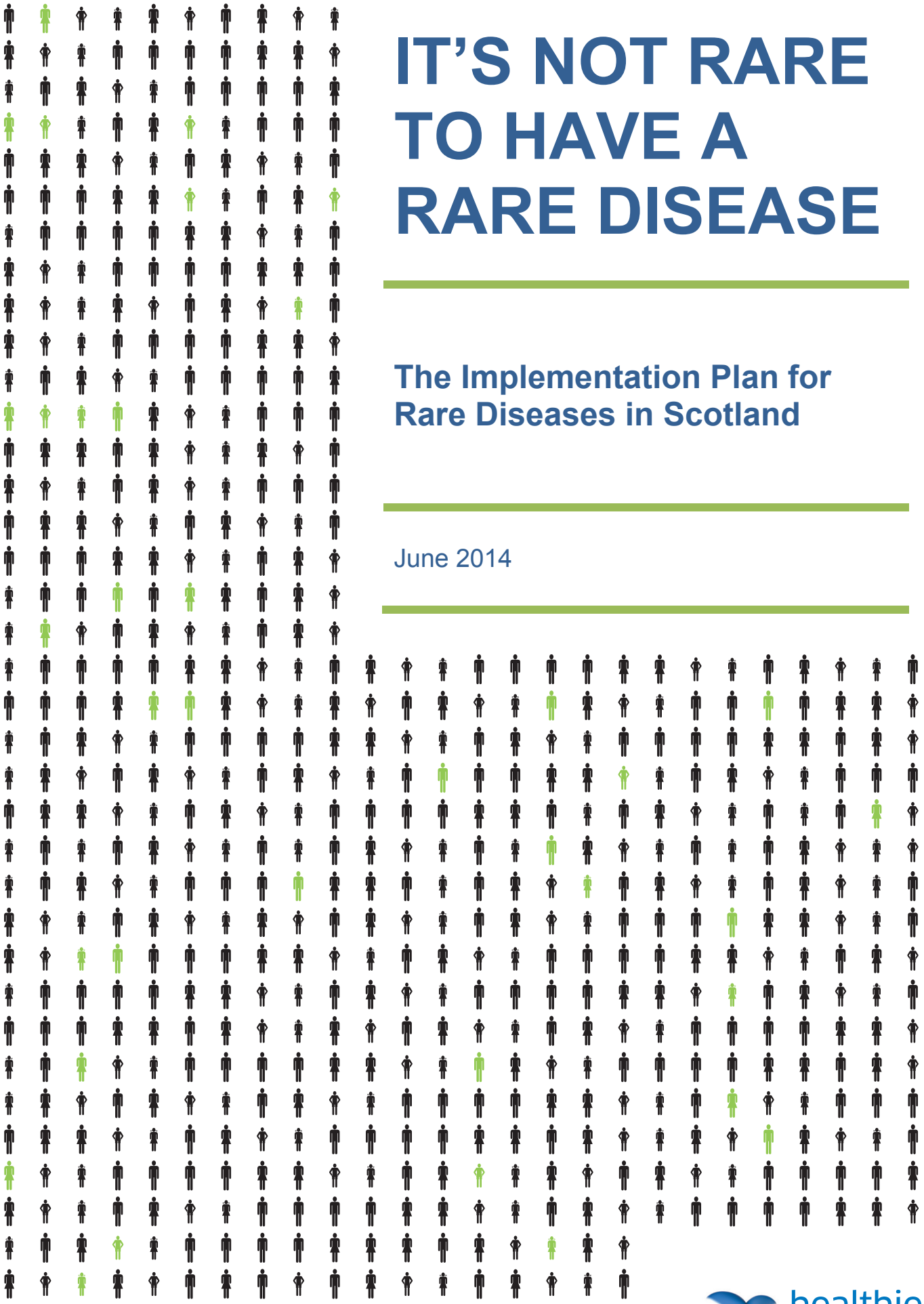


IT'S NOT RARE TO HAVE A RARE DISEASE

The Implementation Plan for Rare Diseases in Scotland

June 2014

healthier scotland
SCOTTISH GOVERNMENT

Contents

Foreword from the Cabinet Secretary for Health and Wellbeing

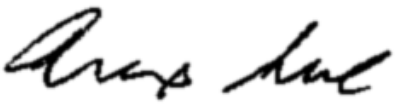
Rare Disease UK estimates that up to 300,000 people in Scotland's population may be affected by a rare disease over their lifetime. This is a significant figure. They also suggest patients with rare diseases in Scotland have experienced delays in gaining a diagnosis, with several getting a number of diagnoses on the way.

It is therefore important to emphasise our continued commitment to pursuing the three Quality Ambitions of safe, effective and person-centred care for all patients including those with rare disease and to deliver our 2020 vision, that by 2020 everyone is able to live longer healthier lives at home, or in a homely setting. In delivering this vision, we are committed to the provision of a healthcare system where we have integrated health and social care with a focus on prevention, anticipation and supported self-management.

Work on a UK Rare Disease Strategy was started as a partnership between the four countries of the UK. We jointly embarked on a consultation on plans to develop a *'UK Strategy for Rare Diseases'*[1], *w*hich was jointly endorsed by me and my UK counterparts at its launch in November 2013.

This Implementation Plan for Scotland reflects some of the excellent work of relevance to people living with rare diseases in Scotland carried out by partners from across the NHS, universities, enterprise, industry, social care and the third sector. It also acknowledges the importance of a timely and accurate diagnosis, allowing appropriate treatment to start as soon as they receive it, and allowing people with rare conditions and their families/carers to access support services across NHS Scotland and the third sector.

This is a challenging plan and it will take time to deliver over the next few years. I am sure however that by continuing to work together with patients as well as NHS professionals and our third sector colleagues we will make a difference.



Alex Neil
Cabinet Secretary for Health and Wellbeing

[1] www.scotland.gov.uk/Topics/Health/Services/RareDiseases

1. INTRODUCTION

The 5.3 million people living in urban, remote and rural settings in Scotland receive health and social care services through 14 NHS Boards and 32 Local Authorities operating across an area of 30,000 square miles, including almost 100 inhabited islands.

Rare Disease UK[2] estimates 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.[3] Many rare diseases present early in life.

A rare disease can be defined as *"…a life-threatening or chronically debilitating disease that affects 5 people or fewer in 10,000 and requires special, combined efforts to enable patients to be treated effectively."*[3]

The diversity of rare diseases, the specialist knowledge required to diagnose and treat the conditions and the geography of Scotland brings challenges for health and social care services and their partners in delivering services. Our remote and rural health delivery challenges can be magnified with rare conditions, as it may not be possible to make highly specialised service provision locally available to the dispersed population of people living with rare conditions.

We therefore worked in partnership with the other UK countries on the development of the UK Rare Disease Strategy[4] aiming to support earlier diagnosis, improve services, research and support for people of all ages living with rare diseases to deliver on the European Union (EU) Council Recommendation of 2009 for Member States to take action on rare diseases, namely:

> *"Establish and implement plans or strategies for rare diseases at the appropriate level or explore measures for rare diseases in other public health strategies, in order to aim to ensure that patients with rare diseases have access to high-quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan drugs..."*[5]

The 2012[6], UK-wide consultation in advance of the UK Rare Disease Strategy received 300 plus responses. The UK Rare Diseases Strategy drew on this consultation in developing the UK response to the issues affecting people living with rare diseases, seeking to:

- promote equity of access – allowing everyone with a rare disease to follow a clear, well defined care pathway, providing high quality services for every individual through integrated personal care plans
- offer a patient centred, co-ordinated approach to treatment services, specialist healthcare and social care support which takes into account the needs of the patient, their family and others who provide essential support
- deliver a holistic approach to diagnosis and treatment of rare diseases, developed through the best use of local resources that are easily accessible by patients and professionals

[2] RDUK – a voluntary sector group representing people with rare diseases and their supporters - www.raredisease.org.uk
[3] www.raredisease.org.uk/documents/Research%20Report/rduk-scotland-survey-report.pdf
[4] http://www.scotland.gov.uk/Resource/0043/00438631.pdf
[5] www.eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF
[6] www.gov.uk/government/uploads/system/uploads/attachment_data/file/146814/dh_132883.pdf.pdf

- support specialised clinical centres offering excellence in clinical care, expertise and patient support
- require these centres to offer care and expertise to families, the multi-professional health care team, and health service management
- promote excellence in research to develop understanding of and treatment for rare diseases
- secure rapid and effective translations of advances in the understanding of rare disease into clinical care by creating appropriate infrastructure, care pathways and clinical competences
- deliver effective interventions and support to patients and families quickly, equitably and sustainably
- promote collaborative working between the NHS, research communities, academia and industry wherever possible to facilitate better understanding about rare diseases and how they can be treated
- have education and training programmes in place that enable health and social care professionals to better identify rare diseases to help deliver faster diagnosis and access to treatment pathways for patients: and
- promote the UK as a first choice location for research into rare diseases as a leader, partner and collaborator.

Each of the four countries committed to developing an implementation plan for the UK Rare Disease Strategy, reflecting their individual health service structures and priorities for action to deliver the agreed commitments.

Work on national rare disease plans is also underway across the EU and many countries have published plans. Where relevant there is existing inter-country and EU-wide collaboration. It is envisaged that more collaboration will develop as rare disease work progresses.

Rare Diseases in Numbers[7]

- An estimated 300,000 people in Scotland are potentially affected by a rare disease (up to 3.5m in rest of UK and up to 29m across the EU)
- There may be between 6,000 and 8,000 rare diseases.
- Around 250 new rare diseases are discovered every year
- Around 80% of rare diseases are genetic in origin
- 350 rare diseases affect 80% of all rare disease patients.

[7] Source Eurostat - http://epp.eurostat.ec.europa.eu

2. THE STRATEGIC CONTEXT

The NHS Scotland 'Quality Strategy'[8] and the associated '2020 Vision and its accompanying Route Map'[9], aim to deliver the highest quality of healthcare to the people of Scotland. Together they provide common principles through which the NHS, Local Authorities and the third sector, working together and with patients, carers and the public, can deliver a shared goal of world-leading healthcare; ensuring services are; safe, person-centred and effective.

Our workforce is key to the delivery of quality services. The 'Everyone Matters: 2020 Workforce Vision Implementation Framework'[10] sets out 5 priorities for action by NHS Boards: healthy organisational culture; sustainable workforce; capable workforce; integrated workforce; effective leadership and management.

This Implementation Plan for Rare Diseases in Scotland supports the drive to deliver the Scottish Government's 2020 Vision that by 2020 everyone is able to live longer healthier lives at home, or in a homely setting. By 2020 we will have a healthcare system where we have integrated health and social care, a focus on prevention, anticipation and supported self-management.

The Scottish Government has also set up a Task Force to consider seven day working. Many services are already delivered seven days a week and changes towards extending care are already happening in some areas, but the Scottish Government wants to accelerate the pace of change, by targeting enhanced weekend and out of hours services that will benefit patients the most. Additional clinical and non-clinical support, at key points over the weekend and overnight, will be focussed towards improving service provision and patient flow.

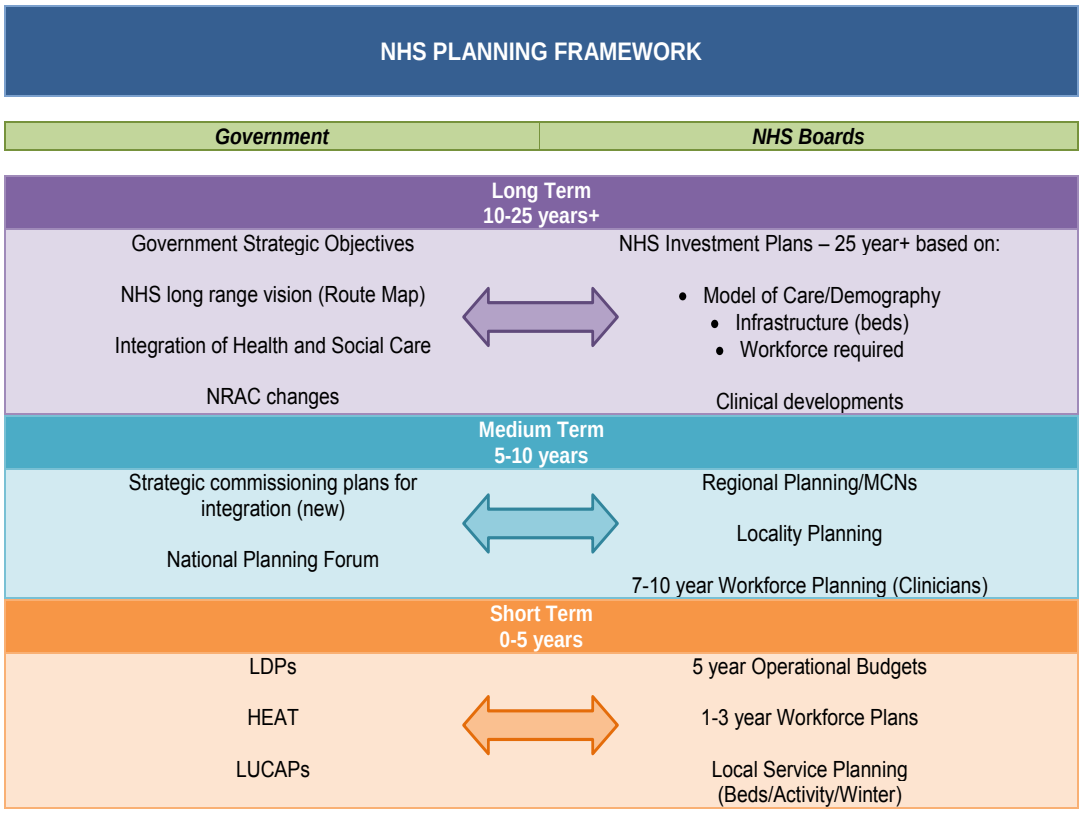
NHS Scotland Organisational Structures – Local, Regional and National

The Scottish Government has responsibility for the National Health Services in Scotland and its resource allocation. Legislation on the NHS in Scotland is made by the Scottish Parliament. The Cabinet Secretary for Health and Wellbeing has ministerial responsibility in the Scottish Cabinet for the NHS in Scotland.

The Scottish Government sets national objectives and priorities for the NHS, agrees delivery plans with each NHS Board and Special Board, monitors performance, and supports the Boards to achieve their objectives (figure 1).

[8] www.scotland.gov.uk/Resource/Doc/311667/0098354.pdf
[9] www.scotland.gov.uk/Resource/0042/00423188.pdf
[10] www.scotland.gov.uk/Resource/0043/00439819.pdf

Figure 1



NHS Scotland strives to deliver equity of access, safe, effective and person centred care for all who need it across all age groups. The existing care and support services reflect many of the elements of the UK Strategy's[11] vision and are delivered through a range of local, regional and nationally provided services through local NHS Boards and NHS National Services Scotland National Specialist and Screening Services Directorate (NSD).

The 14 'territorial' NHS Boards in Scotland are unified organisations which plan, commission and deliver NHS services and take overall responsibility for the health of their populations. Their responsibilities cover hospital and community health services including services provided by GPs, dentists, community pharmacists and opticians, who are independent contractors.

Seven national or 'special' NHS Boards provide national support services and Healthcare Improvement Scotland provides the scrutiny and public assurance of Scotland's health services.

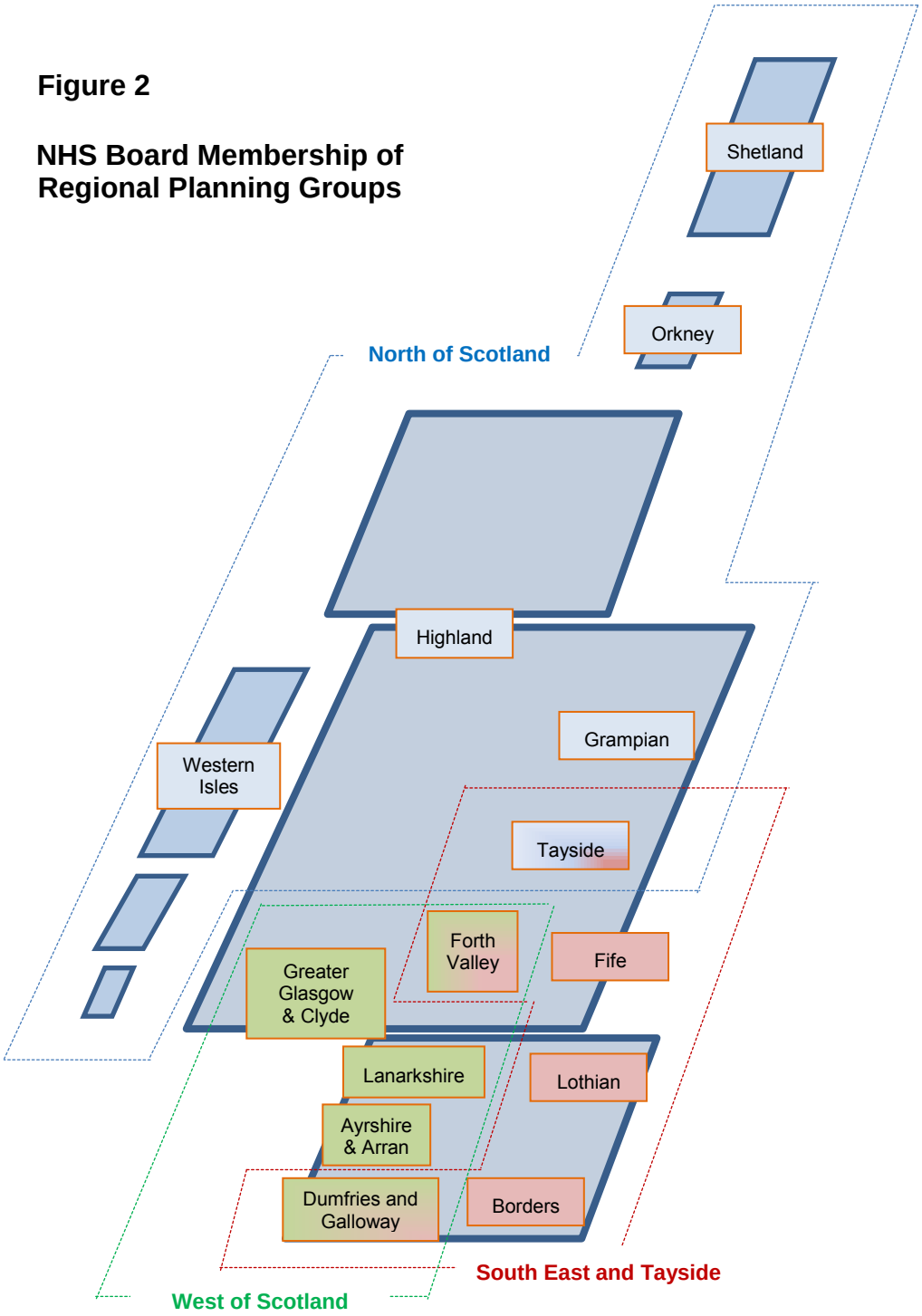
The NHS Boards work closely with partners including patients, staff, local authorities and the third sector to deliver effective healthcare services and to safeguard and improve the health of their residents.

[11] www.scotland.gov.uk/Resource/0043/00438631.pdf

NHS Boards work together regionally, in three planning groups (figure 2) covering: South East and Tayside; West of Scotland and North of Scotland. They also work together nationally to plan and commission specialist healthcare services such as heart and lung surgery, neurosurgery, and forensic psychiatric care. A number of local services are also shared between NHS Boards to maximise efficiency.

Figure 2

NHS Board Membership of Regional Planning Groups



The NHS Boards are guided by an annually reviewed ‘Local Delivery Plan’ (LDP) agreed by each Board with Scottish Government. The LDPs and associated Improvement and Co-production Plans provide a means for NHS Boards to set out and to be reviewed against their plans to deliver against local and national priorities, including service developments.

The Public Bodies (Joint Working) (Scotland) Act was granted royal assent on April 1, 2014. The Act is a landmark in health and social care reform for Scotland which will require NHS Boards and Local Authorities to integrate their services resulting in more joined-up, seamless health and social care provision closer to home to improve people's lives.

There are two integration models available to NHS Boards and Local Authorities allowing the choice of model to be selected based on local needs:

> Option 1 - The NHS Board and Local Authority delegate the responsibility for planning and resourcing service provision for adult health and social care services to an Integration Joint Board.
>
> Option 2 - The NHS Board or the Local Authority takes the lead responsibility for planning, resourcing and delivering integrated adult health and social care services.

These developments and associated guidance also support the new Health and Social Care Partnerships as they take on an important role in delivering integrated services.

NHS National Services Scotland, through NSD manages specialised commissioning and service provision across a wide range of highly specialised services, some of which are nationally designated and provided within NHS Scotland. Others are provided by designated services/bodies in NHS England. Some of these highly specialised services work in the area of one or more rare diseases. All have an important role in supporting people with rare diseases.

It is most likely that anyone requiring investigations to identify a specific disease will be referred to local diagnostic services through a specialist service or directly by their GP (as appropriate) for a specific investigative procedure or tests. However, dependent on the eventual diagnosis, individuals will be assessed to ensure the most appropriate and effective treatment is made available. This may not be available locally if the treatment/service concerned is highly specialised. People may be referred through specialist centres regionally or nationally within Scotland or they may need to access services through designated centres in NHS England, or further afield, dependent on where the required service is provided.

Examples of specialist services in NHS England accessed by people living in Scotland include alkaptonuria, complex neurofibromatosis type 2; rare mitochondrial diseases for adults and children; rare neuromuscular disease; and amyloidosis.

The elements of NSD's work in support of rare diseases are:

National Specialist Services – Before services are nationally designated their clinical and cost effectiveness is considered by two key committees, the National Patient, Public and Professional Reference Group (NPPPRG) who advise the National Specialist Services Committee (NSSC). All NSSC recommendations are referred to NHS Board Chief Executives' prior to submission to Scottish Ministers for approval. NSD also commissions on behalf of all NHS Boards some UK Specialist Services for adults and children. These services cover medical, surgical and scientific provision for those rare conditions where access to specialist expertise and treatment is not available in Scotland.

National Screening Programmes – These are aimed at early identification of a range of diseases/conditions, to allow clinical services to initiate early treatment and follow up. The UK National Screening Committee (UKNSC) provides advice to all four UK Governments on the adoption of new national population screening programmes or changes to existing ones. It is a matter for each country to consider UKNSC recommendations. Newborn screening in Scotland covers; Phenylketonuria; Congenital Hypothyroidism; Cystic Fibrosis; Medium Chain Acyl-CoA Dehydrogenase Deficiency and Sickle Cell Disease.

National Managed Clinical Networks (NMCNs) – Link together health professionals, patients, third sector, service managers and other partners to improve access to and performance of specific services for people, who often have complex needs. The NMCNs are required to deliver continuous quality improvement, pursuing opportunities to achieve better value for money through the delivery of optimal, evidence-based care that adds value from the patient's perspective, optimises productivity and reduces unwarranted variation.

Managed Diagnostic Networks (MDNs) – There are four Scotland-wide networks covering pathology, imaging, clinical biochemistry and microbiology/virology. These networks aim to deliver improved and quicker diagnosis and service efficiencies by quality improvement work and reduction of waste, harm and variation.

Other structures in Scotland have an important role to play in supporting people with rare diseases:

The National Specialist Services Committee (NSSC) considers proposals for national commissioning of highly specialist health services (described above) applications for National Managed Clinical Networks. Each element of this provision is reviewed every 3-5 years to ensure service arrangements remain relevant.

The Scottish Molecular Genetics and Cytogenetics Laboratory Consortium is the forum through which service developments in genetic testing are agreed. The genetic diagnostic laboratory services ensure equitable access to a comprehensive range of diagnostic genetic tests (provided within and outwith Scotland). Developments in this area are overseen by the Molecular Genetics and Cytogenetics Consortium and the Molecular Pathology Consortium.

Many rare diseases are genetic in origin and require specialist genetics input to diagnosis and treatment. The consortia link the four regional genetics centres in

Aberdeen, Dundee, Edinburgh and Glasgow. This approach provides genetic testing for a wide range of conditions.

The Molecular Pathology Consortium is the forum for collaborative discussion and decision making on molecular pathology testing in Scotland agreeing which tests will be provided in Scotland, in which laboratories and the priorities for introducing new tests. The group aims to ensure that everyone in Scotland has access to the same high quality and timeliness of molecular pathology tests, regardless of geographical location.

The Scottish Government's Innovation Partnership Board brings industry and health interests together to advance Scotland as a world leading centre for innovation and health in medicines, medical technology and digital health.

The Scottish Government is a member of the UK Rare Diseases Forum established by Department of Health (UK) to monitor how the vision and actions outlined in the UK Strategy for Rare Diseases are being responded to across the UK.

Palliative Care

Some people with life limiting and progressive rare diseases may require palliative care support. This is defined by the World Health Organisation as *"…an approach that improves the quality of life of patients and their families facing the problem associated with life-threatening illness, through the prevention and relief of suffering by means of early identification and impeccable assessment and treatment of pain and other problems, physical, psychosocial and spiritual."*[12]

Related to this, specialist palliative care[13] is the active total care of patients with progressive, advanced disease. Care is provided by a multi-professional team who have undergone recognised specialist palliative care training to provide physical, psychological, social and spiritual support.

Living and Dying Well[14] was Scotland's first national action plan for palliative and end of life care and set the direction for service planning and delivery. Work carried out as a result clearly demonstrated that there is a strong will to continuously improve and enhance the availability and quality of palliative and end of life care for all.

The Scottish Government through the development of a Strategic Framework for Action will provide a focus to support a wide range of organisations, clinical and care staff to reliably and sustainably spread good practice in high quality palliative and end of life care. The development of the Framework will be linked to the 2020 Vision for Health and Social Care[15].

The Scottish Government will work with a newly constituted National Advisory Group for Palliative and End of Life Care in supporting key stakeholders to apply the '3-Step

[12] www.who.int/cancer/palliative/definition/en/
[13] Tebbit, National Council for Palliative Care. 1999.
[14] Living and Dying Well (2008)
[15] www.scotland.gov.uk/Resource/0042/00423188.pdf

Improvement Framework for Scotland's Public Services'[16] to deliver the changes that they identify to support the Strategic Framework.

Following an announcement in December 2013 that the Liverpool Care Pathway will be phased out in Scotland during 2014, Interim Guidance on Caring for people in the Last Days and Hours of Life[17] was published. In recognition of the impact on staff and teams, work is underway to develop a national statement on the requirements for care in the last days and hours of life. This national statement will reflect the principles set out in the Interim Guidance and support NHS Boards to signpost and prompt clinical and care teams to develop local assessment and care planning materials, policies and guidance to be applied in all care settings.

Training, Education and Information

The UK Plan for Rare Diseases acknowledges that it is unrealistic for most clinicians to have detailed knowledge of treatment and management of every rare disease. This applies across all specialties, across the UK.

It is expected however that clinicians should be aware of the appropriate referral procedures within their own healthcare system. In Scotland this means clinicians should know how to make appropriate referrals for rare diseases within their own NHS Boards or to other NHS Boards and specialist services through NSD.

The Royal College of General Practitioners GP training curriculum includes genetics in primary care. The very rarity of individual conditions makes it unlikely many of these diseases will be seen by individual GPs over their working life. However, across the average practice population, a number of rare conditions may be managed. In some cases, conditions may present within individual families.

Decision support systems have the potential to support GPs to recognise key groups of symptoms which may suggest a rare disease and to refer patients to the appropriate specialists for diagnosis, for treatment plans and when advice is needed on the management of a patient's condition. In support of this, quality assured information on rare diseases is needed for all health practitioners. Such information would also benefit patients and families

NHS Boards, post graduate deaneries, NHS Education for Scotland (NES) and others undertake continuing professional development to meet GPs' educational needs.

Other staff training, education and support needs are addressed through various approaches. The 2020 Vision for Workforce encourages innovation among teams and individuals, supporting them to utilise new technology. It supports 'on-the-job' learning and provides time for and access to learning and development opportunities to develop professional and technical skills, knowledge and competencies. The associated implementation plan for 2014-15 sets out the initial arrangements to deliver workforce developments.

[16] www.scotland.gov.uk/Resource/0042/00426552.pdf
[17] www.scotland.gov.uk/Resource/0044/00444812.pdf

Awareness raising of rare diseases needs to be incorporated into training for the multidisciplinary healthcare team and it would be relevant to direct attention towards childcare and education settings also.

Developments in Education

NES (NHS Education for Scotland) is working to tailor education delivery methods to meet the educational needs of staff in services for remote and rural populations. The RRHEAL (Remote and Rural Healthcare Educational Alliance) provides an education platform through which remote, rural and island healthcare teams can access clinical and other support information.

The Scottish Genetics Education Network (ScotGEN) provides a range of high quality teaching materials for genetics education and patient information and has a wide range of information leaflets and support information on rare conditions of genetic origin.

Within NHS Scotland there are several National Managed Clinical Networks (NMCNs) for rare diseases and complex conditions. These networks are required to develop and implement an education strategy for health professionals and patients/carers. Many are now at the stage of developing online education packages in collaboration with NES.

The NMCNs also support the development and provision of information to support increased recognition. In paediatric epilepsy the network developed an individually tailored patient booklet for a child to take to school to ensure awareness of their needs. The network has also run awareness raising sessions for teachers. Similar work on the dietary needs of affected children was done in the metabolic network.

Infrastructure for Rare Disease Developments

Life sciences and technology innovation

Scotland has ambitions to develop its role as an international centre for innovation in health technologies and in life sciences to improve care for patients. These technologies, some of which are produced by Scottish-based technology and life sciences companies, hold promise to develop support and treatments across a range of health conditions, including for rare diseases.

Many Scottish universities have existing bio-park or similar ventures with NHS Board, life sciences and enterprise partners. These initiatives bring together universities delivering internationally recognised research, a possibly unique integrated health service, with universal population coverage and cutting edge innovation companies.

In 2012, *'Health and Wealth in Scotland: A Statement of Intent for Innovation in Health'*[18] was launched. A blueprint for innovation this consolidates existing partnerships between NHS Scotland, academic institutions, industry and enterprise.

[18] www.scotland.gov.uk/Resource/0039/00396711.pdf

Some of the current work of Scottish life sciences and innovation companies is of potential relevance to rare diseases.

Health Technology Assessment

The Scottish Health Technologies Group (SHTG) provides clinical and cost-effectiveness advice on health-related technologies. This advice supports the planning and decision-making processes nationally and in NHS Boards. SHTG also provides early information on health technologies in development.

The SHTG links to European and international health technology networks and in common with developments in other health technology agencies is moving from the traditional health technology assessment to rapid reviews of evidence.

Telehealth and telecare technology

Telecare systems are becoming established in the community, offering support to and monitoring of elderly and disabled patients and those with long term conditions. Telecare also provides patients with support to self-manage health conditions and to live independently.

Telehealth is increasingly used to link patients to clinical expertise using video and other technologies. Clinician and patient may be some distance apart, but real-time video footage, diagnostic images and test results can be available to inform the consultation and resulting treatment.

As Scotland's health service buildings and facilities are refurbished or replaced, there is investment in the installation of modern health and communication technologies for use in screening, in diagnostics, in treatment, in monitoring and in follow-up.

A National Telehealth and Telecare Delivery Plan for Scotland to 2015 - Driving Improvement, Integration and Innovation[19] sets out actions to develop this provision to deliver a range of services as alternatives to traditional service delivery models.

Following the integration of health and social care and the focus on joint support to patients, shared information systems will be crucial in ensuring support to patients is effectively and efficiently co-ordinated across health, social care and partners.

Information systems, data and registers

Information systems, registers and the data within them are important in the efficient delivery of modern healthcare. With the integration of health and social care, patients with a rare disease will potentially be seen across a number of healthcare and social care settings, which may have different electronic clinical and care systems. The existing variation between systems, confidentiality and firewalls create difficulties in data sharing across NHS Board boundaries.

[19] www.scotland.gov.uk/Publications/2012/12/7791

Through locality planning and joint working between health and social care professionals, information systems are being considered to improve data exchange and to facilitate positive outcomes for people across the spectrum of disease, including rare diseases. As clinical and social care pathways begin to be developed jointly they will need the support of robust IT platforms.

Work on IT developments in this area is covered by the Person-Centred e-Health Strategy[20]. In addition, NHS Boards are progressing the development of patient portals to give access to services in secondary care.

National Services Scotland (NSS) Public Health and Intelligence (PHI - formerly Information Services Division) has a role in developing data standards and coding, building on its well established work in these areas.

Electronic Health Data

Work is progressing well to have a pan-Scotland service available to support the use of routinely collected electronic health data. Through its participation in and co-ordination of the UK e-HIRC (E-Health Informatics Research Centre) network, Scotland is helping to deliver the goal of being able to combine the outputs of the UK datasets.

A Clinical Audit System was developed to provide databases for some conditions covered by National Managed Clinical Networks. This system is used to help health professionals work together in caring for patients.

PHI has considerable experience in linking patient records across healthcare settings. Although data linkage may be possible across different sectors, current variations in the recording of rare diseases and limitations in coding systems like ICD-10 makes it difficult at present to explicitly identify those patients with rare diseases.

PHI will have an important role in monitoring the development of ICD-11, the successor to ICD-10 and any subsequent roll-out across Scotland. As ICD-11 is still under development and may not be published until 2015 at the earliest, its introduction may not commence until 2016-17 or later. This means it cannot be relied on to identify rare diseases in the short term. Without the level of detail which may be available through ICD-11 when it is introduced, there may be limited examples of databases which appropriately record rare diseases. Account also needs to be taken of General Practice coding. In Scotland, the READ codes are used in General Practice. In England there is a planned move to a new system of coding by 2014.

In the main, NHS Boards determine the scope and rollout speed of their various e-Health approaches to suit their local circumstances. Progress is however monitored through the Scottish Government e-Health strategy.

Progress with patient portal developments is picked up during 6 monthly e-Health Plan reviews with each NHS Board.

[20] www.ehealth.scot.nhs.uk/wp-content/documents/personCentredStrategy.pdf

3. DELIVERING THE UK STRATEGY COMMITMENTS

This implementation plan seeks to ensure that the needs of people with rare diseases are reflected in health and social care service planning and delivery across Scotland.

It aims to provide appropriate healthcare support to people with rare diseases through primary care, community, acute and specialist services and social care support, within existing service arrangements and the developing integrated health and social care arrangements.

The plan acknowledges that delivery of the actions requires continuing collaborative working between the NHS, public and third sector partners, researchers, academic institutions and industry. It also acknowledges that partnership working in rare disease research is key to providing diagnostic tools to reduce diagnostic delays and to pilot and develop treatment interventions.

The chapters which follow set out current activity across Scotland and potential actions, with leads and timescales to be agreed, for NHS Scotland and Scottish Government to deliver against the 51 commitments of the UK Strategy for Rare Diseases across its 5 themes:

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

During 2014 Scottish Government will therefore establish an Implementation Oversight Group to among other things key themes/actions and metrics to assess progress/delivery of the Implementation Plan over agreed timescales.

A range of themes have emerged from the work to develop this rare disease plan for Scotland. Many suggested actions have also emerged and these are set out at the end of each chapter under 'Suggested Next Steps'. These suggested actions are grouped under the various themes that have emerged from wide-ranging discussions and feedback from NHS Scotland, third sector colleagues and patients.

It would be neither sensible nor possible to put in motion all the action suggested nor to put timescales against these as part of this plan. It is also clear that many of these themes are a recurring thread throughout and will need to work across several clinical and other areas to achieve.

We are therefore presenting these as a range of suggested actions at the moment, however we will need to agree in collaboration how best to bring these together into a smaller set of deliverables that are capable of being delivered and importantly can be monitored to capture progress over time

The first task for the Rare Disease Implementation Oversight Group will be to bring forward a timed, deliverable and sustainable set of high level actions to encourage, support and deliver the changes we all want to see in rare diseases.

4. EMPOWERING THOSE AFFECTED BY RARE DISEASES

UK Strategy Commitments

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 diseases 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 diseases.

Where we are now

The Scottish Government is committed to supporting person-centred care at all stages of the patient journey and patient and public involvement in the development of healthcare services to help deliver better outcomes.

Through the Patient Rights (Scotland) Act 2011[21] and our work to develop more person-centred health and care services we are giving people more voice on their experiences to drive quality improvement in health, to promote personal responsibility for health and wellbeing and to support self-management. This will include providing support to people to help them navigate and understand the health and social care systems, so that they become more involved and engaged in their healthcare design and delivery of services. This should help people in maintaining their health and in managing periods of ill-health. There are various legislative, policy and structural arrangements which support these approaches.

[21] www.scotland.gov.uk/Topics/Health/Policy/Patients-Rights

Since 2012, the Act has given patients the right that the healthcare they receive:

- considers their needs;
- considers what would be of optimum benefit to them; and,
- encourages them to participate in decisions about their health and provides information and support for them to do so.

The Patient Rights (Scotland) Act 2011 also established and provided access for patients and members of the public to the independent Patient Advice and Support Service (PASS) which provides information and helps raise awareness and understanding of their rights and responsibilities when using health services. PASS also helps and supports patients to give feedback about their healthcare and directs them to other types of support, such as advocacy or communication support services.

The NHS Reform (Scotland) Act 2004[22] places a duty on all NHS Boards to involve the public (patients, carers or members of the public from a range of perspectives) in the planning and design of health care services and policies. This applies to all patients of all ages, including those with rare diseases.

In addition, the Charter for Patients' Rights and Responsibilities published in October 2012[23] states that:

- [patients] *"…have the right to be involved, directly or through representatives in the planning, design and provision of services…"*
- *"Health Boards must involve people who live in their Board area in the planning and development of services, and in decisions that significantly affect the operation of those services."*

The Scottish Health Council promotes public involvement in the NHS in Scotland ensuring that NHS Boards listen to and take account of people's views and work in partnership with patients, carers and the public. The Scottish Health Council's local offices in each of the NHS Board areas link closely with local, regional and national Managed Clinical Networks to promote public involvement in the NHS in Scotland.

The Scottish Health Council provides support to strengthen engagement mechanisms, promotes awareness amongst service providers of the importance of public engagement, encourages liaison between the NHS and local communities and supports patients and the public in having a say in the planning, evaluation and monitoring of services. This work is supported by Participation and Evaluation Toolkits.

Third sector organisations also have an important role to play in supporting patient empowerment and in facilitating peer support.

[22] www.legislation.gov.uk/asp/2004/7/section/2
[23] www.nhsinform.co.uk/Rights/UsingNHS/PatientCharterSummary

Developments in Patient Empowerment

Across a range of health conditions there are a variety of excellent models for patient involvement and information provision which could be extended across rare diseases.

Managed Clinical Networks are key to facilitating and supporting patient involvement in decisions. In some areas, patient representation comes from the third sector, for example in the Scottish Cancer Coalition and the Respiratory National Advisory Group.

The Neurological Alliance of Scotland involves patient representatives in a 'Neurological Voices Programme'. This is run in partnership with NHS Boards. It supports people with neurological conditions and their carers to get involved in planning and improving neurological health services, with a focus on the relevant NHS Scotland Clinical Standards.

In diabetes services, the chair of an active patient subgroup is also on the national Diabetes MCN steering group.

In designated national specialist services patients are involved in annual performance reviews of specialist services and in the regular 3-5 year planning reviews. The Scottish Genetics Education Network has a wide range of information leaflets and support information on rare conditions of genetic origin.

The Person-Centred Health and Care Collaborative[24] is a key part of a Scotland-wide programme of work aimed at improving health and care services to focus on people, their families and carers. The collaborative aims to bring together people from every NHS Board to listen to the experiences of people who use services and to use their feedback to drive improvement and make care more person-centred.

NHS Boards already have structures in place to support the involvement of the people and communities they serve. Currently public partnership fora exist within each NHS Board. With the replacement of Community Health Partnerships by Health and Social Care Partnerships, the future role of Patient Public Forums is being considered, to ensure they continue to discharge their important role in encouraging patient groups to engage with the new bodies.

There are many good examples of patient involvement in the development of NHS services that can be built on. For example NSD's Professional, Public & Patient Reference Group (NPPPRG) which advises on commissioning of highly specialist services for rare diseases and complex conditions has an individual patient member as well as involvement from Genetic Alliance UK.

The Alliance', as Scotland's third sector umbrella organisation, represents the views of its members when collaborating with NHS Scotland and Scottish Government.

[24] www.healthcareimprovementscotland.org/our_work/person-centred_care/person-centred_collaborative.aspx

Capturing Patient Views

Completely independent of government and the NHS, the Patient Opinion website is where patients, their carers or family members can tell other people about their experiences of the NHS. NHS Boards are alerted to stories posted about services in their area and are encouraged by the Scottish Government to post responses saying what they have done in light of what patients have said. It aims to make it easier for people to give feedback and for NHS Boards to get those opinions to the people who need to see them and ultimately, to make services better.

This can be done online at www.patientopinion.org.uk or by telephone on: 0800 122 31 35.

Scotland's four NHS Clinical Genetic Centres aim to help develop innovative, seamless services and to integrate patient and public views into service development and care guidelines. The Single Gene Complex Needs[25] (SGCN) service was established following the recommendations of the 'Review of Genetics in Relation to Healthcare in Scotland'[26]. The review recognised the crucial role that genetic services had in the diagnosis, assessment and treatment of individuals living with complex needs. The review also acknowledged that these patients and their families often had much broader complex health and social care needs.

Support in Genetics

The Single Gene Complex Needs service (SGCN) works primarily with adults living with rare diseases of single gene origin throughout Scotland, supporting individuals and their families to manage their care needs in their local communities.

SGCN works mutually with a wide range of partners from the third, health and social care sectors, to design, develop and deliver anticipatory services to improve outcomes for people living with single gene disorders. The service also acts as a point of contact between the health service, social services, the third sector, education and employers.

Patients who have been referred to the service have presented collectively with 112 different conditions of single gene/chromosomal origin as well as a small number of unspecified diagnoses.

The Key Information Summary (KIS) is in use across all GP practices in Scotland. It contains details of staff involved in a patient's care, main diagnosis and current issues, carer and support details and recommended actions for out of hours services

[25] www.pcgs.org.uk/SGCNcombined.pdf
[26] www.scotland.gov.uk/Resource/Doc/146336/0038294.pdf

The Patient and Public Involvement Group of the Scottish Medicines Consortium (SMC) is reviewing its activities and work plan in response to the Scottish Government's requirements following the Access to New Medicines Review.

Updated guidance issued in July 2012[27], recognises that Managed Clinical Networks (MCNs) have a strong foundation of working effectively with the third sector.

Criteria for National Managed Clinical Networks include requirements to involve service users in their work, to capture service users' views and to have policies to improve access to services and information dissemination (figure 3).

Patient Access to Information

NHS inform is Scotland's national health information service. This web and telephony based service provides quality assured information on a wide range of health and care related topics including information on over 800 medical conditions.

ALISS (A Local Information System for Scotland) is a search and collaboration tool for health and wellbeing in Scotland, helping signpost people to useful community support.

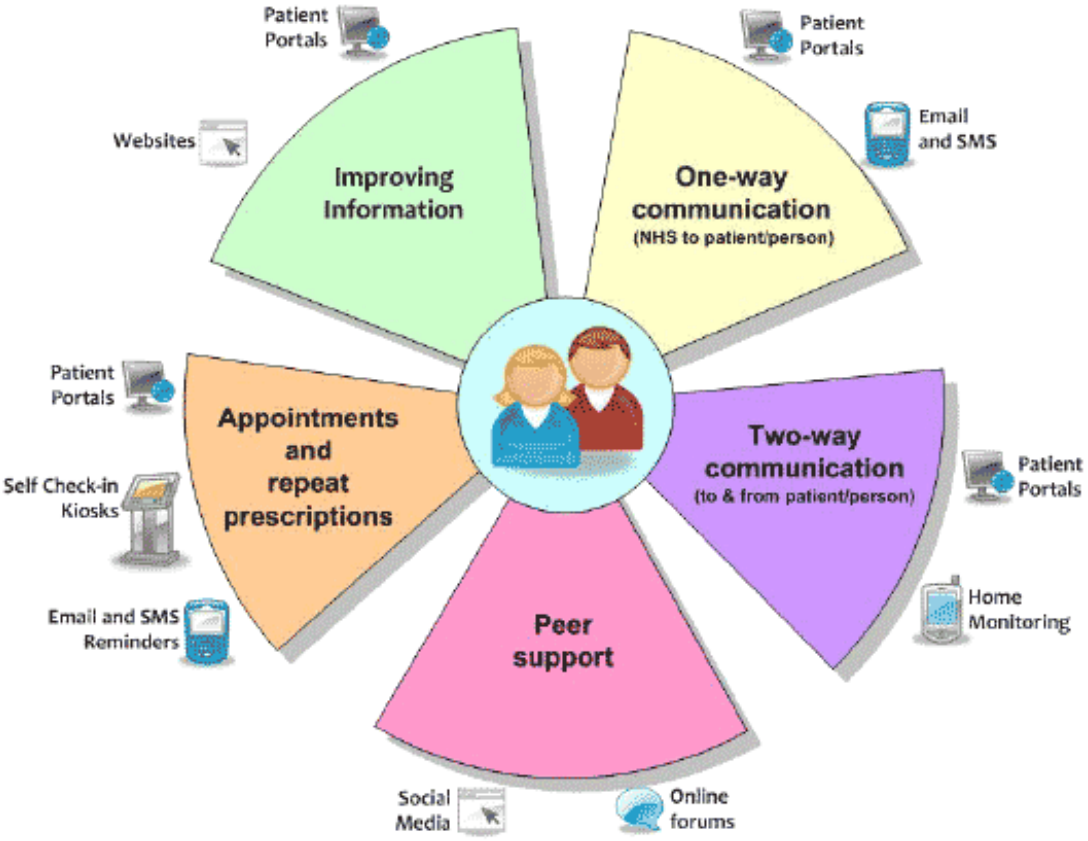
'Renal Patient View' and 'My Diabetes My Way' patient portals provide examples of how to provide more personalised support and information to patients with specific conditions.

Advances in technology and social networking provide opportunities for widening channels for information exchange i.e. Facebook, twitter, blogs and patient forums all provide useful platforms for information and peer support.

[27] www.sehd.scot.nhs.uk/mels/CEL2012_29.pdf

Figure 3

Information and Communication
Support to Patients



Suggested Next Steps Emerging from Discussion/Engagement with Stakeholders

Patient Involvement

We will improve rare disease patient involvement and engage with rare disease service users at all stages in service planning, delivery and evaluation through:

- The creation of opportunities for patients with a rare disease to participate in decisions about all aspects of their care, support and treatment
- The use of existing NHS Board patient and public involvement structures and the Health and Social Care Partnerships arrangements as these develop
- Consideration of how the Person-Centred Health and Care Collaborative can best take into account the needs of people with rare diseases
- Publicising the Patient 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.

Patient Records and Registers

- We will explore with relevant agencies the applicability/practicability of registers and the opportunities to develop a patient electronic health record.

Information for Patients

We will improve information provision to patients with rare disease through:

- Opportunities arising from the Person-centred e-Health Strategy and Delivery Plan
- 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
- Staff in appropriate roles seeking to signpost individuals with rare disease to further information on research and clinical trials and opportunities to participate.

Information for Staff

Staff will be supported to obtain quality information on rare diseases through:

- Consideration by NHS Education Scotland of what information, training and resources on rare diseases may be deliverable to staff, for example via the NES Knowledge Network.

5. IDENTIFYING AND PREVENTING RARE DISEASES

UK Strategy Commitments

9. Continue to work with the UK National Screening Committee to ensure that the potential role of screening in achieving earlier diagnosis is appropriately considered in the assessment of all potential new national screening programmes and proposed extensions to existing programmes.

10. Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular tests are evaluated and recommended by UKGTN (UK Genetic Testing Network) is accessible for at risk relatives.

Where we are now

Many rare diseases are present at birth and are either caused by genetic factors (for example sickle cell disease) or deficiencies or exposures to substances around the time of conception or during pregnancy (for instance, spina bifida is associated with a folic acid deficiency around conception and early pregnancy).

A World Health Organisation report in 2010[28] found that birth defects *"…can be life-threatening, result in long-term disability, and negatively affect individuals, families, health-care systems and societies."* and that early diagnosis and action can prevent complications and illness.

Screening has an important role in allowing early diagnosis of some rare diseases and the initiation of early treatment to reduce complications. The Scottish Government is represented in the UK National Screening Committee which makes its recommendations to all 4 Health Departments across the UK.

Potential new national screening programmes are rigorously assessed by the UK National Screening Committee before their introduction to assess the evidence for programmes against a set of internationally recognised criteria covering the condition, the test, the treatment options and the effectiveness and acceptability of the screening programme. This approach is intended to ensure that screening approaches do more good than harm at a reasonable cost. Any proposed extension to existing programmes is similarly assessed.

In NHS Scotland, screening policy is set by the Scottish Government Health Directorates on the advice of the UK National Screening Committee and other appropriate bodies. NSD commissions and co-ordinates important elements of national screening programmes helping to ensure consistent, effective, coordinated national screening programmes for the people of Scotland.

In some circumstances, case finding approaches may provide an alternative approach to the identification of individuals at risk of a specific disease to offer screening and if appropriate treatment.

[28] http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf

Newborn Baby Screening

In Scotland, newborn baby screening uses a blood spot test (the Guthrie card) for five conditions: Phenylketonuria, Congenital Hypothyroidism, Cystic Fibrosis, Medium Chain Acyl-CoA Dehydrogenase Deficiency and Sickle Cell Disease.

The Laurie report[29] notes that parents have the option to consent to *"…individual tests, all tests, or none of the tests."* If parents agree to screening, the blood spot card is retained, for a 12-month period. This consent can be extended beyond 12-months with parent permission. If no permission is given, the card is destroyed.

Retained blood spots have potential anonymous post-test use for *"…purposes such as comparing different screening methods and developing new tests."* Such usage of the blood spots and the genetic material they contain (dating back to 1965 and containing 2.5 million cards with blood/DNA samples/personal information) was the subject of review, reported on in the Laurie report.[29] This provides a commentary on the storage and potential future uses of the Guthrie cards held in NHS Scotland.

Four National Managed Clinical Networks (NMCNs) provide effective links between the Scottish Newborn Screening Programme and relevant clinical services.

In association with the national pregnancy and newborn screening protocols, counselling is provided to 'at risk' relatives on carrier status if applicable. There is access to genetic testing for at risk relatives in Scotland through the Scottish Genetics Centres.

Within the Scottish Molecular Genetics and Cytogenetics Laboratory Consortium work is focussing on the use of new techniques to allow increased range of genetics panel testing and this is planned to extend to exome sequencing within the next two years.

The Scottish Molecular Genetics and Cytogenetics Laboratory Consortium's current policy is for provision of small Next Generation Sequencing (NGS) devices in all 4 centres. This supports regional subspecialisation within the UKGTN and a distributed pattern of testing close to clinical decision making. Such an approach is sustainable, is considered appropriate to Scotland's geography and fits with the rapid advances in technology. There is close collaboration with Scottish universities.

NHS Education Scotland, National Services Division, the Molecular and Cytogenetics Laboratory and Molecular Pathology Consortia have recently reviewed the future needs for scientists to support future developments in testing and NES is facilitating the implementation of recommendations.

There is an untapped potential for public health services to explore 'upstream' prevention for rare diseases taking into account and acting on social and economic determinants of poor health amongst people with rare diseases.

[29] www.scotland.gov.uk/Resource/0044/00441799.pdf

Suggested Next Steps Emerging from Discussion/Engagement with Stakeholders

Public Health

- Along with their partners public health specialists in NHS Boards should explore options for appropriate interventions to address determinants of health in rare diseases.

Screening for Rare Disease

- Consider the introduction in Scotland of recommendations from the UK National Screening Committee
- Continue to ensure access for people in Scotland and families to UKGTN approved carrier testing, extending coverage as appropriate.

6. DIAGNOSIS AND EARLY INTERVENTION

UK Strategy Commitments

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.

12. Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients, by:

- establishing clear, easily accessible and effective pathways between primary care, secondary care, regional centres and specialist clinical centres, as appropriate
- putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment
- drawing on patients' ability to help inform decisions about referral and diagnosis
- creating effective clinical networks to support this process
- making high quality diagnostic tests accessible through common, clinically agreed systems or pathways
- embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies.

13. Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients.

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.

15. Improve education and awareness of rare diseases across the healthcare professions, including:

- involving patients in the development of training programmes
- encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics
- ensuring awareness of methods and clinical techniques used in differential diagnosis.

16. Monitor the development of ICD-11 in preparation for its adoption.

17. Work with colleagues in Europe in the development of the European Orphanet coding system and consider the adoption of Orphanet coding and nomenclature.

18. Standardise data collection, building upon existing NHS data standards, and develop standards where they do not exist, increasing the reliability of information for use in providing or commissioning care.

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.

Where We Are Now

Delays in diagnosis mean that opportunities for timely interventions can be missed. In addition, relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis. Diagnosis may also require specific expertise to interpret laboratory tests.

Healthcare Improvement Scotland is leading the work package on the development of a collection of good quality clinical guidelines and associated research recommendations for RARE-Bestpractices[30], which was initiated in January 2013 with funding from the EU Seventh Framework Programme. This project is designed to support access to diagnosis and provision of high quality healthcare for patients with rare diseases in line with the EU Directive 2011/24/EU on the application of patients' rights in cross border healthcare. The project is co-ordinated by the Italian National Centre for Rare Diseases.

RARE-Bestpractices will deliver a publically accessible database of good quality guidelines to provide up to date information on disease diagnosis and management for professionals, patients and carers. The project will also deliver: a methodology for the development of clinical guidelines in rare disease topics; a new Journal, 'Rare Diseases and Orphan Drugs'; training activities in the development, appraisal and implementation of guidelines; and assessment of approaches to appraisal of orphan medicines. The expertise within Healthcare Improvement Scotland (including the Scottish Intercollegiate Guidelines Network) in searching for and assessing the quality of guidelines has been pivotal to its involvement in RARE-Bestpractices.

Scotland has well developed diagnostic services, with collaborative working ensuring that there is equitable access to tests across the 14 territorial NHS Board areas. This work is supported by the four Scottish National Managed Diagnostic Networks which focus on delivering service, quality and efficiency improvements in diagnostics, in imaging, pathology, biochemistry and microbiology/virology.

Collaboration through the Scottish Molecular and Cytogenetics Laboratory Consortium brings together the regional genetics centres in Aberdeen, Dundee, Edinburgh and Glasgow to agree how best to provide genetic testing provision for rare diseases and to reduce unnecessary duplication.

NHS Scotland is an active participant in the UK Genetic Testing Network and the 4 genetic testing laboratories work as an integrated service with the 20 laboratories in England.

Work is underway among genetics specialists to establish a series of workshops to support the mainstreaming of genetic testing across relevant medical specialties to improve diagnosis and patient care.

[30] www.rarebestpractices.eu

Application of Genetic Testing

The Molecular Genetics Laboratory at the Southern General Hospital in Glasgow is the lead unit in Scotland for infantile epilepsy. Since 2005 it has tested for mutations in the SCN1A gene which causes infantile epilepsy. In this time the laboratory has maintained close links with the Yorkhill Hospital Paediatric Neurology Department and has as a result expanded the cohort of genes tested to 12. These genes account for some of the commonly associated infantile onset epilepsy syndromes. Almost 500 children a year are tested for genetic abnormalities.

Confirmation of a genetic epilepsy syndrome can have a positive influence on treatment options and clinical outcome for these children, as early diagnosis and subsequent targeted treatment can prevent co-morbidities by reducing seizure frequency.

Without genetic testing it can take several years to get a correct diagnosis. By this time developmental delay (due to prolonged and recurrent seizures) is severe and irreversible.

The laboratory receives specimens from the 4 Scottish Molecular Genetic Consortium centres as well as referrals from the rest of the UK and many international destinations. The testing service for infantile onset epilepsy syndromes has established the Molecular Genetics Laboratory as a centre of excellence for these conditions.

Scotland is represented on the Genetics Specialty Group[31], one of 23 national UK networks of topic-specific expertise, supported by the National Institute for Health Research. The Genetics Specialty Group links as appropriate to other specialty groups and to the NIHR Clinical Research Networks. The group seeks to drive a national approach to research into genetic-related issues covering:

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

Each National Managed Clinical Network also has a dedicated web site and links to the NHS Education Scotland (NES) Managed Knowledge Network where information about relevant conditions and clinical pathways and protocols is published. This may include assessment tools and decision making pathways as well as details of where specialist knowledge may be available locally, for example in paediatric allergy.

[31] www.crn.nihr.ac.uk/genetics/

Suggested Next Steps Emerging from Discussion/Engagement with Stakeholders

Diagnostic Pathways

We will improve diagnostic pathways for people with rare diseases through:

- Review by NHS Boards, NSD and Managed Diagnostic Networks of the input of diagnostic specialties to rare disease diagnosis and consideration of how delays may be reduced
- NSD action to increase awareness among clinicians in Scotland of criteria for patient referral to diagnostic pathways for Scottish or English specialist centres
- Obtaining the input of primary care to the development of appropriate pathways for diagnosis, treatment and support
- Exploration, through NHS Board e-Health leads and NSS Public Health and 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
- 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 diseases
- Exploration by NSS Public Health and Intelligence of how rare disease decision support systems and other rare disease software systems may feature in future development of IT systems in NHS Scotland, ensuring these are interoperable with GP clinical systems.

Training in Diagnostics

We will increase awareness of rare diseases among all relevant staff groups through:

- 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
- Consideration of the inclusion within post-graduate training of appropriate content on diagnostic skills relevant to rare diseases
- Supporting the development of higher specialist training in genetics for medical and scientific staff to develop sustainability in the genetic workforce
- Publicising Orphanet as appropriate to NHS Boards, staff groups, training organisations, universities and others across NHS Scotland
- Developing awareness raising of rare diseases among midwifery, health visiting and GP professions.

Databases

We will take review existing data collection approaches for rare diseases through:

- 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

- Mapping of existing nationally held data-sets to assess which have sufficiently detailed clinical coding to identify patients with rare diseases
- Exploration of opportunities to develop links between UK databases, Scotland specific databases and relevant international databases
- 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
- Consideration by NSS Public Health and 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.

7. CO-ORDINATION OF CARE

UK Strategy Commitments

23. 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:

- an appropriate care plan for all patients with a rare disease
- clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis
- the development of seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age
- access criteria and measures of quality and outcomes.

24. Agree that specialist clinical centres should as a minimum standard:

- have a sufficient caseload to build recognised expertise
- where possible, not depend on a single clinician
- coordinate care
- arrange for coordinated transition from children's to adults' services
- involve people with rare conditions, and their families and carers
- support research activity
- ensure their expertise is available to families and their healthcare teams.

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.

26. Set out clearly the connections to and communications with Specialist Clinical Centres in molecular diagnostics and other forms of diagnostic support.

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).

28. Work with international partners wherever possible and develop UK-wide criteria for centres to become part of an expert reference network to increase the flow of information between patients and professionals in a range of disciplines.

29. Improve systems to record genetic and other relevant information accurately to detail the incidence and prevalence of disease and to support service planning and international planning.

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

Where we are now

The route map to the 2020 vision asserts that there is *"…a strong consensus that to further improve the quality of care provided in Scotland there is a need for an expanded role for primary care and general practice in particular".* Some patients with rare diseases also need support from a range of specialists and hospital departments, making a multidisciplinary and co-ordinated team approach important.

Integration of adult health and social care is a key part of the Scottish Government's commitment to public service reform in Scotland. We will continue to drive forward the widely endorsed commitment to integrating health and social care services in Scotland. Preparatory work is under way with NHS Boards, Local Authorities, third and independent sector partners and including development and delivery against new Single Outcome Agreements and the building of effective Integrated Health and Social Care Partnerships.

The efficient co-ordination of care for people with rare diseases and sharing of relevant clinical information is also important in effectively targeting resources to improve support to patients and families and to improve associated clinical and social care outcomes.

Participation in Rare Disease Registers

Scotland participates in a number of rare disease related registers, including the British Paediatric Orphan Lung Disease Registry (Edinburgh); the European Huntington's Disease Network Registry; the Hereditary Haemorrhagic Telangiectasia Mutation Database (Edinburgh); the UK Cystic Fibrosis Database and the Glasgow Registry of Congenital Anomalies (part of BINOCAR (British Isles Network of Congenital Anomaly Registers) and EUROCAT (European Surveillance of Congenital Abnormalities) networks). An MS register aims to provide an accurate estimate of MS incidence and patient outcomes.

Where services for rare disease are nationally commissioned, each has a clear definition of its function and there are agreed patient pathways. When a patient needs treatment outside Scotland there are pathways through appropriate specialists and NSD to ensure access to highly specialist treatment.

Designated national specialist services in Scotland and the four regional Clinical Genetic Centres already adhere to the minimum standard[32] within the UK Strategy for Rare Diseases to be considered as Specialist Clinical Centres:

- have a sufficient caseload to build recognised expertise
- where possible, not depend on a single clinician
- co-ordinate care
- arrange for co-ordinated transition from children's to adults' services
- involve people with rare conditions, and their families and carers
- support research activity
- ensure their expertise is available to families and their healthcare teams.

[32] Commitment 24 of the Strategy - www.scotland.gov.uk/Resource/0043/00438631.pdf

The Scottish Clinical Genetics Forum brings together the Regional Clinical Genetic Centres and others as indicated from the Genetic Alliance UK (a UK alliance of support groups for individuals and families affected by genetic disorders) National Services Division, and when appropriate healthcare specialities and patient organisations.

There are examples of good practice involving integration of genetic testing and treatment – such as in the paediatric epilepsy service in Scotland, described earlier.

From May 2014, patients and clinicians will be given a greater say on what new medicines are approved by the Scottish Medicines Consortium (SMC) for use in the NHS for end of life care and for treating very rare conditions.

As part of this, the Scottish Medicines Consortium is making two big changes to their drug approval processes. Firstly they are establishing a new Patient and Clinician Engagement (PACE) meeting to give patient groups and clinicians a stronger voice in SMC drug approval decisions. Secondly, a new Peer Approved Clinical System (PACS) will replace the former Individual Patient Treatment Request process for consideration of medicines not approved for regular use in the NHS.

For medicines used to treat very rare conditions (those affecting around 100 people in Scotland) in addition to the patient and clinician engagement approach the SMC will consider more than just the direct health impact on the patient.

Under the new approaches, pharmaceutical companies will have an additional opportunity to put forward a Patient Access Scheme, to make their medicines more affordable.

Co-ordination of care can be more challenging in remote and rural settings. The Scottish Government's 2008 action plan – *Delivering for Remote and Rural Healthcare*[33] guided action to deliver high quality sustainable healthcare to people living in remote and rural communities. All 63 actions were delivered. Since then, continuing developments have been a matter for the North of Scotland Regional Planning Group on behalf of all NHS Boards. However, it has also been recognised that further work needs to be done. To that end, a further, new programme of work funded by Scottish Government and led by NHS Highland is testing models of rural service delivery in primary care.

This work, over three years, involves modernising service delivery approaches to meet patient and community needs and to provide access to the full range of primary care, community care, and acute and specialist care. Associated work is also on-going to recruit and retain the health and care professionals needed to support comprehensive services.

NHS Scotland is committed to moving the upper age limit for its children's hospital services from age 13th to age 16th, with some flexibility up to 18 years. The change has been implemented in many hospitals and will apply in the new children's hospitals in Edinburgh and Glasgow. The adjusted age limits will create an opportunity to ensure that the transition of care to adult services takes place at a suitable time.

[33] www.scotland.gov.uk/Publications/2008/05/06084423/6

Next Steps Emerging from Discussion/Engagement with Stakeholders

Pathways for Rare Diseases

We will improve knowledge and appropriate utilisation by referring clinicians of pathways for rare diseases through:

- Publication of patient pathways for rare disease and the referral requirements for services in and out of Scotland
- Working with specialist services to develop clear diagnostic pathways for molecular diagnostics.

Specialist Centres

We will provide comprehensive support to and clinical management of patients with individual rare diseases through:

- Ensuring Scottish and UK specialist centres bring together all appropriate investigation, diagnosis, treatment, support and research expertise for rare diseases
- Keeping under review the relevance to patients of any potential development of specialist services.

Strategy and Policy on Rare Diseases

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 developments of actions and agreement of timescales to progress support and treatment for rare diseases.

Patient Data and Database Development

We will support service planning, co-ordination of care and monitoring of service delivery through:

- 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
- 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 NHS Scotland clinicians with the opportunity to monitor certain diseases and to produce data of benefit to research and to service planning
- Sharing of anonymised information with partners across organisational and national boundaries in line with data protection requirements and taking particular care where conditions comprise small numbers to ensure individuals are not identified.

8. THE ROLE OF RESEARCH IN RARE DISEASES

UK Strategy Commitments

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

32. Work together to identify a selection of the rare diseases most suited to the development of best-care pathways and propose other rare diseases for possible pathway development, taking on board the needs of patients and carers and the challenges faced during delivery of the first set of pathways.

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 publically, 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.

43. Evaluate different NGS platform configurations, for example:
- NGS for clinical condition-specific sets of genes (such as 100–200 of the 22,000 genes)
- whole exome sequencing (2% of the entire genome)
- whole genome sequencing.

44. Support the introduction of NGS into mainstream NHS diagnostic pathways, underpinned by appropriate clinical bioinformatics, including clinical bioinformatics hubs supported by high performance computing centres, where appropriate.

45. Ensure that training and education are available to the NHS workforce, highlighting the importance of NGS to all aspects of rare disease care, including support for evidence based local counselling for patients and their relatives who receive NGS results.

46. Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration.

47. Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical.

48. Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders.

49. Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS.

50. Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases.

51. Improve engagement between key stakeholders, including:

a. patients and relatives
b. main funding providers
c. healthcare commissioners
d. NHS hospitals and specialist care units
e. industry (pharmaceutical, biotechnology, IT, diagnostics).

Where we are now

Scotland and the rest of the UK are leaders in research into rare diseases. By improving the link between research and services for patients we will promote a culture of innovation. This is expected to lead to faster access to evidence based care. The basic and translational research needed to achieve these benefits for society will also require international collaboration, particularly for extremely rare diseases.

Academic, health and commercial research bodies have an important role in developing research for rare diseases and in translating the research into treatment interventions. Partnership working provides opportunities to combine financial, intellectual and production resources to take research from concept, to the laboratory, to clinical trials, to production of patient treatments.

The Chief Scientist Office (CSO) research strategy for 2010-14 *'Investing in Research: Improving Health'*[34] emphasises the need to fund experimental and translational medicine research alongside health service and health population research. A new CSO research strategy is out for consultation from the beginning of July 2014 and will actively seek the views of the rare disease community.

An example of best practice in research is the Anne Rowling Regenerative Neurology Clinic which combines clinical and laboratory research approaches to investigate neurodegenerative diseases. Opened in October 2013, the clinic currently focusses on multiple sclerosis, motor neurone disease and related neurodegenerative conditions.

The Scottish Health Research (SHARE) Register

The SHARE Register has created a database of up to 1 million people in Scotland prepared to be approached directly about possible participation in research. Through linkages to health records, this facilitates the identification and recruitment of patients into clinical studies. This approach to register development has the potential to be extended to people with rare diseases.

An example of CSO-supported research is the 'Orcades' study looking at genetic factors in complex disorders in the population of Orkney. The Orcades research will provide a better understanding of the genetics of diseases, including MS and their stratification, potentially allowing the development of future targeted therapies. As Scotland has the distinction of having amongst the highest rates of MS in the world and as the rate is the highest in the Orkney Islands[35], this condition is of particular relevance. This approach provides a model for research in other diseases in which genetics feature.

Clinicians, academics, researchers and research institutions in Scotland are active in the field of research in rare diseases, in collaboration across the UK and for some conditions, with EU colleagues.

[34] www.cso.scot.nhs.uk/wp-content/uploads/2013/02/research.pdf
[35] J Neurol Neurosurg Psychiatry. 2012 Jul;83(7):719-24. doi:10.1136/jnnp-2011-301546. Epub 2012 May 10

Collaboration in Research

NHS Research Scotland (NRS) is a partnership involving NHS Boards and the CSO. The NRS Industry Partnership Forum provides a valuable opportunity to discuss with industry future collaborations, including research into rare diseases.

CSO participates in the UK Clinical Trials Gateway along with the other countries of the UK as part of its involvement in the National Institute for Health Research. It has worked with the Scottish Research Ethics Committees to put in place a system of proportionate review. This has been aligned and contributed to the work of the UK-wide compatibility group. CSO has also endorsed the HRA work in creating generic consent forms.

The NHS Research Scotland (NRS) Permissions Co-ordinating Centre based in NHS Grampian is a single, centralised point of contact for researchers and companies wishing to conduct multicentre clinical research in Scotland. It liaises directly with NHS Board Research and Development (R&D) offices to identify investigators and streamline the Scottish R&D permissions process.

CSO is reviewing aspects of the NHS Research Scotland infrastructure in order to ensure that Scotland remains a leader in attracting and delivering clinical research. Scotland is represented by CSO on UK-wide bodies including the Stratified Medicine Innovation Platform, the National Cancer Research Institute and the Medical Research Council UK Dementias Forum. These bodies have a remit which includes the co-ordination of strategic funding initiatives.

In the last year CSO has entered into collaboration with rare disease charities, agreeing for example, to co-fund a Clinical Research Fellowship in partnership with Action Duchenne and the Muscular Dystrophy Campaign.

In Scotland, Research Ethics Committee approval timescales are good and the time taken for Research and Development approval for non-commercial studies continue to fall. Further, NHS Boards have agreed to participate in a rare disease initiative under which certain types of rare disease studies will be expected through the approvals process.

Scotland's forthcoming reorganisation of national network support structures will consider how rare disease research can be better supported.

The development of a single costing model across NHS Scotland for both commercial and non-commercial studies has been implemented.

To ensure success clinical research needs patient participation. In rare disease research the smaller numbers of available patients require particular efforts by research teams to recruit enough research subjects.

To encourage patient participation in research, CSO has supported development of the information leaflets – *'Clinical trials: what they are and what they're not'* and *'Understanding Clinical Trials'*. These are designed to provide potential trial recruits with clear information on taking part in research.

Simplifying Research Approvals in Genetics

The National Institute for Health Research, UK Rare Genetic Disease Research Consortium Agreement (colloquially called the Musketeer's Memorandum[39]) is a national agreement signed up to by all host organisations for the regional genetics centres, including the four in Scotland: NHS Grampian; NHS Tayside; NHS Lothian and NHS Greater Glasgow and Clyde.

The agreement is applicable to non-CTIMP[38] rare disease projects which have minimal local hospital costs. This ensures that when a regional genetics centre, supported by their host organisation, develops a rare disease non-CTIMP project, the local approval is applied to equivalent research in all the other regional genetics centres, without any further administrative work.

The CSO supported Public Involvement Group comprises lay volunteers from a variety of backgrounds. Regular training provides the volunteers with information to support their role in providing a lay perspective to inform CSO policy and in their membership of the Experimental and Translational Medicine Research Committee, the Health Services and Population Health Research Committee, the Chief Scientist Committee, and other working groups. Volunteers provide assistance in ensuring communications on science make use of clear language understandable to the general public.

Stratified Medicines

The application of genetics techniques is increasing understanding of diseases and how individual patients respond to medications, based on their genetic profile. This knowledge has the potential to assist in tailoring drugs to the genetic makeup of individual patients to increase the efficacy of treatment and to reduce undesirable side-effects. Such 'personalised'/stratified medicines are currently in development. The first such medicines are now being prescribed in Scotland after appropriate genetic testing.

A Stratified Medicine Scotland Innovation Centre (SMS-IC) is to be built by 2015 within the new South Glasgow Hospitals Campus. This centre will bring together Scottish universities, NHS Scotland and industry partners to work collaboratively in the area of biomarkers and genomics. The focus on practical application should ensure that the outputs have relevance to the NHS.

[36] Trials that do not involve an Investigational Medicinal Product (IMP) as defined by the MHRA, and therefore do not fall within the scope of the Medicines for Human Use (Clinical Trials) Regulations 2004.
[37] www.bsgm.org.uk/genetics-healthcare-research/nihr-uk-rare-genetic-disease-research-consortium-agreement/

Genetics and Bio-bank

The Scottish Government supported initiative *'Generation Scotland'*[38] is developing a bio-bank of human biological samples which is available to academic researchers and to commercial companies for medical research.

This partnership between the Scottish University Medical Schools and NHS Scotland has accrued samples from 30,000 people from across Scotland. The majority of people providing these samples have consented for them to be linked to their medical records and have agreed to consider possible participation in future research.

There are a number of tissue banks outwith Generation Scotland which accept tissue donations. These are typically partially-funded from CSO, are both University and NHS based and have the potential to add value in rare disease research. In the medical teaching centres these tissue banks are integrated into the Pathology Departments. This provides a link between the Scottish Pathology Network[39] (SPAN), its network members and research and development in rare disease.

Scotland is also participating in the UK Bio-bank. Access to bio-bank resources is important in the study of factors influencing rare diseases.

[38] www.generationscotland.org/index.php?option=com_content&view=article&id=4&Itemid=8
[39] www.pathologyscotland.org/

Next Steps Emerging from Discussion/Engagement with Stakeholders

Research Dissemination

We will support rare disease research through the Chief Scientist Office and publicise to all stakeholders the work carried out in science and research to improve outcomes for people with rare diseases through:

- Continuation of the activity based funding scheme which rewards NHS Boards for recruiting patients into trials
- Continuation of the central management of excess treatment costs to facilitate prompt approval of rare disease studies
- Maintenance of the CSO's success in streamlining research governance through collaboration with the HRA (Health Research Authority) and others
- Maintenance of the current multicentre study mean approval times of 21 working days for commercial and 15 working days for non-commercial studies
- 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
- Continuation of the SHARE Register, maintaining its important role in supporting the participation in research of people from across the population
- Exploration of the development of online application processes and registers of interest for people with rare disease wishing to participate in relevant research
- Ensuring that rare disease research is fully supported in the reorganised clinical research infrastructure for NHS Scotland and that Scotland is represented on any proposed rare disease strategic research initiative.

Genetics

We will support the developing role of genetics in rare diseases through:

- Continuation (and evaluation as the technology develops) of the use of small next generation sequencing (NGS) devices in all 4 Scottish Genetics Laboratories and mainstreaming of NGS as appropriate into NHS diagnostic pathways
- 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
- Maintenance of the commitment of the four regional genetics centres to the UK Rare Genetic Disease Research Consortium Agreement.

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