role for district nurses by 2017, train an additional 500 advanced nurse practitioners by 2021 and create an additional 1,000 training places for nurses and midwives by 2021. This will build on four successive increases in student nursing and midwifery intakes to meet additional demand, especially in primary and community settings.
- By 2021, the aim is to have increased the number of undergraduates studying medicine by 250 as a result of the 50 additional places in Scotland’s medical schools introduced in 2016.

All the actions can be viewed in the plan at: <http://www.gov.scot/Publications/2016/12/4275/downloads>

The Health and Social Care Delivery Plan addresses Commitments 3, 4, 5 and part of 11 (see below) of the UK Strategy for Rare diseases. One of the most common themes in feedback responses from people with rare diseases and their carer/families is that it is very difficult joining up health and social care. This plan for Scotland will mean that the people with a rare disease will be central in the care planning process, with all elements of primary, secondary and social care involved in the journey and planning of care. It ensures that the person is at the centre of all the care and support they require, that they have a say in their care package and are given freedom, choice and say in the decisions that directly affect them.

### **1.3 Realistic Medicine**

In 2016, the Chief Medical Officer’s annual report introduced the concept of “Realistic Medicine”, this highlights that decision making about an individual patient’s healthcare should be focused on the individual and discussed and agreed with them, their family/carer and the clinician. It is about providing people the treatment that is right for them at the right time with the right support. Its aims of reducing harm and waste, tackling unwarranted variation in care, managing clinical risk, and innovating to improve are essential to a well-functioning and sustainable NHS. Realistic Medicine is not about rationing healthcare; it is about changing the overall approach to healthcare.

The full report can be found at: <http://www.gov.scot/Resource/0049/00492520.pdf>

This was followed up in 2017 with ‘Realising Realistic Medicine’<sup>8</sup> which talks about realising the vision of Realistic Medicine for the future – “By 2025, everyone who provides healthcare in Scotland will demonstrate their professionalism through the approaches, behaviours and attitudes of realistic medicine”.

Realistic Medicine addresses Commitment 1, 4 and 5 of the UK Strategy for Rare Diseases. It means that the person with a rare disease and/or their carer/family will have a direct say in their treatment, taking account of what is right for them and their circumstances. No-one should have decisions made about them without them having a say, and they should fully understand what will happen, what their treatment will be and how it will affect their life.

<sup>7</sup> <http://ihub.scot/media/2665/kis-best-practice-statement-from-the-scottish-government.pdf>

<sup>8</sup> <http://www.gov.scot/Resource/0049/00492520.pdf>

## **1.4 Making it Easy: A Health Literacy Plan for Scotland**

In order for patients to feel empowered, they must have the knowledge, understanding, skills and confidence to use health information and be active partners in their care, they must be health literate. The Scottish Government is currently taking forward the ‘Making it Easy: A Health Literacy Action Plan for Scotland’<sup>9</sup> in order to “make Scotland a health literate society that enables all of us to live (and die) well on our own terms and with any health condition we may have.” This has involved developing an online national literacy resource for Scotland (The Health Literacy Place<sup>10</sup>), developing an e-learning module for NHS staff and establishing a national demonstrator site across NHS Tayside. Projects within NHS Tayside have included improving signage and navigation of the hospital environment, using simpler language in appointment letters and utilising the ‘teachback’ technique to check patient understanding. A progress report was published in July 2017 and a new Action Plan was published in December 2017<sup>11</sup>.

This links in with the Scottish Government’s work in enacting the Patient Rights (Scotland) Act 2011 and in particular the legal right for patients to participate as fully as possible in decisions and to be given the necessary information, in a way they can understand, to allow this participation.

Health literacy addresses Commitments 3, 4 and 5 of the UK Strategy for Rare Diseases. It is very important for people with rare diseases and their carer/family members that they understand the condition, the effects, the treatment options available, what they entail and what the outcomes are. Additionally, those living with a rare disease often need to attend numerous appointments, therefore it is important that the person and carer/family member understand what the appointments are for and where the relevant clinic is. This should be a straight forward process that does not add further stress onto what can already be a stressful time for patients and families.

## **1.5 House of Care Model for Scotland**

The House of Care Model for Scotland<sup>12</sup>, developed by the Scottish Government in partnership with The Health and Social Care Alliance Scotland<sup>13</sup> (The Alliance), provides a simple visual model of a house build around collaborative care planning conversations between people and their health care professionals. The care and planning conversations are at the centre of the house/model and are supported by aspects such as health literacy, ‘more than medicine’, committed multi-disciplinary teams of clinicians and effective organisational processes and arrangements.

9 <http://www.gov.scot/Publications/2014/06/9850>

10 <http://www.healthliteracyplace.org.uk/>

11 <http://www.gov.scot/Resource/0052/00528139.pdf>

12 <http://www.gov.scot/Publications/2016/02/8618/7>

13 <https://www.alliance-scotland.org.uk/>



Building the House of Care requires the kind of whole-system transformation needed to recognise the assets, rights and capabilities of people, and place them in the driving seat of their care and support. The Scottish Government has provided £505,000 funding towards implementation of the House of Care model in addition to the £395,000 provided by the British Heart Foundation. The approach has been implemented within 55 GP practices across five adopter sites (Lothian, Glasgow, Tayside, Ayrshire & Arran and Lanarkshire).

The purpose of the care and support planning conversation itself is to empower the patient; it recognises that although healthcare professionals have knowledge and expertise about the clinical care of a condition, the person with the condition knows best how it impacts on their life. The focus should be on what matters to them rather than their condition or disease. It is a real opportunity for them to share information and openly discuss issues and concerns, as well as to get help with accessing the services and support they require to live with their disease or condition. Dr Sue Arnott, Clinical Lead on House of Care for NHS Lanarkshire states that House of Care “is a way to allow patients to have the opportunity just to become involved again, because they were used to having tests and tasks done and things measured, but when you actually get down to it there was very little opportunity for them to tell their story and I really felt that this was a way for them to do that”.

House of Care addresses Commitments 1, 2, 3, 4 and 5 of the UK Strategy for Rare Diseases. Very often a person with a rare disease and/or their carer/family member will become fully knowledgeable in the particular condition. House of Care recognises this and acknowledges that sometimes while clinicians will know about the care and treatment of the condition, the personal experience of the person with the rare disease is not always taken into account. House of Care will mean that the focus of discussion will be on what matters to the person, and will allow them to share information on what they know and have learned about the rare disease, and openly discuss their concerns, preferences and any issues. It should also help the person to access services and support that will help them to live with their rare disease.

## **1.6 What Matters to You?**

In 2016, the Scottish Government and Healthcare Improvement Scotland (HIS) formed a partnership with people who work in care to hold Scotland's first 'What Matters To You?'<sup>14</sup> day on Monday 6 June 2016. 'What Matters to You?' day aims to encourage and support more meaningful conversations between people who provide health and social care and the people, families and carers who receive health and social care. This has now become an international movement with many countries around the world participating.

The 'What Matters to You?' approach goes beyond health and social care, reaching many other parts of the public sector that impact directly on people's health and wellbeing. In 2017, schools, child minders, housing associations and other parts of the public and the third sector in Scotland, all signed up and participated.

By talking to the people they are caring for about what's important to them, listening to the answers and taking action on what they hear, staff can provide the care and support that people really need and want.

The aim of the Scottish Government and HIS is that this becomes an on-going conversation, taking place every day in health and care settings across Scotland.

More information and stories about 'What Matters to You?' stories can be found on the website at: <http://www.whatmatterstoyou.scot/stories-asking-matters/>

The following example is taken from the 'What Matters to You?' day report 2016 and demonstrates that the things that matter to people might not necessarily be medical issues.

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<sup>14</sup> <http://www.whatmatterstoyou.scot/>

## **Staff Involvement with ‘What Matters to You?’**

I spoke with the mother of an 18 year old boy who has Down’s syndrome – he has recently made the transition from school (and children’s services) to a day centre and adult services.

She discussed with me that she felt that it was important that she remains a big part of her son’s care. Although he is 18, he has the mindset of a younger man and still looks to his mother for comfort. Her biggest fear would be if he was told that this was not appropriate.

We discussed how now that he attends the day centre, he would be encouraged to make appropriate relationships (no cuddling, only handshakes etc) but that he would be encouraged to keep his loving embraces for his mother. She seemed very pleased with this.

In contrast to this, she also discussed with me how it was important to her that his sleep pattern gets better again, as recently he has been up a lot through the night and she feels that as he is an adult now, she can’t tell him what to do.

I discussed with her that I would educate him around his sleep hygiene pattern and should the need arise will attend a GP appointment to have this reviewed.

We also discussed how it was important to her that he attended his day centre. She struggles with how much he misses his friends so when he is having a bad day, he refuses to get out of his bed and go to the centre. I discussed how I would create a storyboard for him which she can go over with him which will remind him about all the new friendships he has made at his day centre and also about the groups he attends through his centre at another centre within the area (where he sees some of his old friends from school). I asked Mum if she felt that it would be better to use actual pictures of him at his centre and she agreed that he would respond better to this. I then contacted his centre and clarified that this would be possible and they will supply photographs of him at the centre.

His mother was very happy with our discussion and the planned outcomes and thanked me for my help.

‘What Matters to You?’ addresses Commitments 1, 2, 3, 4 and 5 of the UK Strategy. It is a question that should be asked of people with a rare disease, and their carers/families. Quite often people feel like they are not included in their care and the real issues that are facing them are not discussed in the health care setting. The ‘What Matters to You?’ initiative helps to facilitate this discussion by training staff to really listen to what matters to the individual and improve the care and support that they really need. It will also help to raise awareness of rare diseases as staff will hear first-hand experiences of the rare disease and what it is like to live with the condition.

## 1.7 Care Opinion



Care Opinion<sup>15</sup> (previously Patient Opinion) is a not-for-profit social enterprise that provides an online feedback service that enables people in Scotland to give real-time feedback, and engage in constructive dialogue with healthcare service providers about the services they, their families, or the people they care for, have received.

The Scottish Government has supported the roll out of Care Opinion across Scotland since 2013. A contract was awarded in April 2015, which provides for every territorial NHS Board in Scotland and relevant special boards to be fully registered with the service. Care Opinion have recently been awarded a new contract and the Scottish Government will continue to provide for the NHS Boards to fully register with the service until 31 March 2020.

All Boards subscribe to Care Opinion and are reading and responding to issues directly. There are now around 1000 NHSScotland staff reading stories and we continue to see a higher distribution of staff responding as time goes on. This allows people posting stories to receive a targeted response and provides assurance that their concerns, or messages of thanks, have reached the appropriate staff member(s).

Care Opinion held an event in Glasgow on 17 May 2017 for clinicians wishing to explore how they and their teams can use Care Opinion to support their local learning and improvement work. Some clinicians at this event reported barriers to testing ways of supporting clinicians to respond to postings on Care Opinion. This was subject to social media discussion, which involved some Scottish Government officials emphasising support to empower more clinicians to use Care Opinion if they wish to do so. A further event for Clinicians is planned for February 2018.

In 2017, there were 3029 stories about health and social care services in Scotland shared, and to date over 10,000 stories have been shared. Of these stories, 60 have initiated changes in how health and social care services are being delivered, these include:

- NHS Ayrshire & Arran are making changes to ensure appropriate support is in place for vulnerable people "[Sending a vulnerable adult home alone](#)"
- NHS Borders have created a new information leaflet for people attending ambulatory care appointments "[We came away with no questions answered](#)"

Care Opinion intends to focus on development in the following areas:

- Support Care Opinion and NHS Board clinical leads to explore effective ways to empower and support clinicians to respond to posts and to use information to further refine and develop clinical involvement, that is linked to local learning and quality improvement work.
- Continue to seek time-effective ways for subscription managers across Scotland to share best practice.
- Seek ways to improve site accessibility and enable a wider range of people to share their experiences.

15 <https://www.careopinion.org.uk/info/care-opinion-scotland>

- Create case studies and further exemplar videos to demonstrate the impact of using Care Opinion in order to encourage feedback.
- Continue to raise public awareness through partnerships with third sector patient-led organisations, social and conventional media.

Care Opinion addresses Commitments 2, 3 and 4 of the UK Strategy for Rare Diseases. It allows people to share their experiences of the care they have received from the NHS. These can be either positive or negative experiences which are directly accessed by the relevant staff in the relevant health board. It gives staff the opportunity to hear and address first-hand experiences of care. This can help open a dialogue between the person and staff member(s), or allow staff member(s) to consider making changes to service provisions and/or share good practice with other health boards.

The extracts below are from people and relatives of people with a rare disease.

### **Steven-Johnson Syndrome**

#### **Comment from Patient:**

My son became seriously unwell, developing a nasty condition called Stevens Johnson Syndrome. After several contacts with our local GP practice within a 24 hour period, and a trip to our local out-of-hours service, he was admitted to hospital.

His condition worsened and the situation was distressing for all involved, including the staff, most of whom had never experienced this illness before.

However, I cannot fault the professionalism of the people looking after him, in particular Dr Kelly and an amazing nurse, Heather. Heather clearly does not just do her job, she has an exceptional gift of engaging with children and displaying a level of empathy I have never seen in any professional before.

After six days of not being able to eat, Heather told my son she'd find him anything he wanted, and she did. He was still unable to eat but just being able to have a small taste of something he liked meant a lot. Of note, other fantastic nurses on the ward were Victoria, Hazel and Emma. Committed, talented nurses.

I could give many more examples of how these special individuals went the extra mile but, what was truly the worst week of our lives, could have been much worse had it not been for these fantastic, unsung heroes. The NHS is full of them.

#### **Response from NHS Board:**

Thank you very much for taking the time to share your experience with us at such a difficult time for you and your family. It is always rewarding to have the hard work and kindness of staff recognised. I will ensure all staff involved in caring for your son are aware of your comments.

I hope your son continues to make progress.

Best wishes.

## **Neurofibromatosis**

### **Comment from patient:**

My sister lives in your region, she doesn't drive and has two young children. To get to the hospital, she needs to take two trains and a taxi, and the return journey, including her appointment takes in the region of seven hours. I explained to her at the time that our local hospital doesn't provide neurological services and that is why she has to travel, however, what I am struggling to understand now is why she needs to keep attending there for follow up appointments.

### **NHS Board Response:**

I've passed your feedback to the managers for both of these services to highlight the points you've made. The General Manager for Ophthalmology would like to have a look at this in more detail to see if there's a way we could change this to make it easier for your sister.

### **Patient response:**

Thank you for the quick and helpful response. I have updated my sister tonight and she is really pleased someone is at least willing to properly consider the options around this. She will be in touch via email with her personal details.

## **1.8 Our Voice**

In June 2014 the Cabinet Secretary for Health and Wellbeing said:

**"We must do more to listen to, and promote, the voices of those we care for. We need the voices of patients, those receiving care and their families to be heard in a much clearer and stronger way."**

Lots of different organisations have come together to develop Our Voice<sup>16</sup>, including the Scottish Government, NHS Scotland, the Scottish Health Council, HIS, the Convention of Scottish Local Authorities (COSLA), the Health and Social Care Alliance Scotland (the ALLIANCE) and other third sector partners.

Before developing the Our Voice framework, they wanted to ensure that they were delivering something that people wanted, was useful and would make a difference to how people's voices are heard across Scotland. Many different engagement activities were undertaken, including national events, discussion groups, online surveys and connecting through social media in order to speak to as many people as possible.

Once the feedback was gathered, the team found **7 key themes** that came up time and again.

They were:

- Active Citizenship**

Support to help people develop the skills needed to take an active role in making decisions about their own care and support. A need for more open and useful conversations between patients, staff and the wider health and social care systems.

<sup>16</sup> [http://www.scottishhealthcouncil.org/patient\\_public\\_participation/our\\_voice/our\\_voice.aspx](http://www.scottishhealthcouncil.org/patient_public_participation/our_voice/our_voice.aspx)

- **Benefits and Outcomes**

Supporting people to speak out and have their voice heard brings positive benefits for health and wellbeing. Encouraging health and social care services to act on what they hear should be a goal of the Our Voice programme.

- **Culture Change**

A change of values and culture is needed. People's views should be welcomed and steps taken to remove barriers such as fears about the effect of giving feedback on future treatment.

- **Methods and Approaches**

There should be a range of ways for people to have their views heard. Face-to-face and online methods should complement each other.

- **Information and Support**

Appropriate support should be available to make sure patients and people who use services have equal opportunity to have their voices heard. This includes clear information on how to get involved and communication support if needed. Access to advocacy and peer support helps build up people's ability to speak up for themselves.

- **Principles and Standards**

There should be a single standard for participation across health and social care. This should build on the good practice that already exists such as the Participation Standard and the National Standards for Community Engagement.

- **A Whole System Approach**

An approach which strengthens the voices of patients, people who use services, carers and members of the public within health and social care must run at individual, local and national levels. This approach means people can have their say and influence decision making throughout the whole system.

The Our Voice website provides information on rights and responsibilities for individuals, information on how to make complaints, self-directed support and advocacy services.

It also contains information on the National Voice, which provides opportunities for people to join in with discussions about national health and social care policy. It can involve joining a website for a discussion forum or attending a group. There is also information on consultations and details of support organisations and networks.

For more information on Our Voice and how you can participate, please see the website:

<https://www.ourvoice.scot/>

Our Voice addresses Commitment 1 and 4 of the UK Strategy for Rare Diseases.

## **1.9 National Specialist Services Committee**

The National Specialist Services Committee (NSSC) is responsible for providing recommendations to the NHS Board Chief Executives' Group (BCEs) and to the Scottish Government Health and Social Care Directorates (SGHSCD) on the commissioning of highly specialised services for patients in Scotland with complex needs or rare conditions.

NSSC reviews proposals for national designation of specialist health services and Managed Clinical Networks. They then make recommendations on whether national commissioning is appropriate for each of the proposed national services/networks. Proposals for national services/networks might arise from service planning by SGHSCD, the National Planning Forum, Regional Planning Groups and NHS Boards; or they might be applications from clinicians for designation of specific services/networks which are supported by their NHS Board(s).

In the first instance, NSD supports appropriate applications on new developments for submission to the National Professional, Patient and Public Reference Group (NPPPRG), who liaise with professional, patient and public representative groups, regional planning groups and other relevant bodies. The group gather information in order to provide expert advice to the National Specialist Screening Committee<sup>17</sup>.

## **1.10 National Network Management Service (NNMS)**

The NNMS commissions national clinical and diagnostic networks on behalf of NHS Boards and the Scottish Government. They provide high quality management and operation support to enable them to focus on adding value to healthcare in Scotland through better access to specialist care.

The national networks ensure patients with rare or complex conditions have access to high quality specialist care. In particular, the diagnostic networks support early diagnosis and intervention by ensuring that patients have the right test at the right time. However, they all bring together everyone involved in providing care when the full range of skills required isn't available within a single health board or region – this includes health professionals, carers, patients families and voluntary groups.

This helps to:

- Better understand and meet patients' and carers' needs.
- Facilitate the design and mapping of services across Scotland.
- Measure and improve the quality of care.
- Ensure health professional and carers receive the information and education they need.

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<sup>17</sup> <http://www.nsd.scot.nhs.uk/about/nssc.html>

As such, national clinical and diagnostic networks directly support more than a quarter of all commitments within the UK Rare Disease Strategy – 1-5, 7, 8, 11, 12, 15, 18, 23, 32).

National Networks in Scotland		
For children & adults	For Children	Diagnostic
Burns	Epilepsy	Biochemistry
Inherited Metabolic Disease	Endocrine	Clinical Imagining
Cleft Lip and Palate	Cystic Fibrosis	Microbiology & Virology
Familial arrhythmias	Allergy	Pathology
Phototherapy	Rheumatology	Cancer
Gender identity	HIV/PID	- Sarcoma
Acquired brain injury	Exceptional Healthcare Needs	- Adult Neuro Oncology
Haemoglobinopathies	Disorders of Sex developments	- Pancreatic/hepatobilia
Diaphragmatic Hernia	Renal & Urology	
Inherited bleeding	Visual Impairment	
Perinatal Mental Health		
Neuromuscular Disorders		

Access to individual network websites for information and guidance is available on the NNMS website at <http://www.mcns.scot.nhs.uk/>

Guidance on the role of networks was set out by the Scottish Government in the CEL 29 (2012) "Managed Clinical Networks: Supporting and Delivering the Health care Quality Strategy". This recognised the important role of networks in influencing service re-design, quality improvement, strategy and planning across pathways that may reach across health boards, sectors and agencies. It established a set of core principles for networks that describes some of the governance and structural requirements for networks and outline some specific areas for networks to address, including:

- Working to a documented evidence base. Often for national networks this will be through the development of pathways and guidelines based on the evidence available, the knowledge and experience of the specialist clinicians and the experiences of the patients and their families. This will be complemented by a programme of audit and quality improvement within the network to support the delivery of high quality care.
- Optimising the education and training potential of the networks. Sharing knowledge across Scotland is a key role for national networks and they will develop an education strategy to ensure there are appropriate opportunities available at the right level for the different groups of professional staff within the network and for patients and carers. Examples of ways this may be delivered are through national conferences, online learning, local roadshows and multi-disciplinary case discussion meetings.

- Engaging stakeholders, particularly patients and their families or carers. National networks where possible look to include patients and their representatives as active members of the network alongside clinical stakeholders and try to ensure good communication with all. In addition, national networks will employ a variety of methods to engage with patients and families including through patient and family events, online or clinic questionnaires, newsletters, social media and websites. What matters to patients and families strongly influences the improvement priorities of the network.
- A defined structure setting out the points at which the service is to be delivered and the connections between them. National networks will map out the services available for patients and families across Scotland for the area they support and look to influence any potential improvements in the pathways of care.

On 24 October 2016, the NNMS held an event for health care professionals and partners. Through a programme of presentation and workshops by network leaders and experts, the event focussed on forging relationships to maximise and share the impact of the national networks on health and social care.

More information on the progress of new NMCNs and pathways can be found below at section 3.1.

## **1.11 Health and Social Care Standards**

In 2014, Ministers committed to a review of the Care Standards with the aim of developing new standards capable of being applied across both health and social care services. The new standards 'Health and Social Care Standards: My support, my life'<sup>18</sup> which come into effect on 1 April 2018, will be applied to a diverse range of services from daycare for children, housing support and care at home for adults, to hospitals, clinics and care homes. They will apply to the NHS as well as services registered with the Care Inspectorate and HIS. This sets out the standards that people should expect when using health and social care services.

The Standards are focused on improving people's experience of care and are based on the following outcomes:

- I experience high quality care and support that is right for me.
- I am fully involved in all decisions about my care and support.
- I have confidence in the people who support and care for me.
- I have confidence in the organisation providing my care and support.
- I experience a high quality environment, if the organisation provides the premises.

The Standards are underpinned by five principles: dignity and respect, compassion, be included; responsive care and support and wellbeing; which reflect the way that everyone, including people with rare diseases should expect to be treated. These standards help towards meeting Commitments 3, 4 and 5.

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<sup>18</sup> <http://www.gov.scot/Publications/2017/06/1327>

## 1.12 Provision of Communication Equipment and Support



In March 2016, the Scottish Parliament passed Part 4 of the Health, (Tobacco, Nicotine etc. and Care) (Scotland) Act 2016<sup>19</sup> on the Provision of Communications and Equipment and Support. This gives children and adults across all age ranges and care groups, who have lost their voice, are at risk of losing their voice, or who have difficulty speaking, a statutory right to access the communication equipment and support they need. The legislation gained Royal Assent in April 2016 and is not specific to particular health conditions.

The legislative duty has not commenced as Directions to support the implementation of the legislation are being prepared for the NHS and other key service providers.

Discussions are also taking place with the Scottish Government legal colleagues and Health and Social Care Integration policy colleagues to determine the legal responsibilities and functions that Integrated Joints Boards have before the full legislative process can commence. Although the legislative duty is not complete, specialist augmentative and alternative communication services are being, and continue to be, provided across Scotland.

This legislation addresses Commitments 2 and 4 of the UK Strategy for Rare Diseases. It is an important step forward for people with progressive rare diseases such as Parkinson's Disease, Motor Neurone Disease, and Huntington's disease. It will allow people to be able to communicate and have their "voice" heard when they are no longer able to speak or have difficulties speaking, ensuring that they receive the care and support that they want and need.

## 1.13 Frank's Law

The First Minister, during her Programme for Government (PfG) announcement on 5 September 2017, announced that the Scottish Government would, over the next year begin work to fully implement Frank's Law<sup>20</sup>, by extending Free Personal Care to those aged under 65 who are assessed as needing this service, regardless of their condition. Extending Free Personal Care to people under 65 will benefit around 9,000 people in Scotland.

<sup>19</sup> <http://www.legislation.gov.uk/asp/2016/14/contents/enacted>

<sup>20</sup> <https://news.gov.scot/news/franks-law>

## **1.14 National Patient Portal**

The Scottish Government has commissioned a programme of work to advance the development of a national patient portal which will benefit all people in Scotland, but this will also help to meet Commitment 6 and 19 in the UK Strategy for Rare Disease. The programme is currently working on the delivery of a technical proof of concept, developed in collaboration with an agreed supplier, as well as an outline business case. This work is expected to be concluded by March 2018 in line with the existing aim of launching a portal with initial functionality by 2020. This initial functionality, which would be subsequently expanded, is likely to include access to a summary patient record, online GP services, electronic communications and personalised health information for every citizen in Scotland.

People with rare diseases often have to attend a number of different clinics in order to manage their treatment. This can also include having numerous tests carried out or providing various clinicians with information. The national patient portal will allow people with a rare disease and their carer/family access to information such as appointment letters, access to GPs or other services, a summary of their information and some as well as personalised health information, for example, some test results. This will give the person more flexibility in managing their condition and being able to access services when they need it.

## **1.15 Scottish Genetics Speciality Group**

The role of NHS research is to support the delivery of clinical research in Genetics. It also manages participant recruitment ensuring that they are recruited in time and on target. The research includes Scottish led studies and ones which Scotland is participating in. At the moment there are approximately 30 studies and about one third of these are led from Scotland.

The Genetics Speciality supports the delivery and promotion of clinical research in a wide range of areas, including:

- rare diseases;
- causes and prevention of birth defects;
- common disorders such as familiar cancer; and
- genetic approaches to treatment and prevention.

In terms of rare diseases, the national approach made possible by the National Institute for Health Research (NIHR) Clinical Research Network Portfolio, and supported by the Scottish Genetics Speciality Group means that together, they are able to collect enough people with a particular condition to make research more feasible. This approach provides more opportunities for people with rare genetic diseases to participate in research, which will lead to improved care and the development of new treatments.

A number of studies have been available to people with a rare diseases, but the majority to date, have been observational (understanding the cause and natural history of a rare disease). However, there are a small number of interventional studies – there is Scotland-wide participation in the Association for Improvements in the Maternity Services (AIMS) study for Marfan syndrome and the Cancer Prevention Programme3 (CaPP3) for Lynch syndrome.

Other studies which have been carried out and the outcomes of which are due to be implemented are:

### **The DDD Study**

The Deciphering Development Disorders (DDD) study aimed to find out if using new genetic technologies could help doctors understand why patients get developmental disorders. All of the Scottish Genetic Centres participated in the study, recruiting some 1400 participants. The outcome demonstrated a three-fold increase in diagnosis using Exome enriched Next Generation Sequencing compared to using conventional technology. Previous figures showed that 9% of patients obtained a successful diagnosis using the conventional methods, but by using Exome enriched Next Generation Sequencing, 27% of participants received a diagnosis for a rare development disorder.

The Scottish Genetics Laboratory Consortium (see Section 4.4) is planning to implement this as standard across Scotland, however, there are serious cost implications attached to the test, and NHS Boards have to manage this along with all the other services that they offer. As a result, Scotland is not as far advanced as their English counterparts in implementing this, nevertheless, the Consortium is aware that this is the only test that will lead to a diagnosis in many children with severe developmental disorders and are looking into the matter very carefully.

### **Meaning**

#### **Exome enriched Next Generation Sequencing**

Next Generation Sequencing is the term used to describe modern technology that enable scientists to sequence DNA faster and more cheaply than previous sequencing techniques. It has revolutionised the study of genomics, by allowing us to sequence every gene in a person. This is particularly important as many rare diseases are caused by faults in genes that can only be detected using this technology.

## **The RAPID Study**

The RAPID study Dundee participated in, demonstrated the clinical utility and cost saving of non-invasive prenatal testing (NIPT) for chromosomal trisomies compared to conventional technology which requires amniocentesis.

The new form of testing is non-invasive as it only requires a blood sample being taken from the mother. It can be carried out at 10 weeks of pregnancy and there is no chance of miscarriage. The test uses fragments of fetal DNA, that is found in the mothers blood stream during pregnancy, to check for Trisomies. Trisomies means that there are three copies of one particular chromosome, instead of two (one from each parent), examples of Trisomies include Down syndrome, Edwards Syndrome and Patau syndrome.

Following participation in this study, Dundee piloted this approach demonstrating a cost saving and reduced risk to pregnancies. The Scottish Screening Committee is now overseeing the implementation of this and have commissioned a business case from NHS National Services Division, which was considered at their meeting in November 2017. Next steps involve seeking agreement from Directors of Finance and Board Chief Executives on the investment required to make this available across Scotland.

For more information about NIPT visit Antenatal Results and Choices: <http://www.arc-uk.org/tests-explained/non-invasive-prenatal-testing-nipt>

The Scottish Genetics Speciality Group procedures help towards meeting Commitment 7 and 8 of the UK Strategy for Rare Diseases.

## **2. IDENTIFYING AND PREVENTING RARE DISEASES (9 – 10)**

<b>Commitment</b>	<b>Description</b>
9	Continue to work with the UK NSC to ensure that the potential role of screening in achieving earlier diagnosis is appropriately considered in the assessment of all potential new national screening programmes and proposed extensions to existing programmes.
10	Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular tests are evaluated and recommended by UK Genetics Testing Network (UKGTN) is accessible for at risk relatives.

### **2.1 The Scottish Screening Committee**

The Scottish Screening Committee (SSC) was established in 2016 to improve the strategic management of screening services in Scotland.

The primary objective of the SSC is to consider advice and recommendations which flow from the UK National Screening Committee<sup>21</sup> (UK NSC) - an independent expert advisory group which advises Ministers and the NHS in the four UK countries about all aspects of screening. The role of the SSC is to consider UK NSC recommendations in the context of the specific Scottish circumstances.

Following Ministerial approval, the SSC nationally commissions the implementation of new screening programmes or changes to existing programmes and provides high level oversight of all development and performance in the Scottish screening programmes, based on reporting from National Services Division.

The SSC currently oversees six national screening programmes:

- Abdominal Aortic Aneurysm (AAA) screening
- Bowel Cancer Screening
- Breast Cancer Screening
- Cervical Cancer Screening
- Pregnancy and Newborn Screening
- Diabetic Retinopathy Screening

<sup>21</sup> <https://www.gov.uk/government/groups/uk-national-screening-committee-uk-nsc>

With regards to rare diseases which are covered within the remit of the SSC, Newborn Blood Spot Screening<sup>22</sup> identifies babies who may have rare but serious conditions. The programme includes screening for:

- Sickle cell disease (SCD)
- Cystic fibrosis (CF)
- Congenital hypothyroidism (CHT)
- Phenylketonuria (PKU)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
- Maple Syrup Urine Disease (MSUD)
- Isovaleric Acidemia (IVA)
- Glutaric Aciduria type 1 (GA1)
- Homocystinuria (HCU)

The UK NSC is currently assessing a number of other rare conditions that could potentially be included in the NHS Newborn Blood Spot Screening Programme. As with all screening programmes, these would only be introduced if it is clear that the benefits to newborn babies outweigh any potential harm.

## 2.2 Introduction of new screening tests



Following a review conducted by the UK NSC, the Scottish Government extended the Newborn Blood Spot Screening programme to test for Homocystinuria (HCU), Maple Syrup Urine Disease (MSUD), Glutaric Aciduria type 1 (GA1), and Isovaleric Aciduria (IVA) to all newborn babies up to one year old, extended from six months old.

The four additional conditions are very rare. They typically occur in between 1 in 100,000, and 1 in 200,000 births. In general, early dietary-based treatment for these conditions is effective. If untreated, babies with MSUD, IVA and GA1 can become suddenly and seriously ill, while symptoms of HCU can take up to one or two years to emerge.

Prior to the introduction of the new four tests, babies in Scotland were screened for Phenylketonuria (PKU), Congenital hypothyroidism (CHT), Sickle cell disease (SCD), cystic fibrosis (CF) and Medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

Since the new screening programme began on 20 March 2017, 20,748 babies have been tested, with 24 samples referred for either further testing or clinical referral.

22 <http://www.nsd.scot.nhs.uk/services/screening/newbornscreening/index.html>

## 2.3 UK Genetic Testing Network



The United Kingdom Genetics Testing Network (UKGTN)<sup>23</sup> evaluates genetic tests for scientific validity and clinical utility to ensure that all tests undertaken by member laboratories are evidence based. The Scottish Genetics Laboratory Consortium Molecular and Cytogenetic laboratories<sup>24</sup> are currently members of the UKGTN, and as such can provide genetic services for the NHS throughout the UK. It is the expectation of commissioning arrangements, that Consortium laboratories (in relation to molecular genetics and cytogenetics) only undertake UKGTN (or for future nationally approved alternative) approved tests thus ensuring all tests undertaken are evidence based.

NHS England have recently taken over responsibility for the UKGTN. As part of the reconfiguration of NHS England genetic laboratories, a consultation on the future of the UKGTN evaluation process is anticipated in early 2018. In the meantime, NHS England is consulting with Scotland and other devolved nations on the best way forward.

There is currently no UKGTN equivalent body in respect of the evaluation of molecular pathology testing. Therefore, as part of the implementation of the nationally commissioned service, the Molecular Pathology Evaluation Panel (MPEP)<sup>25</sup> was established to support the introduction of new tests and review and evaluate established tests. MPEP makes recommendations to the Molecular Pathology Consortium<sup>26</sup> on the clinical utility and validity of tests that should be provided on a national basis. This framework is based on UKGTN Test Evaluation Process and Scottish Medicines Consortium submission process.

NSD has established the Genetics Evaluation Panel for Scotland to complete the initial assessment of test requests (in anticipation of the changes to UKGTN), and to provide clinical advice to the Genetics Consortium on the most appropriate tests to be made available in Scotland.

<sup>23</sup> <https://ukgtn.nhs.uk/>

<sup>24</sup> <http://www.nsd.scot.nhs.uk/services/specserv/molgen.html>

<sup>25</sup> <http://www.mpep.scot.nhs.uk/>

<sup>26</sup> <http://www.pathology.scot.nhs.uk/molecular-pathology-consortium/>

To ensure appropriate carrier testing can be rolled out, examples of ongoing work to assess the clinical utility of carrier testing for patient groups are as follows:

### **BRCA testing in serious ovarian tumours**

NHS Grampian launch the introduction of the somatic BRCA1/2 service; Testing of the genes BRCA1 and BRCA2 in individuals affected with breast/ovarian cancer and their family members has been available as a blood test in Scotland for a number of years. Finding a cancer causing sequence difference (mutation) in a patient's blood (the germline) indicates future risks of developing additional cancers, is invaluable for guiding preventative treatments for members of the family and informs treatment options.

In particular, women with ovarian cancer who have a germline BRCA1/2 mutation can benefit from treatment with a new type of drug therapy called PARP inhibitors. Recent research has shown that even when a woman with ovarian cancer does not have a germline BRCA1/2 mutation detectable in her blood, if such an alteration is found in the tumour itself – an acquired ‘somatic’ BRCA1/2 mutation - she may benefit from treatment with PARP inhibitors. Due to the benefits described above, somatic BRCA1/2 testing is now available in Scotland for selected patients who might benefit from the test.

### **Primary Immune Deficiency (PID) Genetic Service**

Following discussions with the Scottish Paediatric and Adolescent Infection and Immunity Network (SPAIIN) and agreement from NSD, a new service has been introduced in the Scottish Genetics Laboratory Consortium. The testing for PID is delivered by the Aberdeen Laboratory and aims to improve the treatment of children and young people with PID through the development of genetic testing. This testing will enable equity of access for Scottish patients to genetic testing, leading to improved diagnosis, care, and treatment.

### 3. DIAGNOSIS AND EARLY INTERVENTION (11 – 22)

Commitment	Description
11	<p>Work to achieve reduced times for diagnosis of rare diseases, whilst acknowledging that more needs to be done to ensure that undiagnosed patients have appropriate access to co-ordinated care e.g. to help disabled children who are thought to have a genetic syndrome or condition that science has not yet identified.</p>
12	<p>Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to , and understood by, professionals and patients, by:</p> <ul style="list-style-type: none"> <li>• establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and specialist clinical centres, as appropriate;</li> <li>• putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment;</li> <li>• drawing on patients' ability to help inform decisions about referral and diagnosis;</li> <li>• creating effective clinical networks to support this process;</li> <li>• making high quality diagnostic tests accessible through common, clinically agreed systems or pathways; and</li> <li>• embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies.</li> </ul>
13	<p>Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.</p>
14	<p>Where appropriate, support the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered.</p>
15	<p>Improve education and awareness of rare diseases across the healthcare professions, including:</p> <ul style="list-style-type: none"> <li>• involving patients in the development of training programmes;</li> <li>• encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics; and</li> <li>• ensuring awareness of methods and clinical techniques used in differential diagnosis.</li> </ul>
16	<p>Monitor the development of ICD-11 in preparation for its adoption.</p>
17	<p>Work with colleagues in Europe in the development of the European Orphanet coding system and consider the adoption of Orphanet coding and nomenclature.</p>

<b>Commitment</b>	<b>Description</b>
18	Standardise data collection, building on existing NHS data standards, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.
19	Explore options to improve the link between existing patient data and electronic health records.
20	Assess the potential for rare disease databases where they do not exist.
21	Agree international standards, building on existing UK standards.
22	Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally.

### **3.1 National Managed Clinical Networks & Pathways**

As noted in section 1.9 the National Managed Clinical Networks<sup>27</sup> have a key role to develop patient pathways and clinical protocols, often for rare disease conditions. Following from the last biennial report, National Services Division has been supporting the development of pathways for a number of rare diseases. This work has involved patients and clinicians mapping existing services, highlighting gaps and facilitating solutions for identified issues. The common themes from the five rare diseases looked at (Acute Porphyria, Atypical haemolytic-uremic syndrome (aHUS), Neurofibromatosis type 2, Vasculitis, including Behçets, and Ehlers Danlos Syndrome) are access to specialist services and drugs as well as the coordination of care for complex decisions.

The updates below demonstrate the progress made and challenges faced:

#### **Acute Porphyria**

##### **Explanation: What is Acute Porphyria?**

Porphyria is the name given to very rare metabolic disorders that occur when your body is unable to produce enough haem. Haem is a component of haemoglobin which helps transport oxygen around your body and helps form many other important proteins found in all body tissues, but mostly in red blood cells, bone marrow and liver.

Acute Porphyria are Acute Intermittent Porphyria (AIP). It is the most common form in the UK and the most severe. It can cause serious damage to the liver and kidneys, and in some cases the person may need a liver transplant. Attacks or seizures can also lead to complications such as respiratory failure and can be fatal if not treated.<sup>28</sup>

The overall objective of this work was to improve the clinical management and care of NHSScotland patients with AIP who experience an attack or attacks. This involves providing immediate specialist clinical advice to acute care physicians; as well as clinically appropriate support on follow-up after discharge via telephone advice and the delivery of collaborative outreach clinics.

<sup>27</sup> <http://www.mncs.scot.nhs.uk/types-of-network/national-networks-in-scotland/nmcns/>

<sup>28</sup> <https://www.britishlivertrust.org.uk/liver-information/liver-conditions/porphyria/>

The outcomes are as follows:

- Scottish Clinicians and patients now have access to NHS England's National Acute Porphyria Service (NAPS).
- A service specification has been agreed between NHS England, NHS Scotland National Services Division and the NAPS.
- NAPS Scotland has been operational since April 2016, giving clinicians and patients in Scotland access to specialist advice during acute episodes and access to specialist follow-up in Scotland.
- Work is on-going with NHS Lothian to arrange six monthly NAPS outreach clinics for Scottish patients.

### **Atypical haemolytic-uremic syndrome (aHUS)**

#### **Explanation: What is Atypical haemolytic-uremic syndrome (aHUS)**

The aHUS website describes aHUS as a very rare disease caused by a fault in the complement system. The complement system is part of the body's immune response. Your body has a built-in system of protector proteins that stop complement from attacking your own cells. In aHUS, complement starts to attack your own cells, especially those that line the blood vessels, and this leads to clots forming within the small vessels.

The most commonly affected organ is the kidney, but all organs can be affected. If aHUS is not treated it can be life-threatening and the majority of people would develop end stage kidney failure.<sup>29</sup>

The overall aim for this project is to enable access for NHSScotlandclinicians to expert diagnostic and management advice from the aHUS service regarding treatment, including the prescription and withdrawal of Eculizumab. This pathway includes access to confirmatory genetic testing.

The outcomes are as follows:

- It was agreed that NSD should seek access to the aHUS service commissioned by NHS England, similar to the agreement for Acute Porphyria.
- It is proposed that the decision to commence a patient on Eculizumab would continue to be subject to an IPTR request made to patients' health board of residence; however, the assessment for starting a patient on this drug should be carried out by the specialist aHUS service in Newcastle.
- A service specification and pathway has been developed with clinical input outlining which service components should be delivered in Scotland and the specialist centre respectively.
- Access to the aHUS service in Newcastle to be added to the Block Service Agreement with NHS England.

29 <http://www.atypicalhus.co.uk/ahus-2/what-is-ahus/>

## **Neurofibromatosis type 2 (NF2)**

### **Explanation: what is Neurofibromatosis type 2?**

Neurofibromatosis is the general name for a number of genetic conditions that cause tumours to grow along the nerves.

Tumours are abnormal tissue growths. In neurofibromatosis, the tumours are usually non-cancerous (benign).

Almost everyone with NF2 develops tumours on the nerves responsible for hearing and balance.

Tumours can also develop inside the brain or spinal cord, or the nerves to the arms and legs. This can cause symptoms such as weakness in the arms and legs, and persistent headaches.<sup>30</sup>

Issues to be addressed by this work stream include improving the coordination of care, including access to Avastin, as well as improving access to specialist advice for patients in Scotland.

- NSD and the Managed Services Network (MSN) for Neurosurgery held a stakeholder meeting in August 2017 to review a draft pathway for Scotland. Ways to improve co-ordination of care to ensure access to appropriate specialist advice for all patients was discussed at the meeting.
- There was agreement at the meeting that all treatment decisions for people with NF2 should be subject to input from a specialist Multi-disciplinary team, and all patients should receive appropriate follow-up and monitoring.
- Work is on-going to set up a national Multi-disciplinary team, to agree a standard of care, which eventually could be audited, and to scope the need for a care co-ordinator.

## **Vasculitis (including Behcets)**

### **Explanation: What is Vasculitis?**

Vasculitis means inflammation of the blood vessels – any blood vessels in any part of the body can be affected. Vasculitis UK suggest that it affects about 2-3000 new people each year in the UK.

Acute Vasculitis can be caused by infections or by reactions to drugs/chemicals. This is usually quite localised (like a rash) and does not need treatment. Other types can be caused by other illnesses such as rheumatoid arthritis or some cancers.<sup>31</sup>

Others can be rare, such as Behcets disease, which can be difficult to diagnose as symptoms are so wide ranging. Symptoms can include genital and mouth ulcers, painful eyes/blurred vision, headaches, painful, stiff and swollen joints. In severe cases there can be life-threatening problems, such as vision loss and strokes.<sup>32</sup>

This work was commenced to address a perceived lack of access and clear signposting to specialist services for people with Behcets Disease in Scotland.

- A workshop with clinicians in October 2015 highlighted the substantive overlap between this condition and other forms of systemic Vasculitis as these are often managed by the same team of specialists.

30 <https://www.nhs.uk/conditions/neurofibromatosis-type-2/>

31 <http://www.vasculitis.org.uk/content/downloads/understanding-vasculitis-factsheet-june-2014.pdf>

32 <https://www.nhs.uk/conditions/behcets-disease/>

- The Short-Life Working Group (SLWG) on Vasculitis was set up by NSD in May 2016 to assess possible options for improving the care for adults with Vasculitis, by improving collaboration between all professionals involved in delivering care for this patient group across Scotland.
- The SLWG concluded that of the estimated 2250 adult patients in Scotland with Vasculitis, up to 75% have currently no access to specialist services.
- A draft report outlining the SLWGs recommendations is currently out for consultation.
- The SLWG concluded that a national managed clinical network (NMCN) or other officially endorsed network would facilitate access to specialist support and improve outcomes for all people who live with these conditions in Scotland.
- This would mirror developments in England where the roll out of Coordinated Networks for Specialised Rheumatology is being supported to improve patient care through the application guidelines, treatment protocols, and policies for access to high cost drugs.

### **Patient Involvement in Pathway Development**

The Scottish Plan for Rare Diseases committed to improving knowledge and appropriate care by developing and publicising of patient pathways for rare conditions and the referral requirement for services in and out of Scotland.

This work has been led by National Services Division, with collaboration where possible with patients and patient organisations. One such example is the development of a patient pathway for Vasculitis.

Vasculitis patient organisations have raised concerns about the difficulties in obtaining correct and timely diagnosis of Vasculitis and subsequently, difficulties in accessing referrals to appropriate care and treatment. To address these issues, the development of a patient pathway was considered.

In 2016, National Services Division established a SLWG to produce a patient pathway for Vasculitis and, importantly, the SLWG has included patient representation throughout the process.

Lynn Laidlaw, a patient with Vasculitis, has been involved in the SLWG and has said ‘the meetings that I have attended have been positive - I have been able to contribute and feel that the other members have both listened and had a genuine interest in what I have had to say’.

Lynn also stated that ‘it was good that patients have been involved in this process - patients often bring different perspectives to clinicians and offer insight to what would improve patient experience and outcomes.’

The patient pathway for Vasculitis development work has also led to the development of regular cross-Scotland clinical meetings to discuss service delivery and complex cases. These meetings will also include involvement from patient representatives, ensuring that the patient experience is reflected in its work.

## Ehlers Danlos Syndrome (EDS)

### Explanation: What is Ehlers Danlos Syndrome?

NHS Choices describe Ehlers Danlos Syndromes (EDS) as “a group of rare inherited conditions that affect connective tissue. Connective tissues provide support in skin, tendons, ligaments, blood vessels, internal organs and bones.

There are several types of EDS that may share some symptoms, including an increased range of joint movement; stretchy skin; and fragile skin that breaks or bruises easily.

EDS can affect people in different ways, for some the condition is mild, while for others their symptoms can be disabling. Some of the rare severe types can be life-threatening.”<sup>33</sup>

In 2016, it became apparent that there is, at present, a lack of clarity as to the referral and treatment pathways for those affected by EDS in Scotland. Patient feedback received also highlighted the difficulties faced when individuals try to access specialist support and care for this condition.

- A meeting took place in May 2017 to gather and review information regarding the patient needs and identify the current options for people with EDS in Scotland (and the UK). There was agreement at the meeting that the main issues were co-ordination of care and access to specialist advice for people who have been discharged from rheumatology or genetics clinics following a diagnosis of EDS.
- There was also agreement that expertise and services to manage the condition exist in Scotland. The main issues were lack of sign posting, referral pathways and overall co-ordination of care. NSD will produce a paper for regional directors of planning, highlighting what issues and service gaps people with EDS, and hypermobility spectrum disorder encounter.

## Huntington's disease (HD)

In addition to the work carried out by NSD, the Scottish Government has funded work on Huntington's disease and Motor Neurone Disease.

### Explanation: What is Huntington's disease?

The Huntington's Disease Association describe the disease as “an illness caused by a faulty gene in the person’s DNA. It affects the body’s nervous system – the network of nerve tissues in the brain and spinal cord that co-ordinate the body’s activities. Huntington’s can cause changes with movement, learning, thinking and emotions. Once the symptoms begin, the disease gradually progresses, so living with it means having to adapt to change.”<sup>34</sup>

In 2015 the Scottish Government commissioned the Scottish Huntington’s Association (SHA)<sup>35</sup> to lead on the development of a National Care Framework for Huntington’s disease (HD)<sup>36</sup>. £180,000 was provided to SHA in order for them to produce a National Framework by early 2017 and to begin work to develop localised versions for each NHS Board area by 2019.

A National Care Framework Lead was appointed, who pulled together a multi-disciplinary development group to devise and consult upon the Framework. The group included representatives of HD families and carers, psychiatry, psychology, neurology, neuropsychology, genetics,

<sup>33</sup> <https://www.nhs.uk/conditions/ehlers-danlos-syndromes/>

<sup>34</sup> <https://www.hda.org.uk/huntingtons-disease/what-is-huntingtons-disease>

<sup>35</sup> <https://hdscotland.org/>

<sup>36</sup> <http://care.hdscotland.org/>

rehabilitation, dentistry, GPs, speech and language therapy, dietetics, physiotherapy, occupational therapy, care homes, palliative care, social work, and academic and research institutions.

Further to a public consultation in Autumn 2016, the Framework was published in March 2017 and formally launched at an event in the Scottish Parliament in May that year. It can be viewed at: <http://care.hdscotland.org/>

The Framework seeks to ensure that the care and support provided to families living with HD takes account of their specific health & social care needs throughout their experience. It does not advocate a “single pathway” or “one-size-fits-all” approach, making clear that every person’s experience is unique and requires to be treated as such.

The Framework makes clear that individual care and support packages require to be created in partnership with families, and should have four key elements.

1. Care coordination should be provided by a single named specialist as determined by each Health & Social Care Partnership (HSCP) who has a key role in assisting families to navigate their way through their HD journey.
2. Care should be provided by a clearly defined multi-disciplinary team consisting of core members whose roles are essential in managing health and social care needs.
3. Access is required to a wider and clearly mapped network of services where well-developed referral and liaison arrangements are in place.
4. Specialist staff play a central role in providing training and education to the wider support network. Each HSCP area should have arrangements to deliver training, or work with other HSCPs to deliver training, appropriate to its own workforce.

The Framework advocates a Person Centred Approach, a Family Systems Approach, a Biopsychosocial Model of Health & Disability, Personalisation and a Palliative Care Approach.

The Framework itself is a flexible, interactive online resource designed to evolve along with changes to health and social care systems, structures and personnel – rather than a printed document that can quickly become obsolete. It provides a wealth of information and resources under the following 16 topics:

- Deciding whether to have the genetic test
- Finding out if you have symptoms of HD
- Getting help with the symptoms and associated difficulties of HD
- Getting information and advice about HD and someone to support you
- Support with employability when you need it
- Getting financial help when you need it
- Getting advice about having a family
- Getting support for children, young people and parents
- Support if you are a caregiver and getting a break from caring
- Getting support with day-to-day living and reducing isolation
- Getting support to adapt your home or live where you choose
- Support to plan for the future
- Moving to supported accommodation and long term care

- Care at the end of life
- Getting support out of hours
- Participation in research

SHA has started the first phase of the localisation work in NHS Ayrshire & Arran, NHS Fife & NHS Grampian. An analysis of local services compared to the Framework has taken place in each of these areas and Local Action Plans have been drafted. It is anticipated that localised versions of the Framework will be in place in these areas by autumn 2017, at which point localisation work will proceed in NHS Greater Glasgow & Clyde, NHS Lanarkshire and NHS Lothian.

People living with neuro-progressive disorders, like Huntington's disease, can face many common challenges and it is anticipated that much of the learning that will emerge from this work will be transferable to supporting people with other long term conditions.

The Framework has been well received by many stakeholders, including the Cabinet Secretary for Health and Sport:

**“From the outset we were hopeful that the HD Framework could have the potential to be used as a template for other neurological and long terms conditions. Widespread feedback from stakeholders representing such conditions appears to have borne this out. We look forward to seeing how this exciting prospect develops, and are pleased to continue working with the Scottish Huntington’s Association and others to make this a reality.”**

*Shona Robison MSP, Cabinet Secretary for Health and Sport*

The Nationally Managed Clinical Networks and pathways relate to Commitments 12, 23 and 32 of the UK Rare Disease Strategy. However, they also relate to Commitment 1 as they include patient groups as key partners in the development of the service and/or pathway.

## 3.2 Decision support tools for health and social care

The Scottish Government eHealth strategy 2014-2017<sup>37</sup> has a commitment to provide decision support tools. It states:

**Clinicians, Social Care Staff and other third sector partners** will be able to:

- have quick and easy access to increasing amounts of clinical guidance and decision support that is relevant to the specific patient context, including highlighting any substantial variation from expectations, and generating appropriate prompts and alerts.



In 2015, Scottish Government produced a Decision Support Roadmap to guide delivery on this commitment. This roadmap highlighted the need for a range of different types of decision support tools, including websites, mobile apps, and the most effective method - integrating prompts into the clinical and care systems that practitioners use on a day to day basis.

There are a number of decision support tools available some of which are purchased or developed for national or local use in NHS Scotland. Some mobile decision support apps and other tools are also available for practitioners to purchase independently or access freely through the Internet. A recent survey of what clinicians need from decision support tools confirmed that clinicians are overwhelmed by the vast range of tools available and that they would value a “go-to” place – a single delivery platform - for quality assured decision support tools that meet NHS Scotland standards. This survey also highlighted the value that clinicians place on decision support delivered through clinical systems and in the form

of downloadable mobile apps. These apps are valued for quick access to reliable guidance, algorithms and calculators, particularly in the community and other locations with limited Internet access. Examples of decision support apps developed through the Decision Support Roadmap include the Antimicrobial Companion and the app for sepsis diagnosis and management. Work will begin in 2018 to look into suitable apps for rare diseases.

In response to the needs identified through this survey and the earlier consultation on the Decision Support Roadmap, the Scottish Government Digital Health and Care Team is developing a quality assurance framework to give assurance that the tools promoted in NHS Scotland are safe and clinically effective. Initially this quality assurance framework will focus on mobile apps for health and social care professionals. The Scottish Government is also working with the Digital Health and Care Institute to create a delivery platform for quality assured decision support tools, linked to a common knowledge base to ensure that all tools are based on best available evidence.

The Scottish Government has also procured a decision support platform that can interact dynamically with data in an individual electronic patient record, run that data through algorithms based on best practice evidence and guidance, and push prompts and recommendations specific to that patient's needs to the clinician, in the context of the patient record. In the first instance, this platform will be evaluated working with primary care electronic health record systems, however it has the capability to interact with any clinical or care system which can transfer the relevant data to the decision support platform. This is an especially important development as the research evidence indicates that this is the form of decision support with the highest impact on healthcare practice.

In secondary care in Scotland, the limitations of the clinical systems such as Trakcare mean that this type of patient-specific decision support is usually harder to implement. A simpler alternative that has been introduced in TrakCare is a hyperlink from the Trakcare menu to a webpage that provides access to quick reference sources such as the British National Formulary, and collections of action-focused summaries of evidence provided by publishers such as BMJ.

While some of these support tools are relatively new, they offer opportunities to help people with a rare disease to get a faster diagnosis and receive fast and tailored treatment plans. In partnership with a range of stakeholders, the Scottish Government is developing a new, integrated Digital Health and Social Care Strategy that will build on achievements to date and set out future development and priorities

These decision support tools are the start of meeting Commitments 12, 14, 30 and 34 of the UK Rare Diseases Strategy.

### **3.3 Cross Border Guidance for Clinicians**

NSD manages the process by which NHS Boards in Scotland can access national risk share funding for referrals to specialist and highly specialist healthcare which is not available in NHS Scotland. Referral is normally to a NHS Trust in England which is recognised as a provider by the NHS England specialist care commissioners. In very rare circumstances, consideration will be given to funding highly specialised care within an internationally recognised healthcare provider overseas.

This ensures that patients in Scotland can access specialist treatment that may not be available in NHS Scotland. This is of particular importance to patients with very rare diseases, where there may be a limited number of expert clinicians in the UK able to offer diagnosis and advice.

NSD have updated the Cross Border Guidance for Clinicians in order to improve knowledge of services available. It has also published NHS England's Highly Specialised Services report in Scotland which gives detail on many of the newer services available. The report can be viewed on the NSD website at: <http://www.nsd.scot.nhs.uk/services/specialised/index.html>

### **3.4 Scottish Clinical Genetics Forum**

The Scottish Clinical Genetics Forum is the national group for Clinical Genetics in Scotland. It was created following the Calman Review of Clinical Genetics services. The purpose of the forum is to develop and share good practice, and develop policies from EU or UK guidelines (for example, EU guidelines for inherited cardiac conditions or NICE guidelines for managing the risk of breast cancer) to ensure that there is equal access of care for all.

The work of the Forum contributes to Commitment 15 of the UK Strategy for Rare Disease.

### 3.5 The Montgomery Review



On 14 December 2016, the Cabinet Secretary for Health and Sport announced that the Scottish Government will take forward the recommendations of Dr Brian Montgomery's independent Review of Access to New Medicines<sup>38</sup>. These reforms will help more patients get better access to the treatments they need.

Since the publication of the report, the Scottish Government has been working closely with relevant partners, stakeholders, patient representatives, the third sector and the pharmaceutical industry to consider the most appropriate and effective way to take forward the recommendations. A large amount of progress has been made.

#### Patient and Clinician Engagement in Decision Making

The SMC are working on developing public friendly summaries of their decisions that will give a brief explanation of the condition, medicine, decision and rationale. Additionally, the role of the public partner has been reviewed and clarified. Since August 2017, they are no longer required to present the patient group submissions. Patient group representatives have also been attending SMC meetings from July 2017. Further work will be done to consider clinician engagement in the SMC decision making process, as there is a need to strike a balance between the time required to attend an SMC meeting versus the time they spend in the clinical environment.

#### Transparency of SMC decisions

The role of the secret ballot has been reviewed. It was felt that this process was necessary and should continue in order to protect the confidentiality of both individuals and SMC decisions until they are officially released.

SMC will also make decisions 'not to accept' medicines clearer on their website.

The Association of the British Pharmaceutical Industry (ABPI) Scotland has led on minimising the inclusion of commercial in confidence information in SMC submissions. The preference of the ABPI is to encourage optimal public disclosure whilst appreciating an individual company's need to redact genuine in confidence information. They have therefore modified existing NICE guidance regarding the management of confidential data to match SMC's requirements. The intention is to have this completed in early 2018.

#### End of life, orphan and ultra-orphan medicine

The SMC has reviewed the definitions of end of life, orphan and ultra-orphan medicines. A new pathway is being developed, and an announcement will be made in early spring 2018 on any revisions to the definitions and the ultra-orphan pathway.

<sup>38</sup> <http://www.gov.scot/Publications/2016/12/9192/0>

## **Managed Access Schemes and Conditional Acceptance Options**

Work is underway to enable the SMC to have an additional decision option of “recommend for use, subject to on-going evaluation and future assessment”. This will allow the SMC to accept a medicine for a time period to allow the collection of real world data on its effectiveness. It has been agreed this would be difficult to apply to all medicines currently due to limited IT capabilities, including the associated administrative/clinical burden of collecting outcome data. It should therefore be tested in the first instance to those medicines that are applicable only for a conditional acceptance. Initially, this will encompass medicines who have received a conditional marketing authorisation (CMA) by the Europeans Medicines Agency (EMA).

Additionally, NHS National Procurement are working closely with the SMC to progress plans on conditional acceptance options as well as how Managed Access Schemes can be adopted early in NHS Scotland. In the first instance Managed Access Schemes will only apply to medicines that have received a conditional acceptance as defined above.

## **Learning from other Health Technology Assessment process**

The Scottish Government has commissioned a comparative review of the health technology assessment (HTA) processes across the world, including end of life, orphan and ultra-orphan medicines. Once completed, the output from this work will assist in delivering recommendations 27 and 28 of the review, which are associated with new developments such as the development of a Scottish Model of Value.

## **Remaining Recommendations**

There are a number of recommendations such as the implementation of the Single National Formulary; the standardisation of data collection for non-formulary requests (recommendations 15 & 19); the future arrangements for the New Medicines Fund (r16); making better use of negotiations, including a pause in the HTA process (r22 & 24); and the development of a new Scottish Model of Value (r27 & 28) all of which are either reliant on the alignment of a number of other recommendations or linked to other policy timelines. Work is in hand to take all of these forward and the Cabinet Secretary for Health and Sport will provide an update to the Health and Sport Committee in spring 2018.

## **Peer Approved Clinical System (PACS)**

Alongside the publication of the Montgomery Review in December 2016, the Cabinet Secretary for Health and Sport also announced changes to the Individual Patient Treatment Request (IPTR) process for patient access to medicines not recommended for routine use in NHSScotland on an individual basis.

These new changes will introduce a new National Appeal Panel, refreshed guidance and standardised national paperwork.

IPTRs will be replaced with PACS Tier Two which is due to be implemented nationwide from 1 February 2018. In October 2015, Tier One PACS for ultra-orphan medicines was piloted in Glasgow. This pilot was rolled out to all other Health Boards across 2016, this will be evaluated to ensure it is achieving its intended aims, in line with recommendation 20 of the review. This will align with the implementation of the new ultra-orphan pathway.

The work being undertaken in this area may benefit people with rare diseases greatly in terms of the medication that is available to them. It also goes towards meeting Commitment 13 of the UK Rare Disease Strategy.

### **3.6 Data Short Life Working Group**

In July 2016, a Short Life Working Group (SLWG) was established by the Rare Disease Implementation Oversight Group to consider how information on rare diseases can be captured in Scotland to enable monitoring and reporting.

The SLWG produced a report in June 2017 that included a recommendation to establish a congenital anomalies register in Scotland. Scotland does not currently have a register.

Congenital anomalies comprise a wide range of abnormalities of body structure or function that are present at birth and are of prenatal origin. Congenital anomalies account for approximately 80% of all rare diseases and affect approximately 2-4% of pregnancies in Scotland.

A congenital anomalies register would:

- provide a resource for clinicians to support high quality clinical practice;
- provide epidemiology and monitoring of the frequency, nature, cause and outcomes of congenital anomalies;
- support research into congenital anomalies and precision medicine including basic science, cause, prevention, diagnostics, treatment and management;
- inform the planning and commissioning of public health and health and social care provision; and
- provide a resource to monitor, evaluate and audit health and social care services, including the efficacy and outcomes of screening programmes.

This proposed method is similar to that used by Public Health England's National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)<sup>39</sup>. NHS Information Services Division will liaise with NCARDRS to gain further understanding of how this works in NHS England.

A copy of the summary report from the SLWG can be found at Annex 2.

The work going forward from the SLWG will help to meet Commitments 7, 16 to 22 and 31 of the UK Strategy for Rare Diseases.

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<sup>39</sup> <https://www.gov.uk/guidance/the-national-congenital-anomaly-and-rare-disease-registration-service-ncards>

### 3.7 RARE Best Practice

#### Managing knowledge to improve care of people with rare diseases



HIS was a partner in the EU funded RARE-BestPractices<sup>40</sup> research project with responsibility for developing two information databases – a database of research gaps RAREGAP and a database of appraised clinical guidelines covering 44 disease areas RAREGUIDELINE <http://www.rarebestpractices.eu/>. This work highlighted the need to improve awareness of access to, and use of existing guidance on the diagnosis and management of rare diseases across those caring for people with rare conditions.

In order to maximise the awareness and application of knowledge and information available to support practice and education regarding rare diseases within Scotland, HIS together with NHS Education for Scotland (NES) are scoping the potential development of a Managed Knowledge Network for Rare Diseases.

A survey will be distributed to a number of clinical communities in Scotland, including general practitioners and paediatric specialists, to assess where clinicians look for and

source information on rare disease management to support their practice. Intelligence collected via this survey, together with a review of current sources of rare disease information, will be collated into a report.

Provision of information alone is not sufficient to ensure use or practice change, and work previously undertaken by HIS and NES on approaches to getting knowledge into action will be used to develop a series of recommendations on what might work best in terms of rare disease information provision for clinicians in Scotland.

The work carried out on the RARE-BestPractice project goes towards meeting Commitments 14 and 22 of the UK Strategy for Rare Diseases.

## 4. CO-ORDINATION OF CARE (23 – 30)

Commitment	Description
23	<p>Continue to develop service specifications for rare diseases. This will include country specific care pathways and a ‘generic’ care pathway that sets out best practice that can be applied to all patients with rare diseases in the UK (particularly where there are no disease specific pathways). The generic care pathway will include:</p> <ul style="list-style-type: none"> <li>• an appropriate care plan for all patients with rare disease;</li> <li>• clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis;</li> <li>• the development of seamless pathways for transition, from childhood to adolescence, and onto adulthood and older age; and</li> <li>• access to criteria and measures of quality and outcomes.</li> </ul>
24	<p>Agree that specialist clinical centres should, as a minimum standard:</p> <ul style="list-style-type: none"> <li>• have a sufficient caseload to build recognised expertise;</li> <li>• where possible, not depend on a single clinician;</li> <li>• co-ordinate care;</li> <li>• arrange for co-ordinated transition from children’s to adults’ services;</li> <li>• involve people with rare conditions, and their families and carers;</li> <li>• support research activity; and</li> <li>• ensure their expertise is available to families and their healthcare teams.</li> </ul>
25	<p>Ensure that the relationship between the specialist clinical centres and science and research is explained to and understood and put into practice by: practitioners delivering local health and social care; the research community; industry; academia.</p>
26	<p>Set out clearly the connections to, and communications with Specialist Clinical Centres in molecular diagnostics and other forms of diagnostic support.</p>
27	<p>Ensure that specialist clinical centres are as concerned with research as with health and social care support, and that they develop networks that provide professional-to-professional dialogue and collaboration across a wide range of experts, including internationally (especially for those conditions that are ultra-rare).</p>
28	<p>Work with international partners wherever possible and develop UK-wide criteria for centres to become part of an expert reference network to increase flow of information between patients and professionals in a range of disciplines.</p>
29	<p>Improve systems to record genetic and other relevant information accurately to detail the incidence and prevalence of disease and to support service planning and international planning.</p>
30	<p>Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments.</p>

## 4.1 European Reference Networks

Some rare diseases can affect just one or two people in the whole of a country, which can make it difficult for clinicians and people to get information, support and treatment.

The European Commission is required to support Member States in the development of European Reference Networks<sup>41</sup>, or ERNs.

The Second Report from the UK Rare Disease Policy Board contains the following section on ERNs:

- The total number of patients who collectively suffer from diseases classified as rare is significant. However, in any given country the number of patients who suffer from a specific rare disease may be small. The scarcity of rare disease expertise and patients in any single country means that diagnosis, treatment and management of rare diseases strongly benefit from cross-border collaboration.
- ERNs are centres of knowledge, skills and expertise in the field of rare diseases and complex conditions. They function as virtual networks that provide a platform to create partnerships between healthcare providers. They enable the principles of better access for patients to highly specialised services and support European co-operation on highly specialised healthcare, knowledge sharing and improved diagnosis and care in medical domains where expertise is limited.
- ERNs can also be focal points for medical training and research. The diagnosis, treatment and management of rare diseases require the highest level of partnership working to remove unnecessary barriers and facilitate access to high quality care and treatment.
- There are currently 24 ERNs across the EEA (European Economic Area), with 26 Member States (MS) participating, creating a large network of over 300 highly specialised healthcare providers (HCPs).

The UK is heavily involved by:

- participating in 23 of 24 networks involving 113 separate UK groups in this initiative of pan-European action on rare diseases; and
- leading six networks (more than any other Member State).

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<sup>41</sup> [https://ec.europa.eu/health/rare\\_diseases/european\\_reference\\_networks\\_en](https://ec.europa.eu/health/rare_diseases/european_reference_networks_en)

Scotland is contributing as a partner organisation to the following ERNs

ERN	CODE	NHS BOARD/HOSPITAL
Rare Bone Diseases	BOND	Greater Glasgow & Clyde
Rare Craniofacial anomalies & ENT Disorders	CRANIO	Tayside
Rare Endocrine Conditions	ENDO	Greater Glasgow & Clyde
Rare and Complex Epilepsies	EpiCARE	Greater Glasgow & Clyde
Rare Respiratory Diseases	LUNG	Ninewells Hospital & Medical School, Dundee. Royal Infirmary of Edinburgh
Rare & Undiagnosed Skin Disorders	SKIN	Tayside

A full list of the ERNs can be found at Annex 3.

The work of the ERNs goes towards meeting Commitment 28 of the UK Strategy.

## 4.2 Specialist Care in the UK

Through National Service's Division (NSD) and the National Specialist Services Committee, NHS Scotland receives and considers application for new specialist services and networks. NSD currently scans for:

- the impact of new medical technologies for existing specialist services;
- the need to develop pathways for access to new specialist services in England; and
- the need to commission new specialist services or Nationally Managed Clinical Networks in Scotland.

Of particular relevance to the Rare Disease agenda is the proposed development of Inherited Metabolic Disease Service in Scotland. The term Inherited Metabolic Diseases (IMD) covers a group of over 650 individual conditions, each caused by a defective single enzyme or transport protein. Individually each condition is rare; collectively IMDs are a significant cause of morbidity and mortality. Early identification and introduction of specialist diet or drug treatments is crucial as patients otherwise face severe health complications. Without treatment many IMDs can lead to severe learning or physical disability and death at an early age.

Around 1000 people in Scotland attend services for IMD. Due to the rarity of individual metabolic conditions and their complex nature, treating IMDs requires an integrated specialised clinical and laboratory service to provide satisfactory diagnosis and management.

Over 2016, a review of the IMD services in Scotland highlighted the need to assess the sustainability of the current provision of services in Scotland on account of:

- The greater number of IMD patients surviving into adulthood. The majority of the over 1000 people with IMDs known to the service in Scotland are 16 years of age or over.

- The impact of orphan drugs and the potentially significant increases in patients who meet clinical criteria for use of these drugs.
- The implementation of the UK Strategy for Rare Diseases.

The Expert Review Group recommended the national designation of an integrated IMD Service for Scotland. An outline application for a nationally commissioned IMD service was endorsed by the NSSC in June 2017. A full application is expected to be considered the first half of 2018. This work helps towards meeting Commitment 23 of the UK Strategy for Rare Diseases.

### **4.3 Specialist Centres**

In Scotland, all organisations involved in the care of patients with rare diseases should work in an integrated way. NSD works with NHS boards and other highly specialised service commissioners across the UK to ensure that patients have access to appropriate treatment across the UK. All specialist centres have a detailed specification which outline the expected service to be delivered and are reviewed regularly by NSD in Scotland and by counterparts for services accessed in NHS England. Any new specialist services would be expected to adopt the same principles. In addition, all national specialist service in Scotland is involved in research – there is a section in each Annual Report.

As a result, Scotland considers that Commitments 24 and 27 of the UK Strategy for Rare Diseases have been met and are now complete.

### **4.4 National Demand Optimisation Group**



The National Demand Optimisation Group (NDOG)<sup>42</sup> was set up by the Scottish Government in 2016. The remit of the group was to review existing practices and information on the use of diagnostic tests across the NHS in Scotland. It was recognised that there was considerable variation in the use of tests, some which could be attributed to differing clinical circumstances or demographic differences.

It is a multi-disciplinary group that contains representatives from Laboratory services,

Radiology, National Services Scotland and Nationally Managed Diagnostic Networks.

The group reviewed existing practices and information, and explored links with local and national initiatives. The main focus of the group was Demand Optimisation. Demand Optimisation is the process by which diagnostic test use is optimised to maximise appropriate testing, which in turn optimises clinical care and drives more efficient use of scarce resource.

The recommendations included:

- NHS Boards should adopt the General Demand Optimisation Guidance and IT Guidance produced by the group.

42 <http://www.gov.scot/Publications/2017/02/5322/1>

- Action on a number of work streams to be taken forward in collaboration with local Health Board leads, for example:
  - Data collection and reporting to allow national collation and analysis.
  - A focussed and collaborative workstream aimed at facilitating the introduction of new tests within definitive clinical pathways.
  - The Scottish Clinical Imaging Network should establish a subgroup to consider the recommendations made, in regard to imaging.
- NHS Boards and Diagnostic Networks should continue to provide support to healthcare science leads, managers and diagnostic staff to work with the national healthcare science leads and diagnostic networks to collectively progress Demand Optimisation work.
- NHS Boards should mandate that diagnostic service providers, and users should develop a focus on demand optimisation, engage in data collection in order to identify variation in user practice, monitor the effect on local improvement in clinical outcomes; and feedback good practice examples.
- The National Diagnostics Managed Network should provide national oversight and consistent quality.

The group will continue their work to allow co-ordination and support for Demand Optimisation work streams and strategy implementation. The Scottish Government have commissioned NSD to take forward further work in relation to An atlas of Variation in the use of Diagnostic tests and supporting local change through the National Diagnostic Networks. The work of the NDOG contributes to achieving Commitment 26 of the UK Strategy for Rare Diseases.

## 4.5 Genetics Laboratory Consortium

In 2016 National Services Division (NSD) undertook a review of the Nationally Designated Genetics Laboratory Testing Services. The review highlighted that the genetics laboratory consortium (GLC) should work towards the introduction of whole exome sequencing in Scotland within the next five years as well as development of multi gene panels in the shorter term. It was also recommended that the service should plan for the introduction of non-invasive prenatal testing (NIPT), non-invasive prenatal diagnostics (NIPD) and circulating tumour DNA (ctDNA) testing in line with professional guidelines and Government policy. As the laboratories develop new testing techniques it is anticipated that this will instigate a reduction in the demand for traditional testing. Therefore the consortium will continue to offer a range of approaches to genetic testing to meet testing requirements for patient care in Scotland.



The review also recommended that the evaluation of the clinical usefulness and cost effectiveness of molecular pathology tests should continue to be undertaken by the Scottish Molecular Pathology Evaluation Panel (MPEP) – involving oncology, haematology, pathology and related disciplines. MPEP recommendations would be made to the Scottish Molecular Pathology Consortium Steering Group for consideration of whether tests should be commissioned routinely by NHS Scotland.

The reworked Genetics Consortium structure will be adapted to mirror these processes and ensure the same rigour is applied to developments in testing practice throughout the consortia. Work is currently underway to rework the Genetics Evaluation Panel (GEP) (formerly the Genetics Laboratory User Group) into a similar panel as MPEP with the skills and knowledge to evaluate genomic/cytogenetic tests and advise the Genetics Laboratory Consortium Steering Group on the clinical utility of tests.

The consortia have been successful in enabling equity of access for all patients in the complex NHSScotland infrastructure to genetics testing. A communications strategy will be developed by National Genetics Laboratory Management Committee; this will include utilisation and expansion of the website currently used for the MPC/MPEP to provide a platform suitable for the combined service/consortia. This will be to provide links to regional services to improve accessibility of testing provision.

In addition, both the Genetics Evaluation Panel (GEP) and the MPEP provide a forum for all clinical users to provide evidence on clinical needs and priorities in relation to genetic testing. MPEP will review evidence on the clinical utility and validity of proposed new genetic tests. GEP will assess proposals tests based on clinical need and recommend which tests should be developed to UKGTN standard.

This helps towards completing Commitments 26, 42, 43, 44 and 45 of the UK Strategy for Rare Diseases.

## 4.6 Laboratory Information Management System (LIMS)

The Tayside genetics laboratory began a procurement exercise in 2015 to develop a fully integrated LIMS system for all aspects of their genetics and molecular pathology service. This went live in June 2017 and is the first fully integrated LIMS system for all three elements of the service in Scotland. Work is underway to activate the LIMS in Raigmore laboratory and colleagues at Tayside have agreed to work with the NHS Grampian laboratory to develop LIMS solution for their needs.

**The VarSeq (VS) Warehouse Project:** The Scottish Genetics Consortium laboratories will pilot specialist software to facilitate the efficient interpretation of NGS data and allow them to share this data securely. The data analysis for NGS is a considerable challenge for two reasons: firstly, the amount of data that is generated by NGS is vast and therefore creates an analysis bottle-neck which is highly labour-intensive; and secondly, the clinical utility of the generated data can only be maximised where all related information is shared amongst the laboratories to aid interpretation. The initiative has the potential to future-proof the laboratories and makes larger NGS analysis such as whole exome sequence (WES) and whole genome sequence (WGS) potentially deliverable.

## 5. RESEARCH (31 – 51)

<b>Commitment</b>	<b>Description</b>
31	Look at how the four UK countries develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways.
32	Work together to identify a selection of rare diseases most suited to the development of best-care pathways and propose other rare diseases for possible pathway development, taking on board the needs of patients and carers and the challenges faced during delivery of the first set of pathways.
33	Examine how they can encourage service providers to involve patients in research and to ensure appropriate funding for excess treatment costs for research in rare diseases.
34	Make better use of online applications to give patients information about their condition so that they can develop a personalised care path plan with their clinical and social care team.
35	Use portals to connect patients and relatives to enhance research participation and, where appropriate, promote self-enrolment to approved research studies with online consenting, self-reporting and use of social media.
36	Encourage patient groups to get involved with regulatory bodies.
37	Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.
38	Explore the feasibility of the UK Clinical Trials Gateway including experimental medicine trials for rare diseases to provide information for patients and their families about research trials.
39	Work with the research community, regulators, providers of NHS Services and research funders to develop risk-proportional permission systems.
40	Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.
41	Promote good practice and the use of systems which facilitate a consistent and streamlined process to local NHS permissions of publicly, charitably and commercially funded research with an aim to reduce timescales.
42	Begin and complete next generation sequencing (NGS) demonstration projects to: evaluate their usefulness, acceptability and cost-effectiveness; develop effective health economic assessments (for example through Health Technology Assessments) and similar initiatives.

Commitment	Description
43	<p>Evaluate the different NGS platform configurations, for example:</p> <ul style="list-style-type: none"> <li>• NGS for clinical condition-specific sets of genes (such as 100-200 of the 22,000 genes);</li> <li>• whole exome sequencing (2% of the entire genome); and</li> <li>• whole genome sequencing.</li> </ul>
44	<p>Support the introduction of NGS into mainstream NHS diagnostic pathways, underpinned by appropriate clinical bioinformatics, including clinical bioinformatics hubs supported by high performance computing centres, where appropriate.</p>
45	<p>Ensure that training and education are available to the NHS workforce, highlighting the importance of NGS to all aspects of rare disease care, including the support for evidence-based local counselling for patients and their relatives who receive NGS results.</p>
46	<p>Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.</p>
47	<p>Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.</p>
48	<p>Set out the benefits of collaboration (besides producing specific treatment) for all stakeholders.</p>
49	<p>Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.</p>
50	<p>Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.</p>
51	<p>Improve engagement between key stakeholders, including:</p> <ol style="list-style-type: none"> <li>a. patients and relatives</li> <li>b. main funding providers</li> <li>c. healthcare commissioners</li> <li>d. NHS hospitals and specialist care units</li> <li>e. industry (pharmaceutical, biotechnology, IT, diagnostics).</li> </ol>

## 5.1 SHARE

SHARE<sup>43</sup> is a new NHS Research Scotland<sup>44</sup> initiative created to establish a register of people interested in participating in health research who agree to allow SHARE to use the coded data in their various NHS computer records to check whether they might be suitable for health research studies. This access can be incredibly useful when it comes to developing new treatments and cures for a wide variety of health conditions.

SHARE aims to have 1 million people registered by 2023, there are currently over 188,000 people registered.

<sup>43</sup> <https://www.registerforshare.org/>

<sup>44</sup> <http://www.nhsresearchscotland.org.uk/>

Work has started on raising awareness in the Rare Disease community, and the existence of SHARE has been communicated to the Scottish network within Genetic Alliance UK.

SHARE has also recently started publishing live studies. People can read about the study, and if they are interested in participating can enter their details into the assessment questionnaire, and it will tell them immediately if they are able to participate.

More information is available on the website: <https://www.registerforshare.org/index.php>

SHARE addresses Commitments 7, 35, 37, 48, 49 and 50 of the UK Strategy for Rare Diseases.

## 5.2 The Scottish Genomes Partnership

The Scottish Genomes Partnership (SGP)<sup>45</sup> was established in January 2015, with a £15m investment from the Universities of Edinburgh and Glasgow. Ten state-of-the-art Illumina HiSeq X genome sequencing instruments were installed at their sequencing hubs. These are Edinburgh Genomics and the Translational Research Centre at the Wolfson Wohl Cancer Research Centre.

In February 2016, the SGP received £6m in research funding from Scotland's Chief Scientist Office (£4m) and the UK's Medical Research Council (£2m). This created a Scotland-wide research partnership to pioneer precision medicine and human genome discovery through academic, NHS clinical and industrial collaborations. The aims of the partnership are to:

- Test the utility of whole genome sequencing for (a) diagnosis of rare diseases and (b) clinical trial stratification for cancers.
- Add value to Scotland's strengths in genomic medicine and science, building a strong foundation for health and wealth creation.
- Expand Scotland's capacity to lead national and international collaborations in human genomics.
- Provide a supportive environment for Scottish academics, SMEs and industry to engage in genomics and bioinformatics research.

### The 100,000 Genomes Project

In March 2017, the SGP Rare Disease collaboration with Genomics England 100,000 Genomes Project<sup>46</sup> opened for recruitment. One thousand participants will be recruited through the nationally designated NHSScotlandgenetic clinics in Aberdeen, Dundee, Edinburgh and Glasgow by the end of March 2018.

The SGP-NHSScotlandcollaboration with the rare disease arm of the 100 Genomes Project has been established with the direct aim of providing evidence to inform the future delivery of NHS genetics services. SGP is contributing 1,000 genomes to the project from patients with rare diseases and their family members to investigate the extent to which Whole Genome Sequencing could improve genetic diagnosis and clinical follow-up for rare disease patients in Scotland. Through this research, genomics services in Scotland are working with clinical genomics researchers in England, Northern Ireland and Wales. This ensures that Scottish patients will benefit from a UK-wide approach to the analysis of WGS, improving rare disease genetic testing and clinical follow-up for patients.

<sup>45</sup> <https://www.scottishgenomespartnership.org/>

<sup>46</sup> <https://www.genomicsengland.co.uk/the-100000-genomes-project/>

### **The SGP describe the study as follows:**

"NHSScotlandGenetics Services will identify potential participants and their families, manage the consent process, collect samples and data, extract DNA and provide de-identified data on participant symptoms to Genomics England. The Universities of England and Glasgow will carry out Whole Genome Sequencing on de-identified DNA samples, before sequence data are sent to Genomics England from the Edinburgh Parallel Computing Centre. Genomics England will analyse variants in the Scottish Genome data alongside the data in the 100,000 Genomes Project, to enable comparisons to be made of symptoms and genomic variants across the dataset. Analyses will continue in the future as more data is obtained from genetics centres around the UK. NHSScotlandGenetics Services will check results received from Genomics England, and provide feedback and clinical results to participants. Clinical management will be carried out within the NHS in the normal way."

As part of the SGP, the NHS National Services Division, which is responsible for commissioning and performance managing Genetics and Molecular Pathology Laboratory services in Scotland, has been leading an alliance of Health Boards across Scotland to facilitate joint working between academic researchers, the four regional clinical Genetics units and four nationally-commissioned Genetic laboratories. A Memorandum of Understanding and Data Sharing Agreement with Genomics England have been put in place to formalise these arrangements.

Intensive working between January 2016 and February 2017 was required to align the Scottish protocol with the Genomics England approach, as well as gaining regulatory approvals from the Public Benefit and Privacy Panel for Health and Social Care and NHS Research Scotland.

With the approval in place, the SGP Rare Disease collaboration with the 100,000 Genomes Project opened for recruitment in March 2017.

Participants are approached by clinical geneticists who will ensure that the participation criteria are met. Potential participants must:

- Have clear evidence of a genetic or family condition which has not been diagnosed through existing test.
- Be part of a family group, which is important for genetic analysis.
- Be eligible according to the list of agreed rare disorders specified by Genomics England.
- The test result will have clinical utility for the patient and/or their family.

The first set of reports from Genomics England are expected back to regional genetic laboratories around December 2017, with reporting back to all participants completed by the end of February 2019.

## Rare Disease Research Projects

The SGP Rare Disease academic studies build on Scotland's outstanding academic track record in gene identifications and functional analysis of single gene human disorders. Four SGP academic-led studies are underway at the Universities of Edinburgh and Glasgow, which focus on molecular influences on motor neurone disease, eye malformation, microcephalic dwarfism and disorders of sexual development. While these are academic-led studies, the Principal Investigators are all part of Multi-Disciplinary Teams which review local clinical cases, and so the learning from all of these studies will be directly transferrable into clinical decision making. The sequencing of around 800 genomes for these studies is 80% complete, with initial analyses underway prior to the selection of final cases.

## SGP Programme developments

Members of the SGP's internationally respected Scientific Advisory Board have commended the exceptional high quality of the on-going work, and noted the cohesive nature of the cross-Scotland working on cutting edge issues of clinical and biological relevance. At its meeting in April 2017, the Board made several recommendations for going forward, which are relevant to the Scottish Rare Disease Implementation Plan.

- *Develop a single Scottish genome archive.* Ensuring that the data collected or generated across the different work streams is integrated and broadly available for clinical and research use as possible, including developing IT tools and infrastructure to facilitate and empower access to the data.
- *Achieving further integration across Scotland.* Capitalising on the opportunities for working together within a small geographical area for rare disease genetics research.
- *Inform downstream delivery of healthcare and future services.* Considering the way in which genomics will be used to deliver healthcare in the future, including commissioning of services, training the workforce in analysis and interpretation of WGS data, and collection of appropriate data to assess usefulness and cost-effectiveness of WGS for diagnosis, treatment and counselling in families affected by rare diseases.
- *Engage the public in genomics.* Advancing understanding and promoting participation are key to future research and genomically-informed care.

The SGP have considered and are now acting on these recommendations.

The work of the SGP addresses Commitments 29, 42, 43, 44, 45, 47, 49 and 50 of the UK Strategy for Rare Diseases.

## Patient Involvement in Research

Scotland and the rest of the UK are leaders in research into rare conditions. Partnership and collaboration with patients and patient groups in developing research initiatives is necessary to ensure high quality research projects continue to be developed.

An example of positive collaboration in research is the Chief Scientist Office (CSO) work in partnership with rare diseases charities Action Duchenne and Muscular Dystrophy UK to co-fund a collaborative clinical research fellowship.

This project began when a grandfather of a child with Duchenne Muscular Dystrophy raised the issue of disparity in access to research with Scottish Ministers - patients were unable to access a number of important clinical trials in England. This led to a meeting with the CSO and the agreement to work collaboratively with Action Duchenne and Muscular Dystrophy. Importantly for the charities involved, this provided an opportunity to have a say in shaping the research fellowship. It was important to the charities that the post holder would have a clinical focus, and as such a fellowship which was 70% clinical and 30% research focus was agreed.

A further benefit for the charities involved was that, as grant holder, the CSO's robust recruitment procedures were used and that the role was to be managed directly by the CSO. The project began approximately one year ago and the charities continue to be involved. Charities are in regular contact with the post holder and have the opportunity to contribute to the project's development. The CSO also maintains contact with the charities, providing regular updates. This ensures transparency and ongoing patient involvement.

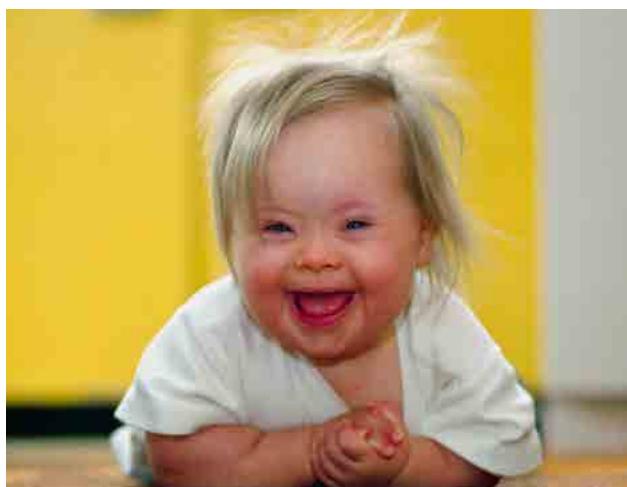
Dr Diana Ribeiro, CEO of Action Duchenne has said 'This has been a ground breaking collaborative model - this should be used as a best practice example for **translational genetic research for genetic conditions like Duchenne** across the United Kingdom'.

### 5.3 Next Generation Sequencing (NGS)

In addition, the Scottish Genetics Centres are now part of the SGP, who will develop the methodology for using Whole Genome Sequencing for diagnosis in Scotland. In the meantime Scottish laboratories continue to develop next generation sequencing for use as a diagnostic tool for genetics. Examples of developments include:

- NHS Grampian's development of BRCA1/2 NGS service has resulted in significant cost and staff time efficiencies, and consistently shorter reporting times. This service has been recently expanded to enable and provide BRCA1/2 somatic testing for eligible patients. Similarly, NGS application has also replaced existing Sanger services for the routine delivery of the national cardiac arrhythmias and familial hypercholesterolaemia services. NGS has also been used in the delivery of a newly commissioned service for Primary Immune Deficiencies (PID)
- NHS Tayside has implemented NGS into their diagnostic service for familial aortic aneurysms and phaeochromocytoma/paraganglioma. This is based on targeted amplification and sequencing of the genes most commonly associated with these conditions; currently there are 10 genes in each panel. In addition the laboratory is working with clinical colleagues to design similar panels for inherited diabetes, genetic skin disease, cystic renal disorders and Parkinson's disease. These panels will offer a better service to patients and improve the laboratory workflow.
- NHS Glasgow Laboratory is delivering Next Generation Sequencing for infantile onset epilepsy (ION). In addition they have evaluated an extended Next Generation Sequencing panel for Disorders of Sexual Development (DSD) which will shortly be available for routine diagnostic service delivery.
- The NHS Lothian laboratory has issued over 2000 NGS reports in the past four years for disease specific panels for Stargardts disease, genes in the RASMAPK pathway, the hypertrophic and dilated cardiomyopathies, Hereditary Haemorrhagic telangiectasia, Alport syndrome, ocular malformations, primary microcephaly, cancer, neurodegeneration. Shifting to NGS technology has increased throughput and decreased costs.

A wide range of Next Generation Sequencing (NGS) tests are available through the Scottish Genetics Laboratory Consortium (SGLC)



In relation to developing and rolling out NGS services as detailed above, the Consortium Genetics Laboratories have undertaken evaluation of the performance of numerous testing platforms. In reference to exome the consortia laboratories have also begun to undertake some evaluative analysis e.g. following the experience gained through NHS Lothian pilot study, to evaluate Whole Exome Sequencing (WES) in relation to Developmental Delay Disorders (DDD) a outline proposal for the implementation of an exome sequencing strategy for the diagnosis of developmental disorders has been drafted.

The review of Genetics laboratory services conducted throughout 2016 included a comparative assessment of workforce across the Scottish Genetics Laboratory and Molecular Pathology Consortium service. It was recognised that the provision and sustainability of the highly specialist

genetics and molecular pathology services was challenging given the lead times for training of skilled staff; and that workforce planning was required on a “Once for Scotland” basis to ensure succession planning in the light of the current age profile of staff in Scotland. The Review Group recommended that the future model of service should ensure adequate training opportunities and succession planning. This work will be taken forward under the revised consortia governance structure.

Recognising the continued need to develop genetics services, the Edinburgh laboratory created a partnership with the Institute of Genetics and Molecular Medicine (IGMM) to devise a strategy to compliment the NES pre-registration scientists training scheme (on which there is a yearly limited intake) for training clinical scientists. Two fixed term posts for translational science initiatives in diagnostic genetics were created using a vacant unfilled clinical scientist post and funding from the IGMM translational Medicine initiative to appoint two IGMM Clinical Translational post-doc posts. A three year training plan has been devised.

The Glasgow laboratory received funding for two research and development scientists to work within the NHS laboratory following the approval of a Joint Working Project, via the NHS GG&C/ Pharmaceutical Industry Alliance Partnership Group. The posts are funded for three years as part of this joint working partner initiative between NHS GG&C and pharmaceutical industry partners for the development and evaluation of services (including NGS) for cancer diagnostics. The long term strategy of the initiative is to build infrastructure and resources for sustained future development and translation into routine genetic cancer services, and it was recognised that there were difficulties in freeing up development time. Since the recruitment of two scientists in 2015, they have been working on a programme to develop the delivery of NGS for cancer which has included the evaluation of secondary and tertiary analysis software for, cross training of scientists in basic bioinformatics, along with the training of technical staff in NGS laboratory work flows.

## 5.4 Co-funding of research

Within the Scottish Government, the Chief Scientist Office (CSO) has responsibility for the funding of clinical research. They work with a number of charities and have awarded over £1 million to studies into rare diseases. They are as follows:

Charity/Organisation	Total Funding	Project Title
Action Duchenne Muscular Dystrophy UK	£225,000	Secondary Osteoporosis and Its Therapy in Duchenne Muscular Dystrophy – ScOT-DMD
Motor Neurone Disease Assoc MND Scotland	£225,000	Population based genotype-phenotype correlation to stratify incident cases of patients with MND in Scotland from 2015-2017
Muscular Dystrophy UK	£216,000	Structural CNS changes, neuropsychological impairment and sleep disturbance in myotonic dystrophy
PSP Association	£195,000	Improving diagnostic and care pathways in progressive supranuclear palsy and corticobasal degeneration
Scottish Huntington's Assoc RS MacDonald Trust	£200,000	Towards improvement in care delivery in Huntington's Disease

This addresses Commitment 46, 47 and 48 of the UK Strategy for Rare Diseases.

## 5.5 Motor Neurone Disease (MND)

### Explanation: What is Motor Neurone Disease?

The MND Association describe Motor Neurone Disease as “a group of diseases that affect the nerves (motor neurones) in the brain and spinal cord that tell your muscles what to do. With MND, messages from these nerves gradually stop reaching the muscles, leading them to weaken, stiffen and waste.”<sup>47</sup>

The Scottish Government announced on 22 February 2017 that they were setting up the Gordon Aikman Scholarship to honour the late campaigner, who lost his fight with MND on 3 February 2017. The £50,000 a year will support individuals and professionals to develop, implement and evaluate practical interventions to improve the quality of life of people affected by MND. The scholarship will be run in partnership with MND Scotland.

The scholarship scheme will accept applications from people working in healthcare and from those affected by MND and their carers whose experience and expertise is invaluable in helping to continually improve care.

<sup>47</sup> <https://www.mndassociation.org/about-mnd/where-do-i-start/what-is-mnd/>

The scholarship will support individuals and professionals to develop, implement and evaluate practical interventions to improve the quality of life of people affected by MND. This may be through travel to identify best practice and/or using improvement and research approaches to test the impact and suitability of those interventions. This could include non-medicine based interventions such as specialised physiotherapy techniques, or non-medicalised assisted breathing techniques.

Projects are expected to be completed in 12 months. Successful applicants will be expected to share their findings, learning and best practice with their peers and across Scotland.

### **MND and MS PhD programmes**

The 2016 SNP manifesto contained the following commitment:

**'We will work to support research into – and treatment of patients with – neurological conditions. Funding will be made available for three research Ph.D.'s in Motor Neurone Disease and a further three in Multiple Sclerosis'**

The Chief Scientist Office was tasked with the delivery of this commitment, and following a rigorous external peer review process, a decision was taken in November 2016 to fund a bid led by the University of Edinburgh to host the above-mentioned PhD Programmes.

The Edinburgh-led bid represents strong multicentre effort that will combine MS and MND in an integrated Scotland wide approach. The participating Universities (Aberdeen, Dundee, Edinburgh, Glasgow, St Andrews) have committed to fund an additional 6 PhD's, bringing the total available to 12. There are plans to recruit to 4 PhD positions per year, thereby building a cohort over the 5 year duration of the programme (each PhD having a duration of 3 years). Recruitment of the first round of PhD's has been completed (109 applications were received and 9 candidates interviewed). There were 4 successful candidates, and 3 of the 4 projects are on MND.

The work undertaken for MND helps to contribute to meeting Commitment 47 of the UK Strategy for Rare Diseases.

## **6. CHALLENGES AND NEXT STEPS**

The Challenges and next steps highlighted in the Second Progress Report from the UK Rare Disease Policy Board are also pertinent to Scotland, most notably the EU Exit and especially the effect that this will have on the European Reference Networks (ERNs) and access to healthcare systems across Europe. The Scottish Government will be working closely with colleagues in the rest of the UK on this matter.

Whilst the wider national issues are of great importance, there are some areas within Scotland which are challenging and which the Rare Disease Implementation Oversight Group intend to focus on over the next two years.

### **6.1 Information Sharing between computer systems**

The digital aspect of sharing information between systems in primary and secondary care is a challenge for the rare disease implementation plan in Scotland. Currently the systems are not linked up and therefore ensuring that key information is shared between primary and secondary care is often left to manual processes. As reported in section 3.2, the Scottish Government, in partnership with key stakeholders, is developing a new, integrated Digital Health and Social Care Strategy that will build on achievements to date and set out future development and priorities, this is a Scotland-wide problem for all health conditions and will need to be considered by Scottish Ministers and NHS Health Boards.

### **6.2 Social Care**

In drafting this report, the Rare Disease Implementation Oversight Group (RDIOG) acknowledged that the group is missing oversight of the implementation plan in the social care setting. Accordingly, in the next few months the group will consider the commitments in the implementation plan and look to how Integrated Joint Boards should be included in the group/plan.

### **6.3 Communication**

The Rare Disease Implementation Oversight Group acknowledge that they have not been actively communicating progress with the Rare Disease community. When the implementation plan was published, there was an expectation from the community that change would happen quickly, and while there has been some communication on major achievements, like the SGP, progress in other areas has not been communicated. As a result, a communications plan has been drafted and work will begin in winter 2017 to increase awareness of rare diseases and to report on the progress of the RDIOG. This will include updating websites, using social media and having a presence at relevant health and social care events.

## **6.4 Genomics**

While work has only recently started in Scotland on the SGP, it has become clear that genomics will become a major focus in health in years to come. Progress is occurring at unprecedented speed and the Scottish Government and NHS Scotland need to consider very quickly how information will be stored and how it can be used routinely. It will also need to consider training a large number of staff in this area. This will be a significant piece of work.

The Scottish Government, with the support of the Chief Medical Officer have requested that the Scottish Science Advisory Council prepare a report entitled “Development of Genomic Medicine in Scotland” to help inform their future activities.

The proposed key areas to be included are:

- The current capabilities for human genomics in Scotland and developments elsewhere.
- The potential to build on existing assets in Scotland.
- The opportunities for, and benefits to, the NHS, research and the life sciences sectors in Scotland arising from further development, including in technology, skills and partnerships.
- The areas where investments may be needed in order to realise these opportunities and benefits.

The report is expected in mid 2018 and will help inform evidence for a future strategy for Genomics in Scotland.

## **6.5 NHS Inform**

NHS Inform is the Scottish equivalent of NHS Choices. The system was redesigned and re-launched in 2017, and unfortunately several of the pages that referred to rare disease were lost or corrupted. Work started in August 2017 to review these pages and consider how the resource could be better used to be of more use for people with rare diseases.

## **6.6 NHS Education training and resources**

The Scottish Implementation Plan “It’s Not Rare to Have a Rare Disease” states that NHS Education Scotland will consider what information, training and resources may be deliverable to staff, with the Knowledge Network being offered as one possibility. The Knowledge Network has become vast and staff at NES are looking to streamline the information available. However, the work being undertaken by RARE BestPractice and the decision support tools will help to address this. Other educational resources have not been considered and this will form part of the work going forward for the next phase of implementing the plan.

# ANNEX 1 – PROGRESS AGAINST UK STRATEGY COMMITMENTS AND ‘IT’S NOT RARE TO HAVE A RARE DISEASE’ OVERVIEW

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
Empowering Those Affected by Rare Diseases				
1	<p>Strengthen the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision and research, recognising patient groups as key partners – including in the development of the four country plans to implement the Strategy.</p>	<p>The use of existing NHS Board patient and public involvement structures and the Health and Social Care Partnerships arrangements as these develop.</p> <p>The creation of opportunities for patients with a rare disease to participate in decisions about all aspects of their care, support and treatment.</p>	<p>1.1 Rare Disease Implementation Oversight Group            1.6 What Matters to You?            1.8 Our Voice            1.9 National Specialist Services Committee            1.10 National Network Management Services            1.13 Franks Law</p>	Ongoing – will be monitored during the next phase of implementation.
2	Improve awareness amongst service providers and others of the effects that rare diseases can have on a person’s education, family, social relationships and ability to work.	Consideration of how the Person-Centred Health and Care Collaborative can best take account the needs of people with rare diseases.	<p>1.5 House of Care Model            1.6 What Matters to You?            1.7 Care Opinion            1.8 Our Voice            1.9 National Specialist Services Committee            1.10 National Network Management Services            1.12 Provision of Communication Equipment            1.13 Franks Law</p>	Ongoing – will be monitored during the next phase of implementation.
3	Encourage effective and timely liaison between the NHS and other public service providers, and encourage providers to consider the effects of rare disease on people’s lives when they are developing and managing services.		<p>1.2 Health &amp; Social Care Delivery Plan            1.4 Making it Easy: Health Literacy            1.5 House of Care Model            1.6 What Matters to You?            1.7 Care Opinion</p>	Ongoing – will be monitored during the next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
Empowering Those Affected by Rare Diseases				
4	Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services.	Publicising the Patient *Care* Opinion website and utilising SMS, social media, 'apps' and other communication technologies to provide patients with rare disease and families with opportunities to feedback on their experience of services.	1.2 Health & Social Care Delivery Plan 1.3 Realistic Medicine 1.4 Making it Easy: Health Literacy 1.5 House of Care Model 1.6 What Matters to You? 1.7 Care Opinion 1.8 Our Voice 1.9 National Specialist Services Committee 1.10 National Network Management Services 1.12 Provision of Communication Equipment	Ongoing – will be monitored during the next phase of implementation via the Communications plan.
5	Consider how to give all patients with rare disease clear and timely information about: their condition and its development; treatment and therapy options; practical support.	Consideration of the development of current NHS Inform resources to provide access for people with rare diseases and their families to clear information on relevant conditions in a range of accessible formats, including links to third sector information resources.	1.2 Health & Social Care Delivery Plan 1.3 Realistic Medicine 1.4 Health Literacy 1.5 House of Care Model 1.6 What Matters to You? 1.11 Health & Social Care Standards 1.13 Franks Law	Work has started and will continue through the next phase of implementation.
		Opportunities arising from the Person-centred eHealth Strategy and Delivery Plan.		The RDIOG has contributed to the Digital Health and Care Strategy for Scotland.
6	Improve access for patients (or where appropriate their parents or guardians) to their personal data.	We will explore with relevant agencies the applicability/practicability of registers and the opportunities to develop a patient electronic health record.	1.12 National Patient Portal	Ongoing – will be monitored during the next phase of implementation.
7	Support patients to register on databases, where these exist.	Staff in appropriate roles seeking to signpost individuals with rare disease to further information on research and clinical trials and opportunities to participate.	1.15 Scottish Genetics Speciality Group 5.1 SHARE	Ongoing – will be monitored during the next phase of implementation.
8	Help patients to contribute to research and other activity related to rare disease.			

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
Identifying And Preventing Rare Disease				
9	Continue to work with the UK NSC to ensure that the potential role of screening in achieving earlier diagnosis is appropriately considered in the assessment of all potential new national screening programmes and proposed extensions to existing programmes.	Consider the introduction in Scotland of recommendations from the UK NSC.	2.1 Introduction of new screening tests 2.2 The Scottish Screening Committee	Ongoing – will be monitored during the next phase of implementation.
10	Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular tests are evaluated and recommended by UKGTN is accessible for at risk relatives.	Continue to ensure access for people in Scotland and families to UKGTN approved carrier testing, extending coverage as appropriate.	2.3 UK Genetic Testing Network	Ongoing – will be monitored during the next phase of implementation.
Diagnosis And Early Intervention				
11	Work to achieve reduced times for diagnosis of rare diseases, whilst acknowledging that more needs to be done to ensure that undiagnosed patients have appropriate access to co-ordinated care e.g. to help disabled children who are thought to have a genetic syndrome or condition that science has not yet identified.	Review by NHS Boards, NSD & Managed Diagnostic Networks of the input of diagnostic specialities to rare disease diagnosis and consideration of how delays may be reduced.	1.2 Health & Social Care Delivery Plan 1.9 National Specialist Services Committee 1.10 National Network Management Services 3.7 RARE Best Practice 4.4 National Demand Optimisation Group	Ongoing – will be monitored during the next phase of implementation.
		To make rare disease guidelines accessible via a single point online database - delivered by the RARE-Best Practice project.		Ongoing – will be monitored during the next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
Diagnosis And Early Intervention			
12	<p>Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients, by:</p> <ul style="list-style-type: none"> <li>• establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and specialist clinical centres, as appropriate</li> <li>• putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment</li> <li>• drawing on patients' ability to help inform decisions about referral and diagnosis</li> <li>• creating effective clinical networks to support this process</li> <li>• making high quality diagnostic tests accessible through common, clinically agreed systems or pathways</li> <li>• embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies.</li> </ul>	<p>NSD action to increase awareness among clinicians in Scotland of criteria for patient referral to diagnostic pathways for Scottish or English specialist centres.</p> <p>Obtaining the input of primary care to the development of appropriate pathways for diagnosis, treatment and support.</p> <p>Consideration of the adaptation of RefHelp (Lothian Referral Guidelines) to include rare disease pathways to assist GPs in the recognition, management and referral of patients with rare disease.</p>	<p>3.1 National Managed Clinical Networks &amp; Pathways 3.2 Decision support tools for H&amp;S care 3.6 Data Short-life Working Group 4.3 Specialist Centres 4.4 National Demand Optimisation Group</p> <p>The Scottish Implementation Plan action is complete.</p> <p>Ongoing through various workstreams. Will continue to be monitored during next phase of implementation.</p> <p>Will be considered during the next phase of implementation.</p>
13	Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.	3.5 The Montgomery Review	Ongoing – will continue to be monitored during next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
Diagnosis And Early Intervention			
14	Where appropriate, support the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered.	Exploration by NSS Public Health and Intelligence of how rare disease decision support systems and other rare disease software systems may feature in the future development of IT systems in NHS Scotland, ensuring these are interoperable with GP clinical systems.	3.2 Decision support tools for H&S care  Ongoing – will continue to be monitored during next phase of implementation.
	Consideration of the inclusion within post-graduate training of appropriate content on diagnostic skills relevant to rare disease.		To be discussed with NES.
	Engagement with NSS Public Health and Intelligence to carry out a stocktake of rare disease databases to scope what existing patient data systems exist, where these are held and how they might be improved.		COMPLETE

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
Diagnosis And Early Intervention			
15	<p>Improve education and awareness of rare diseases across the healthcare professions, including:</p> <ul style="list-style-type: none"> <li>• involving patients in the development of training programmes</li> <li>• encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics</li> <li>• ensuring awareness of methods and clinical techniques used in differential diagnosis.</li> </ul>	Use rare disease information to help improve decision making as part of a learning care system in Scotland.	Ongoing – this will be monitored during the next phase of implementation.
		Exploration of options for inclusion of input from patients with rare diseases into the post-graduate training programmes for doctors, nurses and allied health professionals.	Will be considered during the next phase of implementation.
		Consideration of the inclusion within post-graduate training of appropriate content on diagnostic skills relevant to rare disease.	Will be considered during the next phase of implementation.
		Supporting the development of higher specialist training in genetics for medical and scientific staff to develop sustainability in the genetic workforce.	Will be considered during the next phase of implementation.
		Developing awareness raising of rare disease among midwifery, health visiting and GP professions.	Will be considered during the next phase of implementation.
16	Monitor the development of ICD-11 in preparation for its adoption.	Consideration by NSS Public Health & Intelligence of the best means by which to tackle the limitations affecting data capture arising from the use of existing coding systems such as ICD-10 and link to the roll out of ICD-11.	3.6 Data Short Life Working Group Ongoing – will continue to be monitored during next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
Diagnosis And Early Intervention				
17	<p>Work with colleagues in Europe in the development of the European Orphanet coding system and consider the adoption of Orphanet coding and nomenclature.</p>	Publicising Orphanet as appropriate to NHS Boards, staff groups, training organisations, universities and others across NHS Scotland.	3.6 Data Short Life Working Group	Ongoing – will continue to be monitored during next phase of implementation.
		Consideration of the input Scotland should have into Orphanet, either independently or as part of UK joint work and the feasibility of using Orphanet over and above existing data recording systems, without creating different systems and standards.		COMPLETE. Scoping work undertaken and Orphanet not suitable for the purpose of coding. ICD-11 codes to be used.
18	Standardise data collection, building on existing NHS data standards, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.	Engagement with NSS Public Health and Intelligence to carry out a stocktake of rare disease databases to scope what existing patient data systems exist, where these are held and how they might be improved.	3.6 Data Short Life Working Group	The scoping work for the Scottish action is complete. Work on data will go forward during the next phase of implementation.
19	<p>Explore options to improve the link between existing patient data and electronic health records.</p>	We will explore with relevant agencies the applicability/practicability of registers and the opportunities to develop a patient electronic health record.	1.14 National Patient Portal 3.6 Data Short Life Working Group	COMPLETE - The scoping work for the action is complete.
		Exploration, through NHS Board eHealth leads and NSS Public Health & Intelligence of the feasibility of developing links between NHS Board patient information portals to support patients at diagnosis and to assist in providing co-ordinated and informed care.		Work on data will go forward during the next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
Diagnosis And Early Intervention			
20	Assess the potential for rare disease databases where they do not exist.	Mapping of existing nationally held data-sets to assess which have sufficiently detailed clinical coding to identify patients with rare disease.	3.6 Data Short Life Working Group
21	Agree international standards, building on existing UK standards.	Exploration of opportunities to develop links between UK databases, Scotland specific databases and relevant international databases.	3.6 Data Short Life Working Group
22	Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally.		3.6 Data Short Life Working Group 3.7 RARE Best Practice
Co-ordination Of Care			
23	<p>Continue to develop service specifications for rare diseases. This will include country specific care pathways and a 'generic' care pathway that sets out best practice that can be applied to all patients with rare diseases in the UK (particularly where there are no disease specific pathways). The generic care pathway will include:</p> <ul style="list-style-type: none"> <li>• an appropriate care plan for all patients with rare disease</li> <li>• clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis</li> <li>• the development of seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age</li> <li>• access to criteria and measures of quality and outcomes.</li> </ul>	<p>Publication of patient pathways for rare disease and the referral requirements for services in and out of Scotland.</p>	1.9 National Specialist Services Committee 1.10 National Network Management Services 3.1 National Managed Clinical Networks & Pathways 4.2 Specialist Care in the UK 4.3 Specialist Centres

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
Co-ordination Of Care				
24	<p>Agree that specialist clinical centres should, as a minimum standard:</p> <ul style="list-style-type: none"> <li>• have a sufficient caseload to build recognised expertise</li> <li>• where possible, not depend on a single clinician</li> <li>• coordinate care</li> <li>• arrange for coordinated transition from children's to adults' services</li> <li>• involve people with rare conditions, and their families and carers</li> <li>• support research activity</li> <li>• ensure their expertise is available to families and their healthcare teams.</li> </ul>	<p>NSD action to increase awareness among clinicians in Scotland of criteria for patient referral to diagnostic pathways for Scottish or English specialist centres.</p>	<p>4.2 Specialist Care in the UK 4.3 Specialist Centres</p>	<p>COMPLETE. All national specialist services in Scotland have specifications in place which are reviewed regularly. This is the template which any new centre will also follow.</p>
25	Ensure that the relationship between the specialist clinical centres and science and research is explained to and understood, and put into practice by: practitioners delivering local health and social care; the research community; industry; academia.	Ensuring Scottish & UK specialist centres bring together all appropriate investigation, diagnosis, treatment, support and research expertise for rare diseases.	<p>4.2 Specialist Care in the UK 4.3 Specialist Centres</p>	Ongoing – will continue to be monitored during next phase of implementation.
26	Set out clearly the connections to, and communications with Specialist Clinical Centres in molecular diagnostics and other forms of diagnostic support.	Working with specialist services to develop clear diagnostic pathways for molecular diagnosis.	<p>4.4 National Demand Optimisation Group 4.5 Genetics Laboratory Consortium</p>	Ongoing – will continue to be monitored during next phase of implementation.
27	Ensure that specialist clinical centres are as concerned with research as with health and social care support, and that they develop networks that provide professional-to-professional dialogue and collaboration across a wide range of experts, including internationally (especially for those conditions that are ultra-rare).	Keeping under review the relevance to patients of any potential development of specialist services.	<p>4.3 Specialist centres</p>	<p>COMPLETE. All National Services in Scotland is involved in research. Each Annual Report includes a section on research.</p>

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
Co-ordination Of Care				
28	Work with international partners wherever possible and develop UK-wide criteria for centres to become part of an expert reference network to increase flow of information between patients and professionals in a range of disciplines.	4.1 European Reference Networks.	Ongoing – will continue to be monitored during next phase of implementation.	
29	Improve systems to record genetic and other relevant information accurately to detail the incidence and prevalence of disease and to support service planning and international planning.	Ensuring appropriate data standards and coding methods are adopted and used in any rare disease database which is developed.  Utilisation of existing UK, EU and international databases to provide NHSScotland clinicians with the opportunity to monitor certain diseases and to produce data of benefit to research and to service planning.	3.6 Data Short Life Working Group 5.2 Scottish Genomes Partnership  3.6 Data Short Life Working Group 3.7 RARE Best Practice	Ongoing – will continue to be monitored during next phase of implementation.
30	Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments.	Engagement between NSD, e-Health Strategy and NSS Public Health and Intelligence colleagues to consider how to best develop the IT database requirements for national managed clinical networks to collect systematic patient data.	3.2 Decision support tools for H&S care 3.6 Data Short Life Working Group	Ongoing – will continue to be monitored during next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
The Role Of Research In Rare Diseases			
31	Look at how the four UK countries develop, change or expand information systems to capture, connect and analyse data about clinical and social care pathways.	Exploration of opportunities to develop links between UK databases, Scotland specific databases and relevant international databases.	3.5 Data Short Life Working Group  Ongoing – will continue to be monitored during next phase of implementation.
32	Work together to identify a selection of rare diseases most suited to the development of best-care pathways and propose other rare diseases for possible pathway development, taking on board the needs of patients and carers and the challenges faced during delivery of the first set of pathways.	Publication of patient pathways for rare disease and the referral requirements for services in and out of Scotland.	3.1 National Managed Clinical Networks & Pathways  Ongoing – will continue to be monitored during next phase of implementation.
33	Examine how they can encourage service providers to involve patients in research and to ensure appropriate funding for excess treatment costs for research in rare diseases.	Utilisation of existing UK, EU and international databases to provide NHS clinicians with the opportunity to monitor certain diseases and to produce data of benefit to research and to service planning.	5.4 Co-funding of research  Ongoing – will continue to be monitored during next phase of implementation.
		Continuation of the central management of excess treatment costs to facilitate prompt approval of rare disease studies.	The CSO has continued its policy of centrally managing excess treatment costs to facilitate prompt approval of rare disease studies and activity based funding of NHS Boards.
34	Make better use of online applications to give patients information about their condition so that they can develop a personalised care path plan with their clinical and social care team.	Continuation of the SHARE Register, maintaining its important role in supporting participation in research of people from across the population.	5.1 SHARE  Ongoing – will continue to be monitored during next phase of implementation.
		Exploration of the development of online application processes and registers of interest for people with rare disease wishing to participate in relevant research.	

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
The Role Of Research In Rare Diseases			
35	Use portals to connect patients and relatives to enhance research participation and, where appropriate, promote self-enrolment to approved research studies with online consenting, self-reporting and use of social media.	Exploration of the development of online application processes and registers of interest for people with rare disease wishing to participate in relevant research.	5.1 SHARE  COMPLETE. The SHARE register is the national health research space for Scotland.
36	Encourage patient groups to get involved with regulatory bodies.		Health Improvement Scotland is the regulator for healthcare – consideration will be given to this commitment during the next phase of implementation.
37	Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations.	Continuation of the activity based funding scheme which rewards NHS Boards for recruiting patients into trials.	5.1 SHARE  COMPLETE. The SHARE register is the national health research space for Scotland.
38	Explore the feasibility of the UK Clinical Trials Gateway including experimental medicine trials for rare diseases to provide information for patients and their families about research trials.	Maintenance of the CSO's success in streamlining research governance through collaboration with HRA and others.  Ensuring that rare disease research is fully supported in the reorganised clinical research infrastructure for NHSScotland and that Scotland is represented on any proposed rare disease strategic research initiative.	CSO has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK.
39	Work with the research community, regulators, providers of NHS Services and research funders to develop risk-proportional permission systems.	Maintenance of the current multicentre study mean approval times of 21 working days for commercial and 15 working days for non-commercial studies.	COMPLETE. The Research Active Scottish NHS Boards have signed up to the Musketeer's memorandum thereby facilitating approval of multi-centre rare disease clinical studies.
40	Encourage researchers to use current guidance to produce generic participant information leaflets and consent forms and participate in future guidance reviews.	Continuation of co-funding for research projects and capacity building initiatives with research charities working on rare diseases and pursuit of opportunities for joint working in rare disease research.	Ongoing – will continue to be monitored during next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action	
The Role Of Research In Rare Diseases				
41	Promote good practice and the use of systems which facilitate a consistent and streamlined process to local NHS permissions of publically, charitably and commercially funded research with an aim to reduce timescales.	Continuation of co-funding for research projects and capacity building initiatives with research charities working on rare diseases and pursuit of opportunities for joint working in rare disease research.	4.5 Genetics Laboratory Consortium	
42	Begin and complete next generation sequencing (NGS) demonstration projects to: evaluate their usefulness, acceptability and cost-effectiveness; develop effective health economic assessments (for example through Health Technology Assessments) and similar initiatives.	Continuation (and evaluation as the technology develops) of the use of small next generation sequencing (NGS) devices in all Scottish Genetics Laboratories and mainstreaming of NGS as appropriate into NHS Diagnostic pathways.	4.5 Genetics Laboratory Consortium 5.2 Scottish Genomes Partnership 5.3 Next Generation Sequencing	Ongoing – will continue to be monitored during next phase of implementation.
43	Evaluate the different NGS platform configurations, for example: <ul style="list-style-type: none"><li>• NGS for clinical condition-specific sets of genes (such as 100-200 of the 22,000 genes)</li><li>• whole exome sequencing (2% of the entire genome)</li><li>• whole genome sequencing</li></ul>		4.5 Genetics Laboratory Consortium 5.2 Scottish Genomes Partnership 5.3 Next Generation Sequencing	Ongoing – will continue to be monitored during next phase of implementation.
44	Support the introduction of NGS into mainstream NHS diagnostic pathways, underpinned by appropriate clinical bioinformatics, including clinical bioinformatics hubs supported by high performance computing centres, where appropriate.		4.5 Genetics Laboratory Consortium 4.6 LIMS 5.2 Scottish Genomes Partnership 5.3 Next Generation Sequencing	Ongoing – will continue to be monitored during next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
The Role Of Research In Rare Diseases			
45	Ensure that training and education are available to the NHS workforce, highlighting the importance of NGS to all aspects of rare disease care, including the support for evidence based local counselling for patients and their relatives who receive NGS results.	Participation between NHS Education Scotland, NSD, the Genetics Laboratory Consortium and the Molecular Pathology Consortium in the review of future scientist staffing requirements to support future developments in genetic testing.	4.5 Genetics Laboratory Consortium 5.2 Scottish Genomes Partnership  Ongoing – will continue to be monitored during next phase of implementation.
46	Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.	Continuation of co-funding for research projects and capacity building initiatives with research charities working on rare disease and pursuit of opportunities for joint working in rare disease research.	5.4 Co-funding of Research 5.5 Motor Neurone Disease  Ongoing – will continue to be monitored during next phase of implementation.
47	Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.	Continuation of co-funding for research projects and capacity building initiatives with research charities working on rare disease and pursuit of opportunities for joint working in rare disease research.	5.1 SHARE 5.2 Scottish Genomes Partnership 5.4 Co-funding of Research 5.5 Motor Neurone Disease  Ongoing – will continue to be monitored during next phase of implementation.
48	Set out the benefits of collaboration (besides producing specific treatment) for all stakeholders.	Ensuring that rare disease research is fully supported in the reorganised clinical research infrastructure for NHSScotland and that Scotland is represented on any proposed rare disease strategic research initiative.	5.1 SHARE 5.4 Co-funding of Research  COMPLETE.
49	Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.	Continuation of the SHARE Register, maintaining its important role in supporting participation in research of people from across the population.	5.1 SHARE 5.2 Scottish Genomes Partnership  Ongoing – will continue to be monitored during next phase of implementation.
50	Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.	Continuation of co-funding for research projects and capacity building initiatives with research charities working on rare diseases and pursuit of opportunities for joint working in rare disease research.	5.1 SHARE 5.2 Scottish Genomes Partnership  Ongoing – will continue to be monitored during next phase of implementation.

UK Commitment	It's Not Rare to Have a Rare Disease	Progress Number	Next Action
The Role Of Research In Rare Diseases			
51	<p>Improve engagement between key stakeholders, including:</p> <ul style="list-style-type: none"> <li>a. patients and relatives</li> <li>b. main funding providers</li> <li>c. healthcare commissioners</li> <li>d. NHS hospitals and specialist care units</li> <li>e. industry (pharmaceutical, biotechnology, IT, diagnostics).</li> </ul>	<p>The Scottish Government will continue to participate, as appropriate, in the UK Rare Disease Stakeholder Forum. It will also, through its Implementation Oversight Group, engage with all relevant partners in the development of actions and agreement of timescales to progress support and treatment for rare disease.</p>	Ongoing – will be monitored during the next phase of implementation via the Communications plan.

## ANNEX 2 – SUMMARY REPORT FROM RDIQG SHORT LIFE WORKING GROUP ON DATA



**Rare  
Diseases in  
Scotland**

## What is a Rare Disease?

A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases and around five new rare diseases are described in medical literature each week.

It is estimated that within the Scottish population there are up to 300,000 or 1 in 17 people who may be affected by a rare disease over their lifetime.

## Issues for People with a Rare Disease

Patients with rare diseases in Scotland have experienced delays in getting a diagnosis and treatment, with several getting a number of diagnoses on the way.

## It's Not Rare to Have a Rare Disease

The Scottish Government's implementation plan, It's Not Rare to Have a Rare Disease - The Implementation Plan for Rare Diseases in Scotland was launched in June 2014. This plan highlights the numbers that are affected by a rare disease in Scotland, the difficulties these patients face and the need for action moving forward.

The Rare Disease Implementation Oversight Group was then created to take the Plan forward. One strand of the Group's work has focused on data, tasked to consider how information on rare disease can be captured in Scotland to enable monitoring and reporting.

## Where is Rare Disease Data Held?

Rare disease information may be captured in a number of places, including;



## Congenital Anomalies

Congenital anomalies comprise a wide range of abnormalities of body structure or function that are present at birth and are of prenatal origin. Congenital anomalies account for approx 80% of all rare diseases and affect approx 2-4% of pregnancies in Scotland. There is currently no national congenital anomaly register.

Recommendations from a recent scoping exercise looking at potential data sources to identify patients with a rare disease included a recommendation to establish a congenital anomalies register in Scotland. This register would create a user view of appropriate data held within the NSS Corporate Data Warehouse, ensuring Scotland remains in line with the congenital anomalies work undertaken in England by Public Health England.

## Current Data Sources within NSS (PHI & NSD) Which May Capture Information on Rare Diseases;

### Coding Used

Currently only a small fraction of rare diseases have codes in international nomenclatures, making it difficult to trace patients with rare diseases in health information systems on a national and international level. Having codes for each and every rare disease would help obtain a better knowledge of healthcare pathways and of their impact on specialised health care services and on budgets.

#### ICD10

International Classification of Diseases 10th edition published by the World Health Organisation and used by more than 100 countries across the world. Widely used in the UK, especially in secondary care. Whilst accurate, the value of ICD10 coding for rare diseases is extremely limited as there have been numerous changes and advancements for rare diseases since the inception of ICD10 in 1994. Additionally, ICD10 is not designed to collect at a discreet enough level for rare disease and is not good at separating out the thousands of rare diseases and disorders that exist.

Due to the limitations of the 10th edition, the ICD 11th revision is due by 2018 and may assist in rare disease coding if adopted, however, improved rare disease coding does not only rely on a more suitable coding system but also depends on sufficient clinical, clerical and coding resources that allows detailed, accurate and consistent coding.

#### Read Codes

Standard clinical terminology system used in General Practice throughout the UK and maintained by the UK Terminology Centre, a division within NHS Digital. It supports detailed clinical encoding of multiple patient incidences. The updating of Read codes will cease in April 2018.

#### Hard Coding

Suspected / Carrier recorded against the condition tested for.

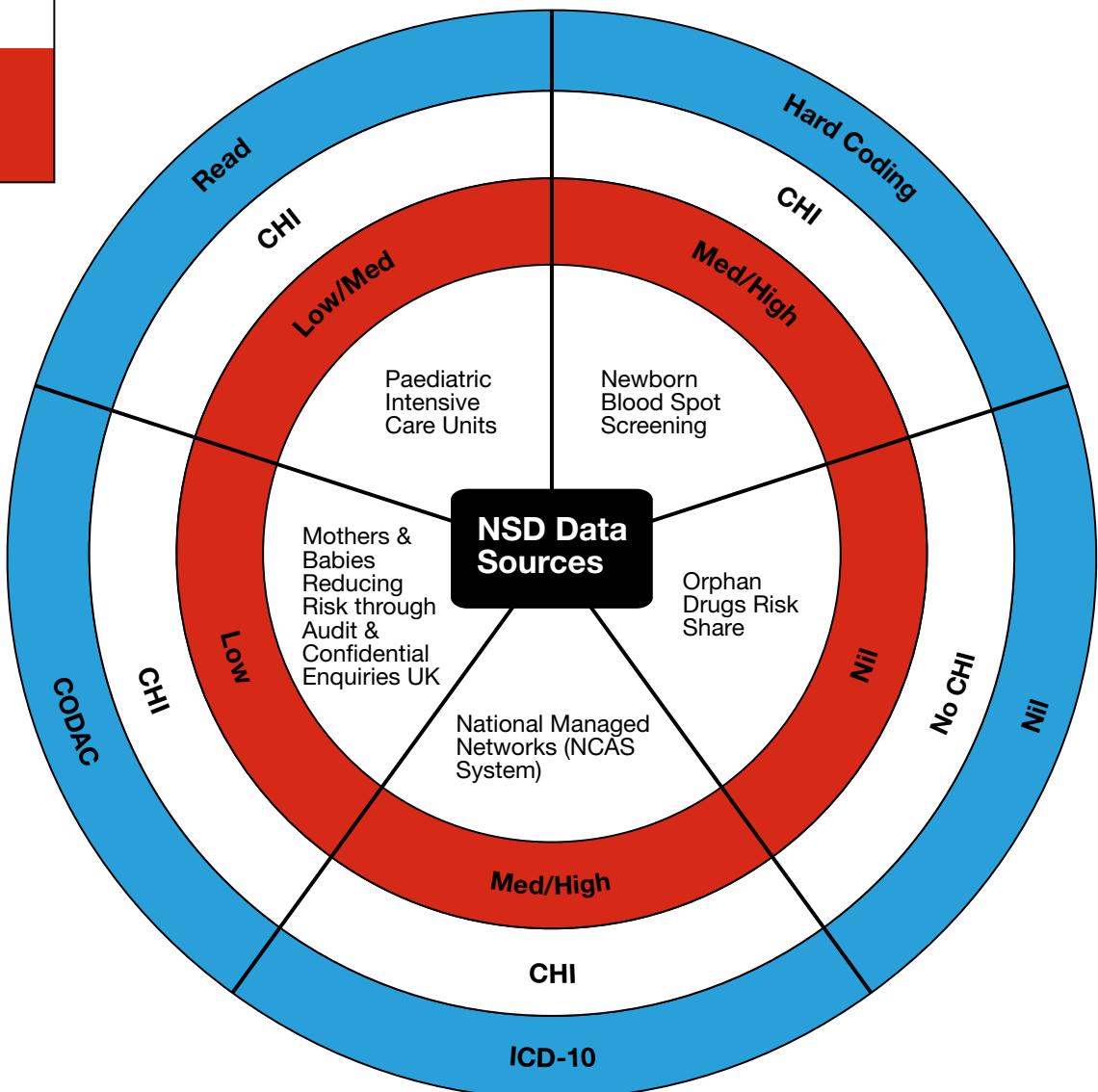
#### ERA-EDTA PRD (Coding for Primary Renal Disease)

The most recent ERA-EDTA coding system for Primary Renal Disease (PRD) includes codes and terms for most kidney diseases and not only for those that may lead to end-stage kidney failure. This coding system has the advantage of including the latest knowledge on kidney disease as well as direct linkage to ICD10 and SNOMED CT concept identifiers.

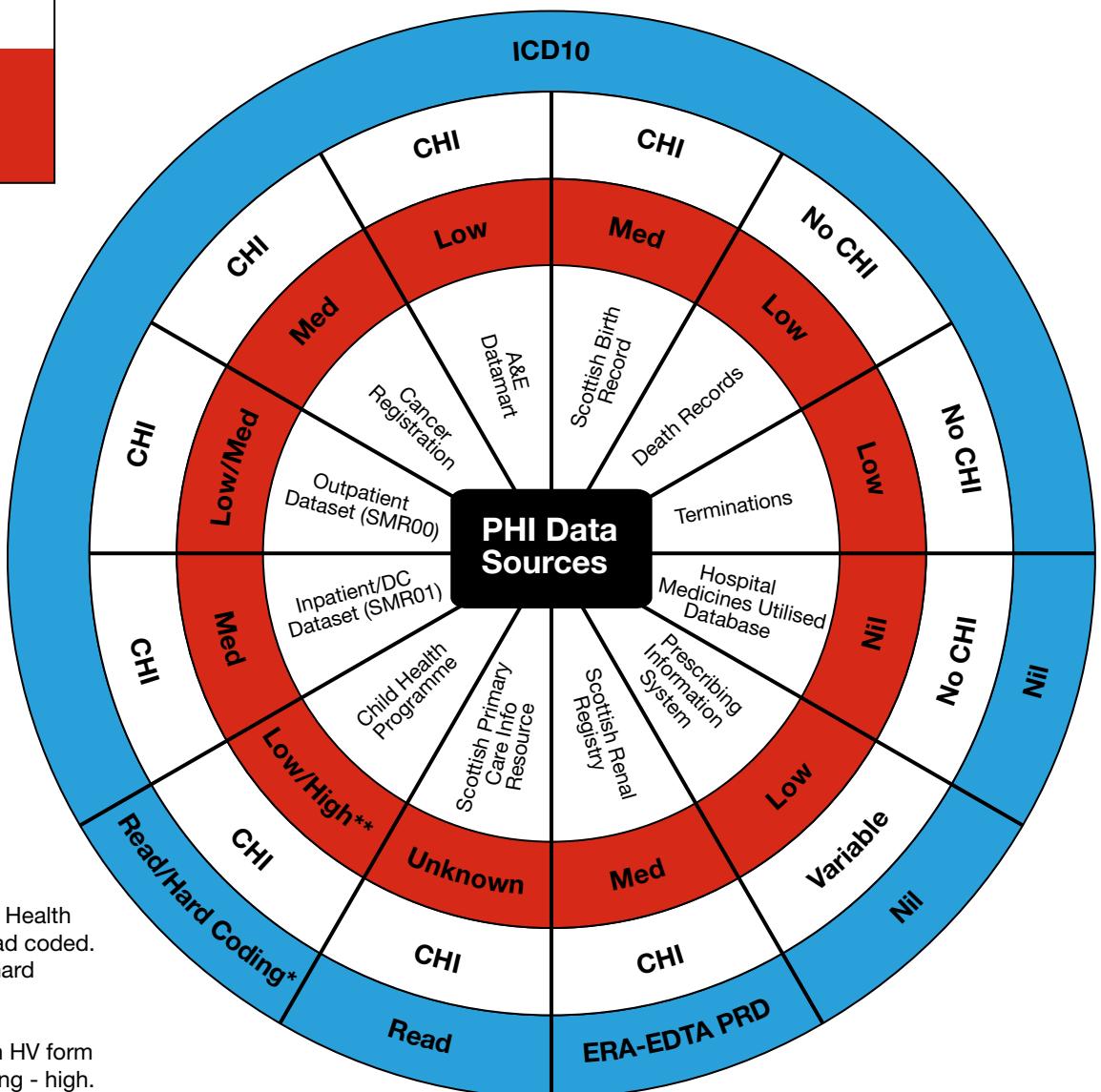
#### CODAC

Cause of Death and Associated Conditions is a tiered approach to the classification of perinatal deaths and can provide details of the stillbirths and neonatal deaths where the main cause of death was attributed to a congenital anomaly (CODAC level 1) showing CODAC level 2, which provides information about the system affected by the anomaly and CODAC level 3, which provides information about the specific anomaly.

Coding Used
CHI Capture
Usefulness for Rare Disease Capture



Coding Used
CHI Capture
Usefulness for Rare Disease Capture



\* Conditions recorded on Health Visitor (HV) forms are Read coded.  
Blood Spot Screening - hard coded.

\*\* Diagnosis recording on HV form  
- low. Blood Spot recording - high.

## ANNEX 3 – LIST OF EUROPEAN REFERENCE NETWORKS

Acronym	Full Title (all start “European Reference Network on”)	Co-ordinator Name	Country of co-ordination
<b>ERN PaedCan</b>	Paediatric Cancer (Haemato-Oncology)	St. Anna Kinderspital and St. Anna Kinderkrebsforschung	AT
<b>ERN LUNG</b>	Respiratory Disease	Universitätsklinikum Frankfurt	DE
<b>ERN-RND</b>	Neurological Disease	Universitaetsklinikum Tuebingen	DE
<b>ERKNet</b>	Kidney Disease	Universitätsklinikum Heidelberg	DE
<b>MetabERN</b>	Hereditary Metabolic Disease	Helios Dr Horst Schmidt Kliniken	DE
<b>ERN TRANSPLANT-CHILD</b>	Transplantation in Children	Hospital Universitario La Paz	ES
<b>ERN EYE</b>	Eye Diseases	Hôpitaux Universitaires de Strasbourg	FR
<b>ERN Skin</b>	Skin Disorders	Assistance Publique-Hôpitaux de Paris, Hôpital Necker-Enfants Malades	FR
<b>EuroBloodNet</b>	Haematological Diseases	Assistance Publique-Hôpitaux de Paris, Hôpital Saint-Louis	FR
<b>VASCern</b>	Rare Multisystemic Vascular Diseases	Assistance Publique-Hôpitaux de Paris, Hôpital Bichat	FR
<b>ERN EURACAN</b>	Adult Cancers (Solid Tumours)	Centre Léon Bérard	FR
<b>ERN BOND</b>	Bone Disorders	Rizzoli Orthopaedic Institute	IT
<b>ERN ReCONNECT</b>	Connective Tissue and Musculoskeletal Diseases	Azienda Ospedaliera Universitaria Pisana	IT
<b>ERN GENTURIS</b>	Genetic Tumour Risk Syndromes	Radboud University Medical Center Nijmegen	NL
<b>ERN GUARD-HEART</b>	Diseases of the Heart	Academisch Medisch Centrum bij de Universiteit van Amsterdam	NL
<b>ERNICA</b>	Inherited and Congenital anomalies	Erasmus Medical Center Rotterdam	NL
<b>Endo-ERN</b>	Endocrine Conditions	Leiden University Medical Center	NL
<b>ERN CRANIO</b>	Craniofacial Anomalies and ENT Disorders	Erasmus MC: University Medical Center	NL
<b>EpiCARE</b>	Epilepsies	Great Ormond Street Hospital for Children NHS Foundation Trust	UK
<b>ERN eUROGEN</b>	Urogenital Diseases and Conditions	Sheffield Teaching Hospitals NHS Foundation Trust	UK
<b>ERN EURO-NMD</b>	Neuromuscular Diseases	The Newcastle upon Tyne Hospitals NHS Foundation Trust	UK

<b>ERN ITHACA</b>	Congenital Malformations and Rare Intellectual Disability	Central Manchester University Hospitals NHS Foundation Trust	UK
<b>ERN RARE-LIVER</b>	Hepatological Diseases	The Newcastle upon Tyne Hospitals NHS Foundation Trust	UK
<b>ERN RITA</b>	Immunodeficiency, Autoinflammatory and Autoimmune Diseases	The Newcastle upon Tyne Hospitals NHS Foundation Trust	UK



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