**Table of commitments 2016-2018**

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| 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. | ENGLAND | In January 2017, NHS England published a 'Framework for patient and public participation in specialised commissioning'. This sets out how 163 patient and public members are involved in NHS England's governance structures. In particular, there are two patient and one voluntary sector/community representatives on each of the 42 Clinical Reference Groups.<br><br>In addition, NHS England involves patients on the working groups that develop 'products' such as clinical commissioning policies and service specifications. As well as involving patients through these mechanisms, NHS England has developed processes for both engaging patients and the public when developing commissioning products (through its registered stakeholder structure) and for public consultation. There are four patient and public voice members on the Rare Diseases Advisory Group. This is a UK-wide group that makes recommendations to NHS England and the three devolved administrations on the commissioning of highly specialised services. | On track |
| | | SCOTLAND | The Scottish Government is fully committed to empowering people in terms of their health and social care. A number of plans and policies have been made since the last biennial report that are being driven and delivered across Scotland. The ones that address this commitment are:<br><br>• The Rare Disease Implementation Oversight Group – a group of clinicians, geneticists, biochemists, patient, NHS and Scottish Government representatives who monitor the implementation of the Scottish plan.<br>• House of Care – provide a simple visual model of a house built around collaborative care planning conversations between individuals and their health care professionals. The approach has been implemented in 55 GP practices across Scotland.<br>• What Matters to You – aims to encourage and support meaningful conversations between people who provide health and social care and the people, families and carers who receive health and social care.<br>• Our Voice – provides information on rights and responsibilities for individuals, information on how to make complaints, self-directed support and advocacy services. It also contains information on the National Voice, which provides opportunities for people to join discussions about national health and social care policy. | On track |

A report on progress will be published by the Northern Ireland Department of Health in 2018.

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| | | | • National Specialist Services Committee / National Professional, Patient and Public Reference Group – this involved patients and clinicians providing expert advice via the NPPPRG to the NSSC on the commissioning of highly specialist services for patients in Scotland.<br>• National Network Management Service – commissions national clinical and diagnostic networks. This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions. | |
| | | WALES | The Health and Care standards for Wales put in place clear standards for health services in Wales. They provide the framework for how services are organised, managed and delivered on a day-to-day basis set around seven themes including safe, effective, timely and individual care. In Wales' recently updated Rare Diseases Implementation Plan, the Welsh Government and NHS Wales continue to commit to both empowering those with a rare disease and ensuring those affected by any kind of rare disease have timely access to high quality pathways of care.<br><br>This has been achieved by:<br>• The Welsh Rare Disease Implementation Group (RDIG) has a patient representative and an officer from the Genetic Alliance UK patient charity. The RDIG oversees the implementation process and works towards keeping the Welsh Delivery Plan updated and relevant. It liaises with the NHS and other interests, taking advantage of developments in Wales and elsewhere in the UK.<br>• Patient representatives are invited to be members of task and finish groups.<br>• Many Welsh health boards have established local rare diseases planning groups, with these involving patient representatives. | On track |
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| **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. | ENGLAND | NHS England is in the process of developing a rare disease insert. This will be a document that will sit alongside those NHS England service specifications for services that treat patients with rare diseases. The document will allow NHS England to hold providers to account for the way in which they treat patients with rare diseases. One of the three areas in the rare disease insert is care coordination. NHS England will expect providers to ensure that there is a person responsible for coordinating the care of any patient with a rare disease, coordination of care to include development and implementation of a care plan and liaison with other providers (and other authorities such as schools). | On track |
| | | SCOTLAND | The Scottish Government is fully committed to empowering people in terms of their health and social care. A number of plans and policies have been made | On track |

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| | | | since the last biennial report that are being driven and delivered across Scotland. The ones that address this commitment are:<br><br>• House of Care – see Commitment 1<br>• What Matters to You – see Commitment 1.<br>• Care Opinion – an online feedback services that enables people to give real-time feedback and engage in constructive dialogue with healthcare service providers about the services they, their families, people they care for have received.<br>• National Network Management Service – see Commitment 1.<br>• Provision of Communication Equipment & Support – this gives children and adults across all age ranges and care groups, who have lost their voice, or at risk of losing their voice or who have difficulty speaking a statutory right to access the communication equipment and support they need. | |
| | | WALES | In collaboration with the Wales Gene Park, Rare Diseases UK established the Welsh Rare Disease Patient Network to engage patients, families and patient organisations to ensure the patient voice is properly informed, and effectively represented in the discussion and development of the implementation of the UK Strategy for Rare Diseases.<br><br>On Rare Disease Day, 29 February 2016, the Rare Diseases Implementation Group held an event to heighten awareness of rare diseases, the Welsh implementation plan and the need for better co-ordination across Wales. Rare Disease UK also held an awareness event in the Senedd on 14 February 2017. | On track |
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| **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. | ENGLAND | NHS England is in the process of developing a rare disease insert. This will be a document that will sit alongside those NHS England service specifications for services that treat patients with rare diseases. The document will allow NHS England to hold providers to account for the way in which they treat patients with rare diseases. NHS England will expect providers to ensure that there is a person responsible for coordinating the care of any patient with a rare disease, coordination of care to include development and implementation of a care plan and liaison with other providers (and other authorities such as schools).<br>NHS England will also expect providers to give every patient with a rare disease an alert card (or similar document) that alerts other providers (and other authorities such as schools) to the patient's rare disease and any special considerations about their care, including medications. | On track |
| | | SCOTLAND | The Scottish Government is fully committed to empowering people in terms of | On track |

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| | | | their health and social care. A number of plans and policies have been made since the last biennial report that are being driven and delivered across Scotland. The ones that address this commitment are:<br><br>• Health & Social Care Delivery Plan – the Scottish Government's plans for enhancing health and social care services by 2021.<br>• Making it Easy: A Health Literacy Plan for Scotland – an action plan that seeks to "make Scotland a health literate society that enables all of us to live (and die) well on our own terms and with any health condition we may have".<br>• What Matters to You – see Commitment 1.<br>• Care Opinion – see Commitment 2<br>• National Network Management Service – see Commitment 1.<br>• Health & Social Care Standards - effective from April 2018. These will set out the standards that people should expect when using health & social care services. They aim to ensure people are fully involved in all decisions about their care and support. | |
| | | WALES | Many health boards have established rare diseases groups. For example, Aneurin Bevan University Health Board (ABUHB) has convened a local Rare Diseases Planning Group (RDPG), incorporating expertise from primary and secondary care, planning and research. This group will draw on local expertise to allow the health board to develop appropriate referral pathways, interface and transition services and individualised care plans for individuals with rare diseases and agree local pathways for these conditions.<br><br>The purpose of the group is to work with clinicians to further develop appropriate referral pathways, interface and transition services, and individualised care plans for patients with rare diseases with agreed local pathways for these conditions. | On track |
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| **4** | Make sure that patients and their families have a say in decisions about treatment and in the planning, evaluation and monitoring of services. | ENGLAND | The NHS England service specification proposition template includes a section on 'Outcomes and Applicable Quality Standards', which include: clinical outcomes; patient outcomes; and structure and process measures. The patient outcomes section can include, for example, indicators to assess how well the service / treatment is meeting the needs and / or aspirations of individual patients, for example shared decision making, patient outcome or experience measures identified via feedback from patient groups or representatives. | On track |
| | | SCOTLAND | The Scottish Government is fully committed to empowering people in terms of their health and social care. A number of plans and policies have been made since the last biennial report that are being driven and delivered across Scotland. | On track |

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| | | | The ones that address this commitment are:<br><br>• Health & Social Care Delivery Plan – see Commitment 3.<br>• Realistic Medicine – the CMO annual report which aims to tackle variation in care, managing clinical risk, reduce harm and waste and innovating to improve in order to provide a sustainable NHS. It puts the individual at the centre of their treatment.<br>• House of Care – see Commitment 1.<br>• What Matters to You – see Commitment 1.<br>• Care Opinion – see Commitment 2.<br>• Our Voice – see Commitment 1.<br>• National Network Management Service – see Commitment 1.<br>• Health & Social Care Standards – see Commitment 3.<br>• Provision of Communication Equipment & Support – see Commitment 2. | |
| | | WALES | The Welsh Government is committed to both empowering those with a rare disease and ensuring those affected by any kind of rare disease have timely access to high quality pathways of care.<br><br>Specific developments:<br>Betsi Cadwaladr University Health Board (BCUHB) supports situations where an individual needs support from different clinicians, possibly far apart or far from home, BCUHB works with WHSSC, as the specialised services commissioner, to develop and agree service specifications for specialised and highly specialised services. Where services are provided in England, WHSSC will work with NHS England to develop these. | On track |
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| **5** | Consider how to give all patients with a rare disease clear and timely information about: their condition and its development; treatment and therapy options; practical support. | ENGLAND | The NHS England service specification proposition template includes sections on: care pathways; evidence base; and patient outcomes. The patient outcomes section can include, for example, indicators to assess how well the service / treatment is meeting the needs and / or aspirations of individual patients, for example shared decision making, patient outcome or experience measures identified via feedback from patient groups or representatives.<br><br>The NHS England policy proposition template includes sections on: evidence base and patient pathway. Each policy proposition also includes a plain language summary of the policy. | On track |
| | | SCOTLAND | The Scottish Government is fully committed to empowering people in terms of their health and social care. A number of plans and policies have been made | On track |

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| | | | since the last biennial report that are being driven and delivered across Scotland. The ones that address this commitment are:<br><br>• Health & Social Care Delivery Plan – see Commitment 3.<br>• Realistic Medicine – see Commitment 4.<br>• Making it Easy: A Health Literacy Plan for Scotland – see Commitment 3.<br>• House of Care – see Commitment 1.<br>• What Matters to You – see Commitment 1.<br>• National Network Management Service – see Commitment 1.<br>• Health & Social Care Standards – see Commitment 3. | |
| | | WALES | Welsh Government expects anyone with a long term condition to have an appropriate individual care plan. In Betsi Cadwaladr University Health Board (BCUHB) their partnership work aims to ensure improvements across the whole 'patient journey', from the first contact with their GP through diagnosis to ongoing management of a rare condition. Patients and their family/carers are actively involved in joint partnership groups emphasising the commitment by BCUHB to the fundamental role that the patient, supported by their family/carer and/or patient organisation will play during this journey. | On track |
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| **6** | Improve access for patients (or where appropriate their parents or guardians) to their personal data. | ENGLAND | Not yet started - see Implementation Plan | n/a |
| | | SCOTLAND | Work ongoing in NHS Scotland around the National Patient Portal. The work is expected to be concluded by March 2018 in line with the existing aim of launching a portal with initial functionality by 2020. | On track |
| | | WALES | Informed health and care – A digital health and social care strategy for Wales sets out the Welsh Government's ambition to build on the progress we have already made and transform how the people of Wales, our citizens and staff, embrace modern information technology and digital tools. To deliver safer, more efficient and joined-up health and social care services to improve outcomes and experiences of patients and service users. Access to patient data is being considered through wider programmes within the NHS Wales Information Service, progress has been made developing integrated care records and My Health Online, certain specialities have also been trialling used of Patient Knows Best. | Progress being made |
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| **7** | Support patients to register on databases, where these exist. | ENGLAND | In October 2016 NCARDRS launched a web based system for electronic notifications. This operates over the N3 network and enables the treating clinician to report cases electronically. We also have in place a subject access request process available for patients. We recognise that we need to deliver further data transparency that meets the needs of patients whilst keeping their data as secure | Progress being made |

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| | | | as possible. We therefore have two current streams of work to further progress this work | |
| | | SCOTLAND | • National Network Management Service – commissions national clinical and diagnostic networks. This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions<br>• Nationally Managed Clinical Networks – following from the last biennial report, NSD has been supporting the development of pathways for a number of rare diseases (Acute Porphyria, Atypcial haemolytic-uremic syndrome (aHUS), Neurofibromatosis type 2, Vasculitis (including Behcets) and Ehlors Danlos Syndrome). This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions. | On track |
| | | WALES | For updates please see the Progress Report. | Progress being made |
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| **8** | Help patients to contribute to research and other activity related to rare diseases. | ENGLAND | NHS England is working to develop Rare Disease Research Networks (RDRNs) and the Rare Disease Research Centres that will be members of the Networks. The working definition of a RDRN is: a recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes. | On track |
| | | SCOTLAND | • National Network Management Service – commissions national clinical and diagnostic networks. This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions<br>• Nationally Managed Clinical Networks – following from the last biennial report, NSD has been supporting the development of pathways for a number of rare diseases (Acute Porphyria, Atypcial haemolytic-uremic syndrome (aHUS), Neurofibromatosis type 2, Vasculitis (including Behcets) and Ehlors Danlos Syndrome). This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions.<br>• SHARE is a new NHS research Scotland initiative created to establish a register for people interested in participating in health research who allow SHARE to use their data. SHARE aims to have 1 million people registered by 2023. The register has recruited over 180,000 participants (as of October 2017), making it the largest resource of it's kind in the UK.<br>• Scottish Genetics Speciality Group – supports the delivery and promotion of clinical research in a wide range of areas including rare disease. A number of studies have taken place including the Deciphering Development Disorders study. There is also participation in the Association for Improvements in the | On track |

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| | | | Maternity Services (AIMS) study for Marfan syndrome and the Cancer Prevention Programme (CaPP3) for Lynch syndrome. | |
| | | WALES | All Health and Care Research Wales activity is underpinned by the belief that the people of Wales have a key role to play in improving the quality and relevance of research. We want to create an environment in which all health and social care research that takes place in Wales happens with the public, for the public. A key priority in the Health and Care Research Wales Strategic Plan is the need to facilitate and enable wider public involvement, engagement and participation in health and social care research. Healthwise Wales was launched in February 2016, HealthWise Wales is an opportunity for everyone in Wales aged 16 or over to take part in research for better health, care and wellbeing**.** | Progress being made |
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| **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. | ENGLAND | On the advice of the UK NSC the following conditions are screened for as part of the NHS Newborn Blood Spot Screening Programme: phenylketonuria, congenital hypothyroidism, sickle cell disease, cystic fibrosis, medium-chain acyl Co-A dehydrogenase deficiency, maple syrup urine disease, homocystinuria, glutaric aciduria type 1 and isovaleric acidaemia. Newborn bloodspot screening in England is offered between 5 and 8 days after the baby is born.<br><br>The UK NSC has a robust evidence review process in place which ensures that each screening topic is addressed in a proportionate manner and to provide reassurance to stakeholders that decisions are grounded in, and informed by, up to date evidence. This includes a process to consider proposals for a new topic which has not been previously reviewed by the UK NSC.<br><br>The UK NSC carries out an annual call for new screening proposals and the Department would encourage the submission of future topics relating to the identification and prevention of rare diseases. Further information on how to submit a screening proposal to the UK NSC is available at https://www.gov.uk/government/publications/uk-nsc-evidence-review-process/appendix-d-how-to-submit-a-proposal-to-the-uk-nsc.<br><br>The UK NSC recently reviewed the evidence submitted for childhood Adrenoleukodystrophy (ALD) as part of the annual call for topics and undertook an external rapid review assessment. The review identifies that ALD is a serious health condition with devastating effects. However, there were significant gaps in the evidence-base around newborn screening for ALD and as a result a population screening programme could not be recommended. ALD will be included as part of the UK NSC's regular update cycle and will be reviewed again | On track |

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| | | | in three years. It is hoped that further information from the New York State pilot and other research will be available at that point.<br><br>DH and PHE are both actively engaging with the Nuffield Council on Bioethics on how important issues of ethics, equality and fairness in society are taken account of in the NHS antenatal screening programme. | |
| | | SCOTLAND | Introduction of screening tests for maple syrup urine disease, isovaleric acidaemia, gultartic aciduria type 2 and homocystinuria to help early detection commenced 20 March 2017. In the first three months, 20,748 babies have been tested, with 24 samples referred for either further testing or clinical referral<br><br>A new Scottish Screening Committee has been established to fully consider implementation of UKNSC recommendations in a specific Scottish context. The first meeting was held on 1 May 2016. | On track |
| | | WALES | Newborn Bloodspot screening service screens for rare but serious diseases that can cause serious illness or even death if not treated early. The screening test is carried out by a midwife and is part of routine post-natal care. It covers all of the conditions that are recommended by the UK National Screening Committee.<br><br>Public Health Wales has published its first Newborn Bloodspot Screening Wales annual report on 28 November 2017, the report shows that 99. 6 per cent of newborn babies in Wales were tested for a number of serious but rare medical conditions in 2016/17. Uptake for this programme is consistently over the target of 95%.<br><br>In the period covered by this report, 1 April 2016 to 31 March 2017, 33,505 babies were tested and the programme identified 39 serious conditions including:<br>• Congenital hypothyroidism<br>• Cystic fibrosis<br>• Medium-chain acyl-CoA dehydrogenase deficiency<br>• Phenylketonuria<br>• Sickle cell disorders | On track |
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| **10** | Initiate action to ensure carrier testing approved by the appropriate commissioning bodies, where the associated molecular | ENGLAND | NHS England is implementing a genomic testing strategy. | On track |
| | | SCOTLAND | A new Scottish Screening Committee has been established to fully consider implementation of UKNSC recommendations in a specific Scottish context. The first meeting was held on 1 May 2016. | On track |

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| | tests are evaluated and recommended by UKGTN, is accessible for at risk relatives. | | UKGTN are currently working to assess the clinical utility of carrier testing for patient groups. | |
| | | WALES | The Chair of the Welsh Rare Diseases Implementation Group also sits on the UKNSC screening board. New genetic and genomic technologies have the potential to revolutionise medicine and public health. Welsh Government's Genomics for precision medicine Strategy published in July 2017, sets out the Welsh Government's plan to create a sustainable, internationally competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales. | Progress being made |
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| **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 coordinated care e. g. to help disabled children who are thought to have a genetic syndrome or condition that science has not yet identified. | ENGLAND | NHS England worked with individual Royal Colleges to identify a list of those conditions that are difficult to diagnose but which have a profound effect if not diagnosed in a timely way. Although NHS England developed a list, this was not universally accepted by the Rare Diseases Advisory Group and further work is planned. This is likely to focus on identifying specific actions that can be taken to reduce delays in diagnosis, for example, identifying additional genetic tests for rare diseases that can be added to existing panel tests. This workstream will dovetail with the Diagnostic Odyssey Task & Finish Group (a sub-committee of the UK Rare Diseases Policy Board) of which NHS England is a member.<br><br>Whole genome sequencing (WGS) will be carried out clinically within the NHS from late 2018. WGS is offering an enhanced diagnostic yield of between 20 and 30%. This will enable more timely diagnosis of rare diseases. In addition the re procurement of the genomic laboratory infrastructure in the NHS will drive up quality and access to genomic testing and this should also have an impact on reducing diagnosis timescales. | Progress being made |
| | | SCOTLAND | • Health & Social Care Delivery Plan – see Commitment 3.<br>• National Network Management Service – see Commitment 1<br>• Decision support tools – SG is working with the Digital Health and Care Institute to create a delivery platform for quality assured decision support tools.<br>• National Demand Optimisation – NDO Group reviewed practices and information on the use of diagnostic tests across NHSS. A report was produced in 2017 that included guidance for NHSS Boards on optimising access to and provision of testing.<br>• Scottish Genomes Partnership – see Commitment 42 | On track |
| | | WALES | Lack of awareness and identification of rare diseases amongst healthcare professionals can often result in a delayed diagnosis or misdiagnosis of rare | On track |

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| | | | disease patients. Education in rare diseases is key.<br><br>The Rare Disease Implementation Group in Wales has set up a Task and Finish group looking at the investigation of children with delayed development. | |
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| **12** | Work with the NHS and clinicians to establish appropriate diagnostic pathways which are accessible to, and understood by, professionals and patients, by<br><br>— establishing clear, easily accessible and effective pathways between primary care,<br>secondary care, regional centres and specialist clinical centres, as appropriate<br><br>— putting protocols in place to identify patients with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment<br><br>— drawing on patients' ability to help inform decisions about referral and diagnosis<br><br>— creating effective clinical networks to support this process<br><br>— making high quality diagnostic tests accessible through common, clinically On trackd systems or | ENGLAND | The NHS England service specification proposition template includes sections on care pathways and patient outcomes. The patient outcomes section can include, for example, indicators to assess how well the service / treatment is meeting the needs and / or aspirations of individual patients, for example shared decision making, patient outcome or experience measures identified via feedback from patient groups or representatives.<br><br>The NHS England policy proposition template includes a section on the patient pathway. | On track |
| | | SCOTLAND | • Nationally Managed Clinical Networks – following from the last biennial report, NSD has been supporting the development of pathways for a number of rare diseases (Acute Porphyria, Atypcial haemolytic-uremic syndrome (aHUS), Neurofibromatosis type 2, Vasculitis (including Behcets) and Ehlors Danlos Syndrome). This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions.<br>• Huntington's disease – the SG provided funding for the Scottish Huntington's Association to develop a National Care Framework. The Framework was published in March 2017.<br>• Decision support tools – see above.<br>• National Demand Optimisation – see above.<br>• Data - the RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond.<br>• Cross Border Guidance – to improve knowledge of access, NSD has updated the Cross Border Guidance for Clinicians and will publish the NHSE Highly Specialist Services report to increase awareness of services available. | On track |
| | | WALES | The All Wales Medical Genetics Service (AWMGS), with support from the National Institute for Social Care and Health Research (now Health and Care Research Wales), is investigating whether clinical Exome sequencing, which allows many genes to be tested at once, can improve outcomes for patients with | On track |

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| | pathway<br><br>— embedding appropriate information in national data systems including measuring equity of access to molecular tests to maintain UKGTN diagnostic studies | | rare diseases, as well as providing a cost-benefit to the NHS. Patient members on the Exome sequencing study steering group has contributed to the study design and lay summary, and will guide the ongoing management of the project. This will include the production of a patient questionnaire examining the acceptability of genomic sequencing and issues related to incidental findings.<br><br>The Rare Disease Implementation Group in Wales has set up a Task and Finish group looking at the investigation of children with delayed development. | |
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| **13** | Ensure that there are appropriate procedures for evaluating the costs and benefits of treatments for patients | ENGLAND | NHS England has clear, published information about how it evaluates the costs and benefits of treatments for patients. | On track |
| | | SCOTLAND | The Cabinet Secretary announced on 14 December that the Scottish Government will take forward the recommendations of Dr Brian Montgomery's independent Review of Access to New Medicines. These reforms will help more patients get better access to the treatments they need. Work is underway in relation to data recording especially in relation to medicines for rare diseases. | On track |
| | | WALES | The All Wales Medicines Strategy Group (AWMSG) changed its process for appraising orphan, ultra-orphan medicines developed specifically for rare diseases to enable even greater involvement of patients and clinicians in Wales. In recognition of the clinical needs of patients with rare diseases, and acknowledging the potentially high costs of treatment, broader considerations are taken into account when appraising.<br><br>Health Technology Wales (HTW) has been established to deliver a strategic, national approach to the identification, appraisal and adoption of new health technologies into health and care settings across NHS Wales.<br><br>The All Wales Medical Genetics Service (AWMGS) provides clinical expertise, information and advice, as well as genetic testing services, for patients across Wales. Genetic testing is a major development area in diagnosing and treating rare diseases. Each year, as knowledge of genetic disorders increases, new genetic tests are commissioned by the UK Genetic Testing Network (UKGTN). These new genetic tests then become available to patients and their families affected by, or at risk of, the associated rare diseases. The right tests at the right time can improve efficiency and reduce anxiety by ensuring quicker diagnosis and avoiding ineffective treatment. | On track |
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| **14** | Where appropriate, support | ENGLAND | Not yet started - see Implementation Plan | n/a |

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| | the availability of computerised prompts to help GPs diagnose a rare disease when a rare disease has not previously been considered | SCOTLAND | Decision Support Tools - please see Commitment 11. | On track |
| | | WALES | One Red Flag tool for motor neurone disease has been developed by RCGP but they have decided not to develop further tools at this point. As a result, this approach has been considered but it was not felt appropriate at this point, consideration is being given to care pathway development and GP education instead. | n/a |
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| **15** | Improve education and awareness of rare diseases across the healthcare professions, including:<br><br>— involving patients in the development of training programmes<br><br>— encouraging medical, nursing and associated health professionals to get hands-on experience in specialist clinics<br><br>— ensuring awareness of methods and clinical techniques used in differential diagnosis | ENGLAND | • Patients are involved in teaching locally within hospital settings through the 13 HEE funded education and training leads. All HEE University contracts contain a requirement to involve patients in the development of programmes and in their delivery.<br>• The HEE Delphi study of GP training needs associated with rare diseases and genomics included patient participants. The RCGP Curricula explicitly mentions Rare Diseases and HEE has been invited to review/refresh the content.<br>• HEE's MOOC (Massive on line open learning course), which achieved more than 14,000 learner registrations in its first year, contains case studies of patients with rare diseases. Patients have also undertaken the MOOC.<br>• The HEE WeNurses webinar on Rare Disease Day 28/2/17 engaged over 6million people.<br>• HEE local teams responsible for medical education and training are facilitating medical trainees to gain experience within specialist clinics as part of the 100,000 Genomes project. By way of example in the East Midlands training is offered to the ENT consultants and teams for specific rare balance conditions where patients would benefit from NGS. | On track |
| | | SCOTLAND | Decision Support Tools - please see Commitment 11. | On track |
| | | WALES | Welsh health boards have developed local Rare Diseases Planning Groups, which incorporate expertise from primary and secondary care, planning and research. To allow the health boards to develop appropriate referral pathways, interface and transition services and individualised care plans for individuals with rare diseases and local pathways for these conditions. | On track |
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| **16** | Monitor the development of ICD-11 in preparation for its adoption | ENGLAND | The strategic intent of NHS England is to adopt SNOMED CT as its standard coding system. SNOMED CT is interoperable with ICD 10, and will be with ICD 11 when developed. | Progress being made |
| | | SCOTLAND | Decision Support Tools - please see Commitment 11. | On track |
| | | WALES | Awaiting progress in England. | n/a |
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| 17 | Work with colleagues in Europe on the development of the European Orphanet coding system and considering the adoption of Orphanet coding and nomenclature | ENGLAND | The strategic intent of NHS England is to adopt SNOMED CT as its standard coding system. SNOMED CT is interoperable with ICD 10, and will be with ICD 11 when developed. | Progress being made |
| | | SCOTLAND | Work is underway to consider the possibility of adding Orphanet to the quick reference section of the secondary care system - Trakcare.<br><br>Consideration of the use of Orphanet coding was considered in the RDIOG Short Life Working Group on data. The report notes that the codes are very useful but are very different from the ICD codes. Therefore, once the ICD-11 codes are known, further work will be carried out to determine if systems would be compatible. | On track |
| | | WALES | Within the Welsh Rare Diseases Implementation Plan is to work with WHSSC and colleagues in Europe on the further development of the European Orphanet. | Progress being made |
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| 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 | ENGLAND | NHS England continues to develop standard data flows to support the commissioning of specialised and highly specialised services. | On track |
| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond | On track |
| | | WALES | The Congenital Anomaly Register and Information Service (CARIS):<br>CARIS was established in 1998 with the objective of assessing patterns of anomalies in Wales, including possible clusters and their causes. Information from CARIS informs planning of wider health services, including screening services. | On track |
| | | | | |
| 19 | Explore options to improve the link between existing patient data and electronic health records | ENGLAND | NCARDRS has a national infrastructure in place with 8 regional offices covering the whole of England. As part of this, there is a single data management system in operation. Work to expand the collection of information on other rare diseases continues apace and a number of NHS trusts have shared information on their rare disease patients with NCARDRS. All new data flows are supported with data sharing. In order to maximise the level of case ascertainment, NCARDRS collects data from a large and diverse array of sources, which it then links to other sources of information (such as ONS and laboratory data) using a range of patient identifiers. | Progress being made |

| Number | UK Rare Disease Strategy Commitment | UK country | Progress update 2016 - 2018 | Progress |
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| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond | On track |
| | | WALES | Informed health and care – A digital health and social care strategy for Wales sets out the Welsh Government our ambition to build on the progress we have already made and transform how the people of Wales, our citizens and staff, embrace modern information technology and digital tools to deliver safer, more efficient and joined-up health and social care services to improve outcomes and experiences of patients and service users. Linkages of health records is being considered through wider programmes within the NHS Wales Information Service, progress has been made developing integrated individual healthcare records and establishing a Welsh Clinical Portal. | Progress being made |
| | | | | |
| **20** | Assess the potential for rare disease databases where they do not exist | ENGLAND | NCARDRS has a national infrastructure in place with 8 regional offices covering the whole of England. As part of this, there is a single data management system in operation. Work to expand the collection of information on other rare diseases continues apace and a number of NHS trusts have shared information on their rare disease patients with NCARDRS. All new data flows are supported with data sharing. In order to maximise the level of case ascertainment, NCARDRS collects data from a large and diverse array of sources, which it then links to other sources of information (such as ONS and laboratory data) using a range of patient identifiers. | Progress being made |
| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond | On track |
| | | WALES | The Congenital Anomaly Register and Information Service (CARIS):<br>CARIS was established in 1998 with the objective of assessing patterns of anomalies in Wales, including possible clusters and their causes. Information from CARIS informs planning of wider health services, including screening services. | On track |
| | | | | |

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| **21** | On track international standards, building on existing NHS standards | ENGLAND | The strategic intent of NHS England is to adopt SNOMED CT as its standard coding system. SNOMED CT is interoperable with ICD 10, and will be with ICD 11 when developed. | Progress being made |
| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond | On track |
| | | WALES | Within the Welsh Rare Diseases Implementation Plan is an action to ensure Wales is involved as a member of UK development of rare disease healthcare coding. | Progress being made |
| | | | | |
| **22** | Support international links to UK databases and build on the work of current funded programmes that aim to link rare disease research internationally | ENGLAND | NHS England will continue to embed the development of European Reference Networks. | On track |
| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond.<br><br>RARE Best Practice – Healthcare Improvement Scotland was a partner in the RARE-Best Practices project with responsibility for developing 2 databases – one of research gaps and one of appraised clinical guidelines. In order to maximise awareness and application of knowledge and information, a number of options are being considered by the RDIOG. | On track |
| | | WALES | Within the Welsh Rare Diseases Implementation Plan is an action to work with WHSSC and colleagues in Europe on the further development of the European Orphanet.<br><br>The Welsh Secure Anonymised Information Linkage (SAIL) databank is a anonymous data linkage system that securely brings together the widest possible array of routinely collected data for research, development and evaluation. The Wales Genomic Medicine Centre will work with SAIL to investigate linking genomic data to other health and social care data to improve the care of patients with rare diseases. | Progress being made |
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| **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:<br><br>— an appropriate care plan for all patients with a rare disease<br><br>— clearly stated principles around the standards of care which patients with a rare disease can expect, including patients with no diagnosis<br><br>— the development of seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age<br><br>— access criteria and measures of quality and outcomes | ENGLAND | The NHS England service specification proposition template includes sections on: care pathways, clinical outcomes, patient outcomes and applicable service standards.<br><br>The NHS England policy proposition template also includes a section on the patient pathway.<br><br>NHS England is in the process of developing a rare disease insert. This will be a document that will sit alongside those NHS England service specifications for services that treat patients with rare diseases. The document will allow NHS England to hold providers to account for the way in which they treat patients with rare diseases. NHS England will expect providers to ensure that there is a person responsible for coordinating the care of any patient with a rare disease, coordination of care to include development and implementation of a care plan and liaison with other providers (and other authorities such as schools). NHS England will also expect providers to ensure that every paediatric patient with a rare disease has an active transition to an appropriate adult service. | On track |
| | | SCOTLAND | • Nationally Managed Clinical Networks – following from the last biennial report, NSD has been supporting the development of pathways for a number of rare diseases (Acute Porphyria, Atypcial haemolytic-uremic syndrome (aHUS), Neurofibromatosis type 2, Vasculitis (including Behcets) and Ehlors Danlos Syndrome). This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions.<br>• Huntington's disease – the SG provided funding for the Scottish Huntington's Association to develop a National Care Framework. The Framework was published in March 2017.<br>• Inherited Metabolic Disease – NSD and the National Specialist Services Committee consider applications for new specialist services and networks. Over 2016, a review of the IMD National Managed Clinical Network highlighted the need to assess the sustainability of the current service provision of services in Scotland. The expert review group recommended the national designation of an integrated service for Scotland. The NSSC will consider the application in December 2017. | On track |
| | | WALES | The updated Welsh Rare Diseases Implementation Plan builds on the first phase and gives the NHS and its partners the vital continuity of approach it needs. The Plan is set out in a similar structure, covering the need to empower, diagnose early, and provide fast, effective and safe care, treatment, and research. | On track |

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| | | | The Rare Disease Implementation Group in Wales has set up a Task and Finish group looking at the investigation of children with delayed development.<br><br>Specific developments in Wales are Aneurin Bevan University Health Board (ABUHB) convening a local Rare Diseases Planning Group (RDPG), incorporating expertise from primary and secondary care, planning and research. This group draws on local expertise to allow the health board to develop appropriate referral pathways, interface and transition services and individualised care plans for individuals with rare diseases and agree local pathways for these conditions. | |
| | | | | |
| **24** | Agree that specialist clinical centres should as a minimum standard:<br>— have a sufficient caseload to build recognised expertise<br>— where possible, not depend on a single clinician<br>— coordinate care<br>— arrange for coordinated transition from children's to adults' services<br>— involve people with rare conditions, and their families and carers<br>— support research activity<br>— ensure their expertise is available to families and their healthcare teams | ENGLAND | NHS England is in the process of developing a rare disease insert. This will be a document that will sit alongside those NHS England service specifications for services that treat patients with rare diseases. The document will allow NHS England to hold providers to account for the way in which they treat patients with rare diseases. NHS England will expect providers to ensure that there is a person responsible for coordinating the care of any patient with a rare disease, coordination of care to include development and implementation of a care plan and liaison with other providers (and other authorities such as schools). NHS England will also expect providers to ensure that every paediatric patient with a rare disease has an active transition to an appropriate adult service.<br><br>NHS England is developing an aspirant market entrance process that will give providers the opportunity to demonstrate a case for there being additional/replacement providers of highly specialised services, for example, because there is inequitable geographical access to a service. | On track |
| | | SCOTLAND | NSD works with NHS Boards and other highly specialised service commissioners across the UK to ensure that patients have access to appropriate treatment across the UK. All specialist centres have a detailed specification which outline the expected service to be delivered and are regularly reviewed. New services would be expected to adopt the same principles. All national specialist services in Scotland are involved in research. | Complete |
| | | WALES | Welsh Health Specialised Services Committee (WHSSC) commission services for people with rare disease and working with providers within NHS Wales and in England to ensure service meet the required standards. Additional funding was secured in the 2016-17 financial year from Welsh Health Specialised Services Committee (WHSSC) for very rare genetic tests that are not available in Wales but available via the UK Genetic Testing Network (UKGTN) The Laboratory has a commitment from WHSSC that there will be further funding as more tests become available in the future. | Progress being made |

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| **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 | ENGLAND | Not yet started - see Implementation Plan | n/a |
| | | SCOTLAND | NSD works with NHS Boards and other highly specialised service commissioners across the UK to ensure that patients have access to appropriate treatment across the UK. All specialist centres have a detailed specification which outline the expected service to be delivered and are regularly reviewed. New services would be expected to adopt the same principles. All national specialist services in Scotland are involved in research.<br><br>• Making it Easy: Health Literacy | On track |
| | | WALES | The relationships between health care and research are set out in the Health and Care research strategic plan and in the updated Welsh Rare Diseases Implementation Plan. | On track |
| | | | | |
| **26** | Set out clearly the connections to and communications with specialist clinical centres in molecular diagnostics and other forms of diagnostic support | ENGLAND | In 2017 NHS England will re procure the genomic infrastructure across the NHS. This will ensure that molecular/genomic testing for rare diseases, and other conditions of a genetic nature, is carried out in a timely manner in laboratories with high levels of expertise and throughput. | On track |
| | | SCOTLAND | NSD undertook a review of the Nationally Designated Genetics Laboratory Testing Services in 2016. Several recommendations were made:<br><br>• The Genetics Laboratory Consortium should work towards the introduction of Whole Exome Sequencing within the next 5 years, as well as the development of multi gene panels in the shorter term. It is anticipated that as new testing techniques are developed, there will be a reduction in demand for traditional testing.<br>• The evaluation of clinical usefulness and cost effectiveness of molecular pathology tests should continue to be undertaken by the Scottish Molecular Pathology Evaluation Panel (MPEP). MPEP recommendations would be made to the Scottish Molecular Pathology Consortium Steering Group for consideration of whether tests should be commissioned routinely by NHSS.<br>• The Genetics Evaluation Panel is being reworked into a similar panel as MPEP. The GEP will assess proposal tests based on clinical need and recommend which tests should be developed to UKGTN standard. It will also advise the Genetics Laboratory Consortium Steering Group on the clinical utility of tests. | On track |
| | | WALES | The all Wales genetic laboratory is a member of UKGTN and communicates with other member laboratories and referring clinical centres on the services it provides and receives. In July 2017 Welsh Government published, Genomics for precision | On track |

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| | | | medicine strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales. | |
| | | | | |
| **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) | ENGLAND | NHS England is working to develop Rare Disease Research Networks (RDRNs) and the Rare Disease Research Centres that will be members of the Networks. The working definition of a RDRN is: a recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes.<br><br>NHS England will also continue to embed the development of European Reference Networks. | On track |
| | | SCOTLAND | NSD works with NHS Boards and other highly specialised service commissioners across the UK to ensure that patients have access to appropriate treatment across the UK. All specialist centres have a detailed specification which outline the expected service to be delivered and are regularly reviewed. New services would be expected to adopt the same principles. All national specialist services in Scotland are involved in research. | Complete |
| | | WALES | The Welsh Government's commitment, for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales, is set out in the Health and Care Research Strategic plan. In addition, the updated Welsh Rare Diseases Implementation Plan sets out the Welsh Government's commitment to research for people with rare diseases and sets out a number of actions for health boards. | On track |
| | | | | |
| **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 | ENGLAND | NHS England will continue to embed the development of European Reference Networks. | On track |
| | | SCOTLAND | Seven European Reference Networks have been established in Scotland. | On track |
| | | WALES | The European Reference Network on Rare and Undiagnosed Skin Disorders is based at Cardiff and Vale University Health Board. | On track |
| | | | | |
| **29** | Improve systems to record genetic and other relevant information accurately to | ENGLAND | NCARDRS has a national infrastructure in place with 8 regional offices covering the whole of England. As part of this, there is a single data management system in operation. Work to expand the collection of information on other rare diseases | Progress being made |

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| | record the incidence and prevalence of disease and support service planning and international planning | | continues apace and a number of NHS trusts have shared information on their rare disease patients with NCARDRS. All new data flows are supported with data sharing. In order to maximise the level of case ascertainment, NCARDRS collects data from a large and diverse array of sources, which it then links to other sources of information (such as ONS and laboratory data) using a range of patient identifiers. | |
| | | SCOTLAND | • The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond.<br><br>• RARE Best Practice – Healthcare Improvement Scotland was a partner in the RARE-Best Practices project with responsibility for developing 2 databases – one of research gaps and one of appraised clinical guidelines. In order to maximise awareness and application of knowledge and information, a number of options are being considered by the RDIOG.<br><br>• The Scottish Genomes Partnership | On track |
| | | WALES | The Congenital Anomaly Register and Information Service (CARIS):<br>CARIS was established in 1998 with the objective of assessing patterns of anomalies in Wales, including possible clusters and their causes. Information from CARIS informs planning of wider health services, including screening services. | On track |
| | | | | |
| **30** | Identify how they can change systems to hold information about rare diseases, including information about the uptake of treatments | ENGLAND | The strategic intent of NHS England is to adopt SNOMED CT as its standard coding system. SNOMED CT is interoperable with ICD 10, and will be with ICD 11 when developed. | Progress being made |
| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond.<br><br>Consideration of the use of Orphanet coding was considered in the RDIOG Short Life Working Group on data. The report notes that the codes are very useful but | On track |

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| | | | are very different from the ICD codes. Therefore, once the ICD-11 codes are known, further work will be carried out to determine how best to capture data on rare diseases<br><br>Many NSD services or services with links to already submit data to UK-wide registries. For instance the Scottish Haemophilia centres enter data onto the UKHCDO database. Nationally commissioned stem cell services submit data to the BSBMT database. However lack of data management resource means that not all clinicians who look after people with rare diseases have the capacity to contribute to these databases and/or query them. This particularly applies to areas without a national service or NMCN. | |
| | | WALES | Data is being submitted to registries where these exist, given the complex nature and number of conditions a Welsh specific registry would not be achievable. If a UK wide or European register was developed consideration would be given to Wales participating.<br>The Welsh Secure Anonymised Information Linkage (SAIL) databank is a anonymous data linkage system that securely brings together the widest possible array of routinely collected data for research, development and evaluation. The Wales Genomic Medicine Centre will work with SAIL to investigate linking genomic data to other health and social care data to improve the care of patients with rare diseases. | Progress being made |
| | | | | |
| **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 | ENGLAND | DH is working with the UK Rare Diseases Policy Board and devolved administrations to facilitate UK-wide collaboration and learning across the four Nations. | Progress being made |
| | | SCOTLAND | The RDIOG established a Short Life Working Group to scope existing data and systems. The draft paper was considered by the RDIOG in December 2016. Work has started on implementing the recommendations which mainly focusses on the creation of a Congenital Anomalies Register. A full business case will be written towards the end of 2017, beginning of 2018 with a view to securing funding for 2018/19 and beyond.<br><br>Consideration of the use of Orphanet coding was considered in the RDIOG Short Life Working Group on data. The report notes that the codes are very useful but are very different from the ICD codes. Therefore, once the ICD-11 codes are known, further work will be carried out to determine how best to capture data on rare diseases.<br><br>Many NSD services or services with links to already submit data to UK-wide | Progress being made |

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| | | | registries. For instance the Scottish Haemophilia centres enter data onto the UKHCDO database. Nationally commissioned stem cell services submit data to the BSBMT database. However lack of data management resource means that not all clinicians who look after people with rare diseases have the capacity to contribute to these databases and/or query them. This particularly applies to areas without a national service or NMCN. | |
| | | WALES | Informed health and care – A digital health and social care strategy for Wales set out the Welsh Government's ambition to build on the progress we have already made and transform how the people of Wales, our citizens and staff, embrace modern information technology and digital tools to deliver safer, more efficient and joined-up health and social care services to improve outcomes and experiences of patients and service users. Linkages of health records is being considered through wider programmes within the NHS Wales Information Service, progress has been made developing integrated individual healthcare records and establishing a Welsh Clinical Portal. The Welsh Secure Anonymised Information Linkage (SAIL) databank is a anonymous data linkage system that securely brings together the widest possible array of routinely collected data for research, development and evaluation. The Wales Genomic Medicine Centre will work with SAIL to investigate linking genomic data to other health and social care data to improve the care of patients with rare diseases. | Progress being made |
| | | | | |
| **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 | ENGLAND | NHS England is developing a number of services for patients with rare diseases. The NHS England service specification proposition template includes a section on patient pathways.<br><br>All service specifications for highly specialised services are considered at the Rare Diseases Advisory Group, which has membership from the four nations. | On track |
| | | SCOTLAND | • Nationally Managed Clinical Networks – following from the last biennial report, NSD has been supporting the development of pathways for a number of rare diseases (Acute Porphyria, Atypcial haemolytic-uremic syndrome (aHUS), Neurofibromatosis type 2, Vasculitis (including Behcets) and Ehlors Danlos Syndrome). This work involves patients and clinicians mapping existing services, highlighting gaps and facilitating solutions.<br><br>• Huntington's disease – the SG provided funding for the Scottish Huntington's Association to develop a National Care Framework. The Framework was published in March 2017. | On track |
| | | WALES | The Welsh Rare Disease Implementation group are working with health boards, | Progress |

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| | | | Wales genetics service and the Congenital Anomaly Register and Information Service (CARIS) to understand numbers of referrals for and incidence of rare diseases. Consideration will be given to as to whether there is a need to develop specific pathways for some conditions. Work is ongoing to develop a pathway for investigation of children with delayed development. . | being made |
| | | | | |
| **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 | ENGLAND | NHS England is working to develop Rare Disease Research Networks (RDRNs) and the Rare Disease Research Centres that will be members of the Networks. The working definition of a RDRN is: a recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes.<br><br>NHS England has a published position on excess treatment costs. | On track |
| | | SCOTLAND | The Research Active Scottish NHS Boards have signed up to the Musketeer's memorandum thereby facilitating approval of multi-centre rare disease clinical studies.<br><br>The Chief Scientist Office has continued its policy of centrally managing excess treatment costs to facilitate prompt approval of rare disease studies and activity based funding of NHS Boards.<br><br>Within the revised NHS Research Scotland research support infrastructure, a Rare Disease Specialty Group has been established to develop and deliver rare disease research studies in Scotland. | On track |
| | | WALES | The Welsh Government's commitment, for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales, is set out in the Health and Care Research Strategic plan. In addition, the updated Welsh Rare Diseases Implementation Plan sets out the Welsh Government's commitment to research for people with rare diseases and sets out a number of actions for health boards. Welsh involvement in 100,000 genomes project will enable rare disease patients to take part in the study. | On track |
| | | | | |
| **34** | Make better use of online applications to give patients information about their condition so that they can develop a personalised care | ENGLAND | Not yet started - see Implementation Plan | n/a |
| | | SCOTLAND | Work ongoing in NHS Scotland around the National Patient Portal. The work is expected to be concluded by March 2018 in line with the existing aim of launching a portal with initial functionality by 2020.<br>Decision support tools – SG is working with the Digital Health and Care Institute to | On track |

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| | path plan with their clinical and social care team | | create a delivery platform for quality assured decision support tools. | |
| | | WALES | NHS Wales Informatics Service (NWIS) are currently delivering a project called NHS Wales/GIG Cymru portal and Interactive digital service project, which will better provide information to patients and health professionals. | On track |
| | | | | |
| **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 | ENGLAND | As of October 2017 responsibility for the UKCTG has been transferred to the NIHR Clinical Research Network Coordinating Centre (NIHR CRNCC). This is intended to utilise maximum benefit of the synergies between the UKCTG and the Join Dementia Research System also operated by the NIHR CRNCC. The CRNCC will be introducing a series of developmental updates to the system in order to improve the usability and flexibility of the Gateway to both volunteers and researchers. As part of this work will be plans on how to engage with the over 10,000 volunteers who tested the registration functionality of the gateway and to consider how to enable researchers to make direct contact with volunteers. Work has also commenced with the Health Research Authority on enabling the Gateway to be recognised as an appropriate recruitment source for future studies.<br><br>NIHR has, with INVOLVE, actively promoted and led on activities to encourage greater patient and public involvement, engagement and participation in research. This includes the 'I Am Research' campaign launched in 2017. In addition, all NIHR supported research projects, programmes and infrastructure are required to include active patient/ public engagement and involvement in their design and conduct. | On track |
| | | SCOTLAND | SHARE is a new NHS research Scotland initiative created to establish a register for people interested in participating in health research who allow SHARE to use their data. SHARE aims to have 1 million people registered by 2023. The register has recruited over 180,000 participants (as of October 2017), making it the largest resource of its kind in the UK. | Complete |
| | | WALES | Within the Welsh Rare Diseases implementation Plan is actions for health boards to:<br><br>• Work with the Health and Care Research Wales (HCRW) genetics speciality lead, researchers and the HCRW Support and Delivery service to increase the number of rare disease research studies undertaken in Wales;<br><br>• Encourage more people with rare diseases in Wales to participate in research activity, for example through working with the Wales Genomic Medicine Centre to enable patients across Wales to take part in the 100,000 genomes | Progress being made |

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| | | | project;<br><br>• Health and Care Research Wales activity is underpinned by the belief that the people of Wales have a key role to play in improving the quality and relevance of research. We want to create an environment in which all health and social care research that takes place in Wales happens with the public, for the public. A key priority in the Health and Care Research Wales Strategic Plan is the need to facilitate and enable wider public involvement, engagement and participation in health and social care research. Healthwise Wales was launched in February 2016, HealthWise Wales is an opportunity for everyone in Wales aged 16 or over to take part in research for better health, care and wellbeing.<br><br>• The Welsh Secure Anonymised Information Linkage (SAIL) databank is an anonymous data linkage system that securely brings together the widest possible array of routinely collected data for research, development and evaluation. The Wales Genomic Medicine Centre will work with SAIL to investigate linking genomic data to other health and social care data to improve the care of patients with rare diseases | |
| | | | | |
| **36** | Encourage patient groups to get involved with regulatory bodies | ENGLAND | The HRA manages the public involvement network which is a virtual network of patients and members of the public (82 public contributors are currently on our network). When opportunities for the public to become involved or contribute their views/insights to our work arise, we advertise opportunities to this network (and/or advertise to connected external patient networks, depending on the opportunity). HRA public contributors have been involved in the recruitment process to appoint a new HRA Director of Policy, co-designed/delivered an induction session for new staff and contributed to a public dialogue workshop set up by our Confidentiality Advice Team.<br><br>HRA has also established a working relationship with NIHR INVOLVE including its work on increasing the diversity of people involved in research.<br><br>HRA has participated in the Arm's Length Strategy Forum on Public Involvement, set up in 2016. One of the aims of this Forum is to increase the diversity of people involved in healthcare service development and research and to influence researchers and research funding organizations to encourage them to take the same approach. | On track |

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| | | | MHRA has run a Patient Group Consultative Forum (PGCF) that provides a framework for patient groups to get involved with the regulator and acts as an agency-wide resource that can be used when the "patient voice" is required to add value to the decision-making process. | |
| | | SCOTLAND | Healthcare Improvement Scotland is the regulator for healthcare in Scotland. Consideration will be given to this commitment during the next phase of implementation. | To be started |
| | | WALES | The Rare Diseases Implementation Group works with the charity Rare Disease UK, patient and Genetic Alliance representatives sit on the Group. Healthcare Inspectorate Wales is the independent inspectorate and regular for Wales they utilise a number of approaches to help them understand people's experiences of healthcare in Wales. | On track |
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| **37** | Help patient organisations and community engagement events develop more formal partnerships with the NHS research-active organisations | ENGLAND | In 2017, the 'I am research' campaign (https://www.nihr.ac.uk/news-and-events/support-our-campaigns/i-am-research/ ) was launched and follows in the footsteps of the 'OK to ask' campaign helping to raise awareness of research. Campaign highlights include:<br><br>• 186 events held in NHS trusts, GP surgeries and community settings across England<br>• 20 pieces of radio coverage, with a 34 million audience reach<br>• 46,220 general leaflets ordered<br>• 7,321 campaign page views<br>• Eight blogs<br>• Three #whywedoresearch tweetchats<br>• Over 1. 5 million social reach for our first ever Thunderclap<br>• 5,032 engagements and 26,921 reach on Facebook<br>• 649 new @OfficialNIHR Twitter followers<br>• 12,997 #IAmResearch tweets (data from Symplor)<br>• 1,015 views for our first ever Facebook Live broadcast | Progress being made |
| | | SCOTLAND | SHARE is a new NHS research Scotland initiative created to establish a register for people interested in participating in health research who allow SHARE to use their data. SHARE aims to have 1 million people registered by 2023. The register has recruited over 180,000 participants (as of October 2017); making it the largest resource of its kind in the UK. | Complete |
| | | WALES | Wales Gene Park plays an active role in community and public engagement events, working with Genetic Alliance UK, and linking with the All Wales Medical Genetics Service. | On track |

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| **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 | ENGLAND | As of October 2017 responsibility for the UKCTG has been transferred to the NIHR Clinical Research Network Coordinating Centre (NIHR CRNCC). This is intended to utilise maximum benefit of the synergies between the UKCTG and the Join Dementia Research System also operated by the NIHR CRNCC.<br><br>The CRNCC will be introducing a series of developmental updates to the system in order to improve the usability and flexibility of the Gateway to both volunteers and researchers. As part of this work will be plans on how to engage with the over 10,000 volunteers who tested the registration functionality of the gateway and to consider how to enable researchers to make direct contact with volunteers. Work has also commenced with the Health Research Authority on enabling the Gateway to be recognised as an appropriate recruitment source for future studies. | Progress being made |
| | | SCOTLAND | CSO has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK. | Progress being made |
| | | WALES | Health and Care research Wales has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK. | |
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| **39** | Work with the research community, regulators, providers of NHS services and research funders to develop risk-proportional permission systems | ENGLAND | HRA Approval provides sites with the information required to assess, arrange and confirm capacity and capability. In some instances there is no requirement to undertake these checks as there is no impact to service. We work closely with sponsor and host organisations to embed this proportionate approach. | On track |
| | | SCOTLAND | CSO has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK. | On track |
| | | WALES | Ongoing work in Welsh Government with the other UK nations and through the Health and Care Research Wales Permissions Service, which helps researchers to obtain NHS permission in Wales. The Service coordinates a streamlined and consistent process on behalf of NHS Wales. | On track |
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| **40** | Encourage researchers to use current guidance to produce generic participant | ENGLAND | HRA guidance is continually reviewed and revised as necessary. The guidance was updated to incorporate more emphasis on proportionate approaches after stakeholder engagement. Guidance relating to submission has been migrated to | On track |

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| | information leaflets and consent forms and participate in future guidance reviews | | Integrated Research Application System. We are currently, with stakeholder input, updating our website and its content to be clearer and more user friendly. We are working with HTA engaging with the public to explore the public's understanding and wishes around consent. We are also working with MHRA on a statement on the use of e-consent. | |
| | | SCOTLAND | CSO has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK. | On track |
| | | WALES | Health and Care research Wales has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK. | Progress being made |
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| **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. | ENGLAND | HRA fully rolled out its new HRA Approval process in March 2016. The new system aims to reduce unnecessary bureaucracy and duplication by incorporating assessments by NHS staff alongside the independent research ethics committee opinion, resulting in a single HRA Approval assessing all practical, legal and ethical aspects of health research studies in the NHS in England. It provides an opportunity for tailored instructions to be given to NHS organisations to address the specific considerations of a study, including handling arrangements for identification and/or referral of patients with rare diseases.<br><br>The HRA continues to engage with NHS R&D and sponsors to support consistent implementation of processes that are streamlined. HRA are also working with sponsors, hosts, funders and regulators to identify any duplication and the best ways to reduce this. | On track |
| | | SCOTLAND | CSO has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK. | On track |
| | | WALES | Health and Care research Wales has been fully involved in the establishment of the HRA procedures and has been working with colleagues across the 4 devolved nations to ensure seamless cross border working. This is of particular relevance to rare disease studies which may need to recruit patients from across the UK Ongoing work through the Health and Care Research Wales Permissions Service, which helps researchers to obtain NHS permission in Wales. The Service coordinates a streamlined and consistent process on behalf of NHS Wales. | On track |

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| **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 | ENGLAND | Through the 100 000 genomes project, the NHS has collected more than 40,000 samples from patients with rare diseases and their relatives. The sequencing of these samples is leading to new discovery and treatment and reducing diagnostic odyssey. Current indications are that there will be a 20 - 30% improvement in diagnostic yield over current testing - clearly demonstrating the value of whole genome sequencing (WGS) for diagnostic purposes in rare diseases. As part of the transition from this discovery stage into the use of WGS as part of mainstream testing within the NHS, NHS England is developing a genomic testing strategy which will be based upon solid clinical evidence and health economic assessments. | Complete |
| | | SCOTLAND | The Scottish Genomes Partnership (SGP), established in January 2015 is a collaboration of Scottish Universities and the NHS Scotland genetics service. The initiative will develop genomic medicine, research and commercial opportunities in Scotland and a Scottish partnership with Genomics England Ltd on genomic sequencing of NHSS rare disease patients.<br><br>In March 2017, the SGP Rare Disease collaboration with Genomics England 100,000 Genomes Project opened for recruitment. One thousand participants will be recruited through the nationally designated NHSS genetic clinics in Aberdeen, Dundee, Edinburgh and Glasgow by the end of March 2018. The first set of reports from Genomics England are expected back around September 2017, reporting back to participants by the end of December 2018.<br><br>A Scientific Advisory Board has been established and members have noted the considerable amount of work that took place between January 2016 to February 2017 to align protocols with the Genomics England approach, and in gaining regulatory approval from the Public Benefit and Privacy Panel for Health & Social Care and NHS Research Scotland. The board also made several recommendations for going forward which SGP are acting on.<br><br>NHSS recognises this is a fast moving area and that staff will need to be trained in this new area of work. As a result, local delivery plans for NHS Education Scotland (NES) include a commitment to provide support for provision of genetic workforce development programmes.<br><br>Next Generation Sequencing – while SGP will develop the methodology for Whole Genome Sequencing, Scottish laboratories will continue to develop NGS for use as a diagnostic tool for genetics. | On track |

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| | | WALES | The All Wales Genetic Laboratory has introduced genomic analysis using Next Generation Sequencing (NGS), and has seen a dramatic increase in the number of patient samples analysed each year. As a result, more patients with rare diseases are able to access genomic sequencing results, and therefore receive a diagnosis, appropriate treatment and management.<br><br>In September 2016, Genomic Medicine Centre was awarded Medical Research Council (MRC) and Welsh Government funding of £1m and £2. 4m respectively to support Wales' involvement in the Genomics England 100,000 genomes project. The Genomic Medicine Centre will work closely with the Wales Gene Park, All Wales Medical Genetics Service and heath boards and trusts across Wales to facilitate the development of genomic medicine in Wales. The 100,000 Genomes Project in Wales will be used as an exemplar towards the integration of genomic medicine into clinical care pathways in Wales and aligns with the Welsh Government Genomics for Precision Medicine Strategy. The project is due to begin in Wales in January 2018.<br>Health Technology Wales (HTW) has been established to deliver a strategic, national approach to the identification, appraisal and adoption of new health technologies into health and care settings across NHS Wales. | On track |
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| **43** | Evaluate different NGS platform configurations, for example:<br>— NGS for clinical condition-specific sets of genes (such as 100–200 of the 22,000 genes<br>— whole exome sequencing (2% of the entire genome)<br>— whole genome sequencing | ENGLAND | As part of developing the genomic testing strategy for the NHS, NHS England has been working with experts to gain consensus on the genetic testing approach for the NHS. This will include determining which tests will be recommended for which condition from single gene tests to multiplex panels and whole genome sequencing. This strategy will be reviewed annually in line with new discovery and research. | On track |
| | | SCOTLAND | The Scottish Genomes Partnership – see above.<br>Next Generation Sequencing – see above. | On track |
| | | WALES | In July 2017 Welsh Government published, Genomics for precision medicine strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales. Additional funding was secured in the 2016-17 financial year from Welsh Health Specialised Services Committee (WHSSC) for very rare genetic tests that are not available in Wales but available via the UK Genetic Testing Network (UKGTN). The Laboratory has a commitment from WHSSC that there will be further funding as more tests become available in the future. | On track |
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| **44** | Support the introduction of | ENGLAND | NHS England is re-procuring its approach to genomic testing across the country. | On track |

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| | NGS into mainstream NHS diagnostic pathways, underpinned by appropriate clinical bioinformatics, including clinical bioinformatics hubs supported by high performance computing centres, where appropriate | | There will be a new genomic infrastructure which will include genomic testing hubs capable of delivering all of the genetic tests required for a population, to defined standards and time scales. This will be delivered against the new genomic testing strategy for rare diseases, which will define which tests will be paid for within the NHS. Each genomic hub will be linked to a national genomic data base with appropriate data and informatics systems ensuring effective communication operating across the whole system. This will include an effective clinical interpretation pipeline to support clinical diagnosis. | |
| | | SCOTLAND | The Scottish Genomes Partnership – see above.<br>Next Generation Sequencing – see above.<br>Genetics Laboratory Consortium – see above. | On track |
| | | WALES | All Wales Medical Genetics service is actively working on a number of projects to enable the introduction of NGS platforms into mainstream diagnostic pathways, supported by the emerging Genomics for Precision Medicine Strategy in Wales. | On track |
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| **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 | ENGLAND | HEE is developing resources to train the workforce in the feedback of WGS results to patients. HEE is training also Genetic Counsellors through a formalised Scientist Training Programme leading to professional registration with the HCPC for the first time. Where relevant other healthcare professionals receive training in genomic counselling skills as part of their professional training programmes - for example GPs. | On track |
| | | SCOTLAND | The Scottish Genomes Partnership – see above.<br>Genetics Laboratory Consortium – see above. | On track |
| | | WALES | For updates please see the Progress Report. | |
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| **46** | Work with industry to set priorities and determine how best to support research into rare diseases and promote research collaboration | ENGLAND | Genomics England works with industry via the Discovery Forum which was launched in July 2017 and builds on the work of the previous GENE consortium. The Discovery Forum provides a platform for collaboration and engagement between Genomics England, industry partners, academia, the NHS and the wider UK genomics landscape. Genomics England also works with companies that specialise in data analysis, so that the 100,000 Genomes Project can benefit from cutting edge advances in handling Big Data.<br><br>The Life Sciences Strategy was published in May 2017, followed by the Life Sciences Sector Deal on 6th December 2017. The Sector Deal will help ensure new pioneering treatments and medical technologies are produced in the UK, improving patient lives and driving economic growth.<br>The NIHR RD-TRC launched an open Collaborative Industry Joint-Funding in | On track |

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| | | | 2015, inviting expressions of interest to facilitate rapid and efficient collaboration between NIHR-funded research infrastructure and industry. There were 23 expressions of interest, 9 of which were funded between 2015 and 2017. | |
| | | SCOTLAND | CSO has co-funded a number of clinical research fellowships with research charities in areas such as Duchenne Muscular Dystrophy, Progressive Supranuclear Palsy, MND and Huntington's Disease. CSO will continue to explore further possibilities for collaboration going forward. | On track |
| | | WALES | Researchers at Cardiff and Vale University Health Board and Cardiff University and have developed a more reliable method of screening for Duchenne muscular dystrophy (DMD) in newborn babies. In collaboration with biotechnology company PerkinElmer, they have developed a diagnostic kit that can accurately screen for the disorder by analysing neonatal dried blood spots. DMD is the most fatal common genetic disorder diagnosed in childhood. The disorder gradually causes muscles to weaken, leading to an increasing level of disability and eventually premature death. DMD almost always affects boys, with around 100 boys born in the UK with the condition each year, and about 2,500 living with the condition in the UK at any one time.<br><br>The new screening test originated from research by Dr Stuart Moat of Cardiff and Vale University Health Board and Professor Ian Weeks from Cardiff University. When PerkinElmer joined the collaboration, the research was successfully adapted to an existing PerkinElmer analyser, allowing it to be translated into a routine test that could be used globally. The new research 'Characterization of a Blood Spot Creatine Kinase Skeletal Muscle Isoform Immunoassay for High-Throughput Newborn Screening of Duchenne Muscular Dystrophy' is published in the journal Clinical Chemistry. | On track |
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| **47** | Support initiatives to facilitate engagement between patients, clinical care teams, researchers and industry wherever practical | ENGLAND | NHS England is working to develop Rare Disease Research Networks (RDRNs) and the Rare Disease Research Centres that will be members of the Networks. The working definition of a RDRN is: a recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes. | Progress being made |
| | | SCOTLAND | Work is ongoing towards a fully developed precision medicine ecosystem in Scotland, involving academia, NHSS and industry, offering world leading support for research in rare diseases. See Scottish Genomes Partnership and SHARE above. | On track |
| | | WALES | In July 2017, Welsh Government published, Genomics for precision medicine strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people | On track |

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| | | | of Wales.<br>Health and Care Research Wales Strategic Plan sets out our vision for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales. | |
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| **48** | Set out the benefits of collaboration (besides producing specific treatments) for all stakeholders | ENGLAND | Genomics England has developed the Genomics England Clinical Interpretation Partnership (GeCIP) to link up researchers and clinicians, from both academia and the NHS, to work together to continually analyse data from the 100,000 Genomes Project and help with the better interpretation of genomic data, better clinical understanding and better patient outcomes.<br><br>Genomics England has also been working with industry via the Discovery Forum which was launched in July 2017 and builds on the work of the previous GENE consortium. The Discovery Forum provides a platform for collaboration and engagement between Genomics England, industry partners, academia, the NHS and the wider UK genomics landscape.<br><br>NIHR also continues to provide world-class research infrastructure in the NHS to support and enable collaboration with research charities, the life sciences industry and other public funders of research. The includes through initiatives such as the NIHR Translational Research Collaborations, and via the NIHR Office for Clinical Research Infrastructure (NOCRI) which enables funders to work in partnership with NIHR infrastructure and ensures that NIHR-supported Centres, Units, Facilities and Networks can work together to help drive the flow of innovative research for patient benefit. | On track |
| | | SCOTLAND | Work is ongoing towards a fully developed precision medicine ecosystem in Scotland, involving academia, NHSS and industry, offering world leading support for research in rare diseases. See Scottish Genomes Partnership and SHARE above and co-funding of research at Commitment 46. | On track |
| | | WALES | In July 2017 Welsh Government published, Genomics for precision medicine strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.<br>Health and Care Research Wales Strategic Plan sets out our vision for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales. | On track |
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| 49 | Continue to build a cohesive infrastructure for implementation and coordination of rare disease research in the NHS | ENGLAND | The NIHR BioResource, NIHR BioResource - Rare Diseases and the NIHR Rare Diseases Translational Research Collaboration are being integrated to form the NIHR BioResource for Translational Research in Common and Rare Diseases. This will create a single national resource founded on the principles of:<br>• A national consent;<br>• A single repository for biosamples<br>• A single database<br><br>This will serve to consolidate and strengthen the nations capacity and capability to support translational research, including for rare diseases. With funding of over £36.5 M over 5 years it will aim to expand the number of patients with rare diseases who are recallable for research and enrich the value of cohorts and the potential for targeted recruitment through genomic and phenotypic profiling. | On track |
| | | SCOTLAND | NHS Research Scotland, formed through a partnership of the NHS Boards and CSO, promotes and supports excellence in clinical and translational research in Scotland so that patients can benefit from new and better treatments. The research support infrastructure provided by NRS includes dedicated rare disease research resource. | On track |
| | | WALES | In July 2017 Welsh Government published, Genomics for precision medicine strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.<br>Health and Care Research Wales Strategic Plan sets out our vision for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales. | On track |
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| 50 | Encourage major research funders to use current structures to coordinate strategic funding initiatives in rare diseases | ENGLAND | The Office for Strategic Coordination of Health Research (OSCHR) has continued its mission to facilitate more efficient translation of health research into health and economic benefits in the UK through better coordination of health research and more coherent funding arrangement to support translation. The Board focuses on coordination and foresight of health research policy as well as addressing key strategic issues. OSCHR continues to maintain a watching brief on rare disease research. | Progress being made |
| | | SCOTLAND | The investments in NHS Research Scotland, the Scottish Genomes Partnership, SHARE and the Precision Medicine Ecosystem are all being actively marketed to both commercial and academic funders of research, including research into rare diseases. | On track |
| | | WALES | In July 2017 Welsh Government published, Genomics for precision medicine | On track |

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| | | | strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.<br>Health and Care Research Wales Strategic Plan sets out our vision for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales. | |
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| **51** | Improve engagement between key stakeholders, including:<br>— patients and relatives<br>— main funding providers<br>— healthcare commissioners<br>— NHS hospitals and specialist care units<br>— industry (pharmaceutical, biotechnology, IT, diagnostics) | ENGLAND | NHS England is working to develop Rare Disease Research Networks (RDRNs) and the Rare Disease Research Centres that will be members of the Networks. The working definition of a RDRN is: a recognised network of member providers, each of which has a demonstrable research-active interest in a rare/very rare disease, the aim of the network being to improve patient outcomes. | Progress being made |
| | | SCOTLAND | The investments in NHS Research Scotland, the Scottish Genomes Partnership, SHARE and the Precision Medicine Ecosystem are all being actively marketed to both commercial and academic funders of research, including research into rare diseases. | On track |
| | | WALES | In July 2017 Welsh Government published, Genomics for precision medicine strategy for Wales, Our plan to create a sustainable, competitive environment for genetics and genomics to improve health and healthcare provision for the people of Wales.<br>Health and Care Research Wales Strategic Plan sets out our vision for Wales to be internationally recognised for its excellent health and social care research that has a positive impact on the health, wellbeing and prosperity of the people in Wales. | On track |