Department of Health & Social Care

The UK Strategy for Rare Diseases

2020 update to the Implementation Plan for England

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Introduction

Document summary

Around 3 million people in the UK will be affected by a rare disease, 80% of which will be of a genetic origin and over 75% of all rare diseases affect children. The publication of the UK Strategy for Rare Diseases (hereafter referred to as 'the Strategy') in 2013 was an effort by all four health departments in the UK to recognise and respond to the needs of all those affected by rare diseases. The Strategy contains 51 commitments setting out a strategic vision for 2013-2020 covering five key areas: empowering those affected by rare diseases; identifying and preventing rare diseases; diagnosis and early intervention; coordination of care; and the role of research.

Implementation plans for England were co-published in January 2018 by the Department of Health and Social Care (DHSC) and NHS England, which together cover actions to achieve the 51 commitments across England. The DHSC 2018 Rare Disease Implementation Plan for England (hereafter referred to as 'the Implementation Plan') covers the 21 commitments in the Strategy for which DHSC have lead responsibility and where we work collaboratively with a range of partners to deliver them. NHS England's Implementation Plan for the UK Strategy for Rare Diseases similarly sets out proposed actions against commitments for which NHS England have lead responsibility. On Rare Disease Day in February 2019, DHSC published an Update to the Implementation Plan for England, which reviews progress and includes actions for 2019/20. NHS England also published a Progress Report on the NHS England-led Implementation Plan in July 2019.

This document is the second update to the DHSC-led Implementation plan. It reviews progress since the last update in February 2019 and takes a forward look of actions for 2020. NHS England commitments are not considered in this document and a second Progress Report is planned for publication by NHS England later in 2020. The main body of this document provides an update on actions for 19/20 outlined in the previous Update across the five themes of the Strategy. **Annex 1** outlines forward looking actions until the end of 2020 for DHSC and partner organisations and **Annex 2** gives a measurement of progress over the last year, where it is possible to do so.

In producing this document, we have worked in close collaboration with our partner organisations and convened a catch-up meeting with delivery partners and patient representatives in November 2019. Partner organisations involved in writing this document include: Genomics England; Health Education England (HEE); Health Research Authority (HRA); Medicines and Healthcare products Regulatory Agency (MHRA); NHS Digital; NHS England; Office for Life Sciences (OLS); Public Health

England (PHE); The National Institute for Health Research (NIHR); UK National Screening Committee (UK NSC).

Policy Context

The rare disease landscape has evolved significantly over the past year. Since the announcement of the NHS Genomic Medicine Service (GMS) in October 2018, there have been major advancements in the development of genomic technologies to improve diagnosis and treatment of patients. The first National Genomic Test Directory was published in March 2019, providing for the first time a comprehensive list of genomic tests available in NHS in England, the technology by which they are or will be available and the patients who will be eligible to access them. The Directory will be updated on an annual basis.

As part of the NHS Long Term Plan commitment to harness the power of genomic testing to rapidly diagnose rare diseases, Government announced in January 2020 that whole exome sequencing would be provided to critically ill babies and children. Whole exome sequencing may more than double the chances of successful diagnosis and are faster than standard practice where multiple tests may be performed one at a time, providing results in days rather than waiting months. It is anticipated that up to 700 babies and children will benefit from the test each year.

Significant commitments to further utilise genomics for prevention of disease have also been made. The Accelerating Detection of Disease (ADD) challenge aims to recruit up to 5 million diverse, healthy participants into a world-leading research cohort in order to shed new light on the detection and treatment of chronic diseases. In September 2019, a £200 million investment from Government, industry and charity was announced to secure the whole genome sequencing of all 500,000 UK Biobank participants, with the first tranche of data available by 2021 and all data available by 2023.

OLS are in the process of finalising a **National Genomics Healthcare Strategy** to be published in early 2020. The strategy will seek to create alignment between our world leading genomics assets and map out a clear direction of travel over the next 5 - 10 years that will enable the genomics community to drive forward innovation for the benefit of all patients.

With the UK Strategy for Rare Diseases coming to an end at the end of 2020, a National Conversation on rare diseases was announced in July 2019 to gather views from the rare disease community to inform a post-2020 framework. To initiate the conversation, a survey was launched in October 2019. The survey was open for 6 weeks and received over 6200 responses from patients, families and carers, patient organisations, healthcare professionals, researchers and life-sciences industry professionals. At its January 2020 meeting, the UK Rare Diseases Policy Board

discussed results of the survey, next steps for the national conversation and development of a framework to follow the Strategy.

An update on progress in 2019/20

Theme 1- Empowering Those Affected by Rare Diseases

At **DHSC** we have continued to engage with patients through the UK Rare Disease Forum and its online platform, Rare Disease UK's Patient Empowerment Group (PEG), and our implementation plan working group which we convene annually to capture progress. We have shared a draft of this document with PEG and are grateful for their helpful comments which made this a better document.

Following feedback from the Policy Board and attendees at the UK Rare Disease Forum Conference in Cardiff in October 2018, DHSC presented a Forum analysis paper to the Board in January 2019, outlining the impact and engagement of the Forum. It was subsequently decided that the Forum would continue. The third UK Rare Disease Forum conference was hosted by the Scottish government in Edinburgh on 29 October 2019 (agenda included in **Annex 3**). The conference provided updates to attendees of developments in rare disease research and genomic medicine, and provided an opportunity for clinicians, service commissioners, patients, patient organisations and government officials to discuss challenges and opportunities for improving rare disease patient care.

As mentioned in the introduction, the **national conversation survey** was launched in October 2019, targeting the rare disease community to identify the major barriers and challenges faced by those living and working with rare disease. The survey was designed and developed with significant input and testing from patient organisations. The survey was open for 6 weeks and we received thousands of responses from patients, family members and carers and patient organisations. Survey responses will be used to inform the development of a post-2020 framework.

At **MHRA**, we have continued to engage with rare disease patients and representatives through the **Patient Group Consultative Forum (PCGF).** The PGCF currently has over 120 individual members, representing a wide variety of health constituencies. Members include Genetic Alliance UK and several groups that represent specific rare disease populations such as Muscular Dystrophy, including Duchenne, and the rarer forms of cancer. The most recent PGCF meeting in January 2020 aimed to seek patient views on the regulation of three 'bio-modifying technologies': 3D printing, gene editing and stem cell therapy, and attendees included representatives from seven different rare disease patient groups. During 2020, rare disease patient groups will be given opportunities to contribute to the development of MHRA's longer-term Strategy for PPE to ensure that it meets their expectations. The PGCF provided patient representatives from three rare disease groups who participated in a joint MHRA/OLS workshop to review the effectiveness of the Early Access to Medicines Scheme since its inception. The Patient, Public and

Stakeholder Engagement team delivered a presentation on the work of the Agency and the PGCF at the Genetic Alliance UK 2019 conference.

MHRA also ran a public consultation, from July to October 2019, on how the Agency should engage and involve patients and the public in its work. A report of the consultation responses will be published in 2020 and the consultation will inform the development of a longer-term strategy for patient and public engagement, including actions to support the increased involvement of those affected by rare diseases.

At the National Congenital Anomaly and Rare Disease Registration Service (**NCARDRS**), we have continued working to strengthen collaboration, including with NICE, NHS Digital, NHS England, clinicians and clinical networks, academics and patient groups. Over the past year we have expanded the number of rare diseases we collect data on and currently collect data on 1,008 rare diseases and congenital anomalies. Data is now collected from 244 healthcare providers across England and we have established automated data feeds from national routine data sources such as Hospital Episode Statistics (HES) and Office of National Statistics (ONS). Though still in a set up phase, we are now in a position to begin to analyse and report on some of the data that we have collected. Two examples published in 2019 include congenital anomaly statistics for 2017 and our first rare disease data briefing on the prevalence of spinal muscular atrophy type 1 (SMA1).

Over the past year, to implement our workplan for rare disease expansion, we have established our rare disease data management system and expanded our partnerships working with a number of clinical and patient groups to establish and validate new data sources and support efforts to improve patient outcomes. One example is the **Registration of Complex Rare Diseases – Exemplars in Rheumatology (RECORDER)** project with the University of Nottingham. We have launched an email-based system to support patient self-reporting and are currently working with two patient groups to pilot the system on a larger scale, including processes for confirmation of diagnoses by treating clinicians. This system will inform the development of a web-based self-reporting system. NCARDRS also continues to support research and have agreed data sharing collaborations with a number of university research projects. Work continuing in 2020 is outlined in Annex 1.

Theme 2 - Identifying and Preventing Rare Diseases

PHE's UK National Screening Committee (UK NSC) ran an open call for new screening proposals between October and December 2018 and received 10 submissions of which five met preliminary criteria for further evidence assessment by UK NSC. This included proposals for newborn screening for two rare diseases: Neurofibromatosis type 1 (NF1) and 22q11 Deletion Syndrome. More details on submission outcomes can be found in the 2018/19 UK NSC annual report. The annual call for 2019/20 screening proposals was open between October and

December 2019. Members of UK NSC met in January 2020 to discuss the submissions with a decision about whether proposals will be taken forward into evidence review expected by June 2020. More information can be found on the PHE Screening website.

Throughout 2019, UK NSC have reviewed evidence for a number of screening programmes including; Biotidinase deficiency, feto-maternal autoimmune thrombocytopenia (FMAIT), Fragile X, Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD), lead poisoning, and severe combined immunodeficiency disorder (SCID) in relation to BCG tuberculosis (TB), spinal muscular atrophy (SMA), sudden cardiac death, thrombophilia, congenital heart disease in newborns and congenital syphilis. Reports have also been published on tyrosinaemia screening cost effectiveness and a model for vasa praevia screening.

Work will continue throughout 2020, including the following programmes: amino acid metabolic disorders; Beta Thalassaemia Major; cervical cancer in vaccinated populations; Congenital Adrenal Hyperplasia; 22q11 Deletion Syndrome; Duchenne Muscular Dystrophy; Newborn- Whole Genome Sequencing (WGS) for Cystic Fibrosis; Glutaric aciduria type 1 (GA1); Galactosaemia; Kernicterus; Klinefelter syndrome; Non-invasive prenatal testing (NIPT) evaluative roll out; Neurofibromatosis type 1 (NF1).

In 2019/20 the NHS and UK NSC have continued to set standards and monitor progress of screening programmes, information of which is publicly available through the NHS Screening Programme annual reports. The most recent report published was for 2017/19 and the 2018/19 report is due for publication in early 2020. UK NSC have also worked closely with clinicians and screening experts to see where genomic technology can be used as a primary screening test, or as a confirmation test, and published the 'Generation genome and the opportunities for screening programmes' report in August 2019.

In light of fast-moving genomic technology developments, the UK NSC reconsidered the original evidence review for the introduction of an evaluative roll out of contingent Non-invasive Prenatal Testing (NIPT) for Down's syndrome, Edwards' syndrome and Patau's syndrome in 2019. The UK NSC agreed that microarray could be used as well as next generation sequencing in the laboratory procurement for the offer of NIPT and that NIPT could be offered in twin pregnancies as part of the evaluative roll out. Given these developments, **NHS England** decided to cancel the existing procurement and will put in place new plans to procure laboratory services for NIPT in line with the latest evidence considered by the UK NSC. More information can be found on the PHE Screening blog.

NHS England published a progress report on the NHS England Implementation Plan in July 2019 and have continued to work on fulfilling action in the Plan, the second update to which is planned for later in 2020.

Theme 3 - Diagnosis and Early Intervention

HEE have continued to deliver the **Genomics Education Programme (GEP)** to meet the needs of the whole NHS workforce to support the implementation of the GMS and the National Genomic Test Directory in the NHS. As part of this work we have undertaken the first phase of baseline workforce data collection for staff in Genomic Medicine Centres involved in rare diseases. This data will be used to inform commissioning of training places, with a focus on specialist workforce such as Genetic Counsellors and Clinical Geneticists, as well as other initiatives around recruitment and retention of staff. The GEP have also been representing the genomics workforce on the Healthcare Science Partnership Board and the Workforce Redesign Partnership Board, to support proposals laid down in the NHS Interim People Plan.

The recommendations for genomics in the independent Topol Review published in February 2019 have been incorporated into the GEP Business Deliverables. Initiatives include the development of a 'just-in-time' digital resource to provide clinicians with relevant and actionable information to be used within the clinical setting. Alongside this, the GEP have continued to meet the training and education needs of the GMS. As well as specific activities related to the new **patient choice model** (discussed under Theme 5) the GEP have continued to develop online resources such as the Genomics 101 series, funded Genomic Medicine Master's tuition fees for over 1600 NHS and PHE staff, supported CPD for variant interpretation, and are in the process of developing cross-professional competency frameworks for the facilitation of genomic testing.

Under the **UK Rare Diseases Policy Board, DHSC** established a **Task and Finish group** in early 2017, chaired by Dr Trevor Cole (Consultant Clinical Geneticist at Birmingham Women's and Children's NHS Foundation Trust), with the aim to further explore and propose improvements to the collection of data to measure the time travelled in the diagnostic pathway for patients with rare diseases. Three conditions, ANCA-related vasculitis, Bardet Biedl syndrome and tuberous sclerosis complex, are being used as exemplars to explore this. In 2019, the group continued their efforts in examining and defining the data for patients with continuing support from **NHS Digital**. The Task and Finish group's report will be published in 2020.

At **OLS** we have continued work to improve the prevention, detection and diagnosis of diseases through artificial intelligence (AI), digital and data. In 2019, we launched the new flagship **Accelerating Detection of Disease (ADD)** challenge, creating a platform for developing effective diagnostics for early, asymptomatic chronic disease

in a joint project with charities, NHS and industry partners. This aims to recruit up to 5 million diverse, healthy participants into a world-leading research cohort. As part of this work, at least three million polygenic risk score assessments will be carried out, and volunteers will each receive personalised feedback on their results. The data created will be made available to researchers from academia and industry creating the largest and deepest data set for medical and diagnostic research in the world.

Work has continued to drive the **Accelerated Access Collaborative (AAC),** which was established in 2018 to identify and support the most transformative health innovations. In 2019, the ACC announced an expansion of its remit to help put the most promising medicines, diagnostic tools and digital services through the clinical development and regulatory approval process faster by providing innovators with support to overcome barriers. Work has also continued in the development of a pioneering programme on digital pathology and radiology using AI, and a competition for a **digital pathology and radiology lab** was launched over the summer 2019. This follows the establishment of five centres of excellence supported by £50m from the Industrial Strategy Challenge Fund over £33m of investment from industry partners. While such programmes are not specific to rare diseases, they work to enable earlier and improved diagnosis of diseases and lead to the development of new processes, practices and products that will benefit the NHS patients, including those with rare diseases.

Theme 4 - Coordination of Care

The majority of actions to improve the co-ordination of care are covered in the implementation plan led by NHS England, however **DHSC** have continued to support work in this area. This included holding a minister-chaired roundtable in October 2019 to discuss early experiences of NHS England's 'rare disease insert' with clinicians and patient groups. DHSC have also continued to support the Diagnostic Odyssey Task and Finish Group (mentioned in Theme 3). Further detail can be found in the 2018 NHS England Implementation Plan and subsequent updates.

In June 2019, the European Court of Auditors published a special report on EU actions for cross-border healthcare, which included an assessment of **European Reference Networks (ERNs)** and made a number of observations and recommendations, including about their impact and long-term sustainability. In autumn 2019, the European Commission opened the call for new members to join existing ERNs. The Rare Diseases Advisory Group ran a process by which interested UK healthcare providers could obtain national endorsement to apply for membership. 841 new clinical units in hospitals from participating countries, including from the UK, requested to become members of the ERNs. The evaluation process started in December 2019 with the outcome unlikely to be known before late 2020. Updates will be published on the European Commission website.

During the transition period, which runs to December 2020, UK healthcare providers remain full members of ERNs. UK participation in ERNs beyond the transition period is subject to on-going negotiation regarding the UK's future relationship with the European Union. In the meantime, we have begun to explore the option of setting up national networks akin to the goal of ERNs to prepare for the possibility of UK exit from ERNs after 2020. This development work will continue throughout 2020 in collaboration with the Devolved Administrations, patient organisations and UK clinicians and other interested parties.

OLS have taken over responsibility from DHSC for providing the secretariat for the minister-chaired **National Genomics Board (NGB)** which met three times in 2019. Further information, including its membership, can be found on the NGB website. Work has also continued on the roll out of the GMS - the first national genomic healthcare service in the world that will incorporate whole genome sequencing and allowing for faster diagnosis and personalised care through equitable access to genomic medicine. As part of the NHS Long Term Plan commitment to harness the power of genomic testing to rapidly diagnose rare diseases, Government announced in January 2020 that whole exome sequencing would be provided to critically ill babies and children.

Theme 5 - The Role of Research in Rare Diseases

The **National Institute for Health Research (NIHR)** have continued to make considerable progress on essential research over the last year. By the end of March 2019, 54 rare diseases had been adopted by the NIHR BioResource for Translational Research, expected to increase to 100 by the end of March 2022. NIHR BioResource have, as of November 2019, recruited over 16,700 patients with rare diseases from 50 NHS Trusts in England with whole genome sequencing data available on over 13,000 of these patients. Research supported by the NIHR BioResource has also identified 99 associations between genes and rare diseases, of which at least 80 subsequently confirmed to contribute to and/or cause the development of the disease or condition.

In February 2019 the NIHR BioResource also secured a £400,000 grant from UK Research and Innovation (led by Health Data Research UK [HDRUK]) as one of ten 'Sprint Exemplar Innovation Projects', aimed at demonstrating the power of data in health research to transform lives. The **Rare Diseases Sprint Exemplar Innovation Project** aims to develop a secure cloud research platform with the potential to transform the understanding of rare genetic disorders, drive improvements in diagnosis and provide proof of principle for use in other diseases. This project will build on the advances which clinical imaging, pathology and genomic technologies have made in understanding rare diseases by creating a secure, anonymised platform to draw together and integrate data from the NHS with research data.

The **UK Standards for Public Involvement in Research** have been developed over the last three years by a four-nation partnership and launched in autumn 2019. The standards have been embraced by research and public involvement networks and organisations and align with international quality standards such as the Patient Focused Medicines Development (PFMD) Patient Engagement Quality Guidance. In 2018/19, more than 40 organisations, groups and individuals were involved in the testing phase of the standards; using the standards in different ways, including as a framework to support reflective practice and future plans for public involvement activities.

At **HRA**, we have continued to work with research communities to provide a consistent and streamlined approach. The 'combined ways of working' pilot for Clinical Trials of Investigational Medicinal Products (CTIMPs) has demonstrated that this integrated process can streamline and speed up approval of clinical trials. The pilot, launched in collaboration with the MHRA in 2018, streamlines processes to a single application and single decision from the regulator and research ethics committee. 100 applications have been submitted to the pilot with input from MHRA, HRA and the devolved administrations, and user feedback has been very positive.

HRA have continued to support the NIHR Clinical Research Network to explore the potential of a 'just in time' model and have worked across the four nations to develop a consistent approach to setting up research studies. This has included the UK roll out of the Local Information Pack in June 2019. The roll out of the pharmacy and radiation single technical assurances has continued throughout the last year and has already demonstrated benefits to support site set-up. HRA have also worked with Cancer Research UK to explore the issues with effective delivery of complex innovative trial designs. Training is being provided to Research Ethics Committee members, and the HRA will continue to support the production of guidance to researchers.

In 2019, **MHRA's** Rare Diseases Interest Group (RDIG) published an article that describes some of the multiple ways that our work helps patients with rare diseases, whether this is to support accurate and timely diagnosis, appropriate treatment, or prevention and surveillance of disease. The paper is open access and published in Expert Opinion on Orphan Drugs.

Genomics England have continued to work with NHS England to engage clinicians, patient groups and participants to develop a '**patient choice mode**l' which covers two key aspects of choice when it comes to the NHS Genomic Medicine Service – clinical care and the option of taking part in research through the 'National Genomics Research Library'. This has been supported by the completion of the Sciencewise Public Dialogue on Genomic Medicine in April 2019, which demonstrated public 'redlines' in relation to genomic data, public ambitions /aspirations for genomic

medicine, concerns the public identifies with genomic medicine, and the language the public understands and uses to talk about genomic medicine.

Genomics England have also continued to work with NHS England and the **NHS Genomic Laboratory Hubs** to develop the systems to facilitate diagnostic whole genome sequencing for rare disease as well as cancer. As of December 2019, the systems are being beta tested by the NHS Genomic Laboratory Hubs. We have also developed proposals for additional research cohorts focussed on a transformation of healthcare linked to the strategic priorities of the NHS Long Term Plan. **NHS Digital** have also continued to work with Genomics England to improve the systems used to record genetic data and other relevant information.

As mentioned under Theme 3, at **HEE**, we have continued to expand the Genomics Education Programme (GEP), developing and implementing training programme for the new hybrid **patient choice model** so that healthcare professionals involved in consenting patients for genomic testing have the knowledge and the skills to guide their patients through this new process. We have been working with NHS England and Genomics England as part of weekly Patient Choice stakeholder group meetings, as well as with clinical services via bi-weekly national Patient Choice WebEx meetings, to inform the development of resources to support the new model. Six outputs have been published on the GEP website, including 'at-a-glance' guides on requesting whole genome sequencing and a cross professional competency framework to support healthcare professionals facilitating patient consent for genomic testing to identify their own learning need and to guide educators and trainers when developing training events.

Finally, **OLS** and Government have continued to make strong progress in implementing both Life Science Sector Deals in collaboration with the life sciences sector, charities researchers and industry, providing the secretariat for the **Life Sciences Council**, which oversees the delivery of the commitments (see also Theme 3). As was cemented in the Sector Deal 2, the Government set out an ambitious vision for genomics in October 2018, announcing a new goal to sequence 5 million genomes over the next 5 years, including 1 million whole genomes from NHS and UK Biobank participants. In September 2019, a £200 million investment from Government, industry and charity was announced to secure **the whole genome sequencing of all 500,000 UK Biobank participants**. The project is unique worldwide – nowhere else is whole genome sequencing being undertaken at this scale, with this level of commitment to open access to researchers.

As part of the **Digital Innovation Hubs** programme, seven Health Data Research Hubs were launched by Health Data Research UK (HDR UK) in October 2019, alongside the first phase of HDR UK's Innovation Gateway – a metadata catalogue held by the 28 members health data custodians in the UK Health Data Research Alliance. This is a first step to enabling a UK-wide life sciences ecosystem that

provides responsible and safe access to health data, technology and science, and research and innovation services. Although not specific to rare disease, these programmes will help to ensure NHS patients can benefit from innovative treatments, including rare disease patients.

Annex 1: 2020 actions to implement the 51 commitments

The Tables below list the specific actions DHSC and its partner organisations will take in the coming year to implement the Strategy commitments across the 5 Themes in 2020. For a full list of the 51 commitments, please see the UK Rare Disease Strategy.

All actions listed below are ongoing throughout 2020, unless a specific date or yearly quarter is provided.

Theme 1 - Empowering Those Affected by Rare Diseases

Actions	Lead	Commitment
Continue to engage patients through the Rare Diseases Forum as well as Rare Diseases UK's Patient Empowerment Group and convene, through the UK Rare Diseases Policy Board, an annual Rare Disease Forum conference. Following approval by the Board to continue with the Forum, aim to expand the membership of the Rare Disease Forum and Platform usage by continuing to: • Identify suitable ways to promote broader forum membership through consideration of current gaps; publish draft UK Rare Diseases Policy Board minutes on the forum platform ahead of formal sign-off; and respond to Forum questions to the Board in a timely fashion. • Work through the UK Rare Diseases Policy Board and with the Devolved Administrations on advice to Government ministers on the future of the Strategy post 2020. • Continue to engage with patients and patient groups as part of the National Conversation and development of a post-2020 framework.	DHSC	C1; C8
Work together on relevant elements in the Life Sciences Industrial Strategy and Sector Deals.	DHSC, OLS	C1; C8

Continue to lead on the implementation of the second sector deal, reporting through the Life Sciences Council which oversees delivery of the commitments and continuing to provide the secretariat to the Council.	OLS	
NCARDRS to continue their work plan (2018-2020) for rare disease expansion. This includes: • **Infrastructure**: Continue ongoing development of data management system including the creation of a guide to coding rare metabolic conditions to underpin standardisation registration from multiple sources with the support of the Metabolic Clinical Reference Group. • **Clinical data liaison**: Continue to work closely with PHE's Office for Data Release, PHE Caldicott Guardian, and IG specialists in other institutions to ensure that data-related activities remain lawful and uphold good practice. • **Multi source approach**: Continue programme of work to validate HES data for sensitivity and specificity for identifying rare disease patients and continue to refine algorithms and improve sensitivity of the rare disease data management system. Publish report on linkage between patients with Wilsons Disease and community prescribing data, combined with hospital reported data. • **Patient portal**: Continue work on piloting the email-based system to support patient self-reporting and development of a web-based self-reporting system. • **Data outputs**: Continue regular meetings with NICE, NHS Digital and NHS England to align objectives and identify issues through horizon scanning. Continue pilot project with Yorkshire and North East and South London Genomic Laboratory Hubs to address some of the technical, ethical and clinical issues arising from the use of molecular diagnostic data to support rare disease registration. • **Research Strategy:** Continue to position NCARDRS as the obvious collaborator for research studies for data storage and linkage.	PHE	C7; C8

Continue to engage with rare disease patients and representatives through the Patient Group Consultative Forum (PGCF) with meetings arranged subject to the requirements of the Agency's regulatory and policy work (an average of three meetings per year). Publication of the public consultation on how MHRA should engage and involve patients and public in its work (January 2020) Development of a longer-term strategy for patient and public engagement, including actions to support the increased involvement of those affected by rare disease. Publication of draft strategy in Q1.	MHRA	C1; C8
Actions regarding the research elements of C7 and C8 are covered below under Theme 5 - The Role of Research in Rare Diseases.		

Theme 2 - Identifying and Preventing Rare Diseases

Actions	Lead	Commitment
Continue with actions from the 2018 Implementation Plan. Continue to review the screening proposals submitted from the open call which ran from October – December 2019. An initial triage review in January will determine whether proposals will be taken on further, with rapid reviews commissioned in 2020 where appropriate. Run an open call for new screening proposals in 2020/21 Continue to regularly assess whether existing screening programmes should be maintained or ceased, including evaluation of maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (HCU) (added to the Newborn Blood Spot	PHE, UK NSC	C9

Actions	Lead	Commitment
Screening programme in 2015) following receipt of health and development outcomes of children who had screened positive. Continue work on the tyrosinaemia screening cost effectiveness, including a workshop and continue work to set up the Severe Combined Immunodeficiency (SCID) evaluation in Q2 /3 (subject to resources). Continue discussions with Genomics England on research activities relating to whole genome sequencing in newborns and continue to consult and examine new methods of non-invasive prenatal testing using genomic technology. Commission further work with affected families and stakeholders to determine acceptability following the presentation of the evaluation results of the use of Whole Genome Sequencing technology for confirmatory diagnosis for Cystic Fibrosis in newborns to the Fetal, Maternal and Child Health Group (FMCH) in January 2020.		
Continue to consider the outcome of any UK NSC recommendation and work to ensure that an appropriate NHS service (where it is the responsible commissioner for that service) is available to treat patients with the identified condition. Evaluation of the roll out of contingent Non-invasive Prenatal Testing (NIPT) following procurement. Continue to review the evidence to consider reflex testing within Down's screening as a major programme modification in accordance to its published evidence review process. This is subject to additional work on practicalities and acceptability by the proposers.	NHS England	C9

Theme 3 - Diagnosis and Early Intervention

Actions	Lead	Commitment
Support the publication of the Diagnostic Odyssey Task and Finish Group report in 2020 and discuss any next steps with the Rare Diseases Policy Boar (Q 2).	DHSC	All relevant commitments
Continue to deliver the Genomics Education Programme to meet the needs of the whole NHS workforce to support the implementation of the NHS Genomic Medicine Service and the new Genomic Test Directory in the NHS. This includes the development of a workforce plan and responding to relevant areas of new health policy and strategy (e.g. NHS Long Term Plan) as well as the independent Topol review.	HEE	C15
Continue to be involved in working group discussions between all partners (including MHRA, NICE, NHS England/I and NIHE) on the development of the Accelerated Access Pathway (AAP).	OLS	C13
Continue the implementation of the ‘Accelerating detection of disease’ challenge.	OLS	C11

Theme 4 - Coordination of Care

Actions	Lead	Commitment
Work with the UK Rare Diseases Policy Board and devolved administrations to facilitate UK-wide collaboration and learning across the four Nations.	DHSC	C31
Work to explore the possibility of setting up national networks in the UK akin to ERNs.	DHSC	C25
Continue to work with PHE via the NHS Digital/PHE Analytical working group. The group promotes a greater understanding and visibility of NHS Digital data developments and analysis	NHS Digital	C25; C29

Actions	Lead	Commitment
and provided early insight into possible emerging issues for PHE uses of NHS Digital data.		
NIHR's actions against commitment C25 are covered under Theme 5.		

Theme 5 - The Role of Research

Actions	Lead	Commitment
Continue to work with the life sciences industry, stakeholder groups and across government to implement the second Life Sciences Sector Deal, including by providing secretariat for the Life Sciences Council which oversees delivery of the commitments.	OLS	C25
Through the NIHR research infrastructure, with support from the NIHR Office for Clinical Research Infrastructure (NOCRI), continue to enable and support collaboration with the life sciences industry, research charities and other public funders of research. Continue to support the NIHR BioResource to recruit patients with rare diseases and increase the number of rare diseases adopted onto the portfolio. Continue work on patient and public involvement, participation and engagement in research; ensuring all research projects, programmes and infrastructure supported by the NIHR have active patient/ public engagement and involvement in their design and conduct. A new contract starting on 1st April 2020 for coordinating this work which will strengthen the impact of PPIPE in research in the coming years.	NIHR	C25; C40; 49; C50
OSCHR Board to hold a further discussion on the co-ordination of rare disease research in June 2020 following discussion at the OSCHR Board in March 2019 on UK rare disease	OSCHR	C50

Actions	Lead	Commitment
research.		
Following EU Exit, MHRA will ensure that the leading role of the UK in paediatric drug development continues by supporting companies and researchers, utilising UK research networks and the input of patient disease groups.	MHRA	C36; C39
Continue roll out of the combined ways of working pilot for Clinical Trials of Investigational Medicinal Products (CTIMPs) to streamline and speed up approval of clinical trials (subject to resource). Continue to work with NHS England and the NIHR Clinical Research Network to support proportionate and efficient site set-up, including costing and contracting processes, and pharmacy and radiation set-up (subject to resource).	HRA	C39; C41; C40
Continue to implement a training programme for the new hybrid (service and research) patient choice model so that healthcare professionals involved in consenting patients for genomic testing have the knowledge and the skills to guide their patients through this new process.	HEE	C45
Continue to work with colleagues in DHSC and the four UK countries to encourage collaboration and data sharing.	NHS Digital	C31
Continue to support the genomics science strand of the National Genomics Board	DHSC	C25
Continue the implementation of the Government's ambition for genomics of sequencing 5 million genomes in the next 5 years including the sequencing of all 500,000 UK Biobank participation. Work with NHS England and Genomics England to maximise the research environment around	OLS	C31

Actions	Lead	Commitment
the NHS Genomic Medicine Service. Publication of National Genomics Healthcare Strategy (Q 1-2).		
Finalise and agree the future Genomics England strategy during Q1 2020. Provisional actions that are subject to agreement of a Genomics England Business Plan: • Work with NHS England and NHS Genomic Laboratory Hubs to go live with diagnostic whole genome sequencing for rare disease as well as cancer commissioned through the NHS via the new NHS Genomic Medicine Service in early 2020. • Work with users to enhance the Genomics England Research Environment to improve researchers' ability to deliver participant benefit from their work. This process has started and will be a major focus of work during 2020. • Work with DHSC, OLS and funders to consider additional research cohorts focussed on a transformation of healthcare linked to the strategic priorities of the NHS Long Term Plan. This process has started and will be ongoing.	Genomics England	C37; C47; C48

Annex 2: Measuring Tangible Progress

The table below shows metrics to measure the progress between January 2019 – January 2020.

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	**Lead**	**Measurement from Jan '19 - Jan '20**	**Commitment(s) being measured**
Deliver Rare Diseases Forum conference to enable attendee-led discussions on highlights, limitations, key challenges and forward view to inform Rare Diseases Policy Board discussions. (A)	DHSC	The third Rare Disease Forum conference was held in October 2019 in Edinburgh, Scotland, with 70 delegates attending.	C1; C36
Number of Forum members active on the Rare Diseases Forum platform. (A)	DHSC	As of December 2019, we have 37 members on the Rare Disease Forum online Platform.	
Publication of minutes from the Rare Diseases Policy Board meetings. (Q)	DHSC	As of December 2019, the Rare Disease Policy Board published the Minutes of each Board meeting (N=4). Following feedback from the Board, a decision was taken to amend the process for minutes publication to improve transparency and throughout 2019 draft minutes were published within six weeks of Board meetings.	
Number of engagements with the Rare Diseases UK's Patient	DHSC	Meeting with delivery partners to discuss document development attended by patient representatives. Engaged PEG in January 2020	

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	Lead	Measurement from Jan '19 - Jan '20	Commitment(s) being measured
Empowerment Group in the review of implementation plan and progress report. (A)		on reviewing this document.	
Number of responses to National Conversation Rare Disease Survey (October-November 2019).	DHSC	The survey was open for 6 weeks and received 6293 responses: • 4303 people living with a rare disease • 1461 family members or carers of someone living with a rare disease • 48 rare disease patient organisations • 426 healthcare professionals including rare disease researchers • 55 life sciences industry professionals and organisations	
Number of engagements with the Rare Disease UK's Patient Empowerment Group in the NCARDRS work plan (O)	PHE	NCARDRS staff attend and present at rare disease meetings, engaging with patients and other stakeholders through meeting stands and presentations (Royal College of Paediatrics Conference, BPSU Tea Party and the Rare Disease Summit). We meet with patient groups on a one-to-one basis including, Shine, Ring 20, NSPKU, Aplastic Anaemia Trust, Wilson Disease Support Group and Alström UK to better understand how we might work together to strengthen national registration of rare diseases.	
Number of patients involved in MHRA and HRA groups and committees.	MHRA/ HRA	MHRA examples include: The Patient Group Consultative Forum – 123 patient representatives; Valproate Stakeholders' Network – 14	

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	**Lead**	**Measurement from Jan '19 - Jan '20**	**Commitment(s) being measured**
(A)		individual patient representatives affected by valproate plus 9 patient groups/charities; Lay Members' Forum – 11 lay members, 3 of whom are patient representatives; Safer Medicines in Pregnancy Consortium – 2 groups that represent women who are pregnant or breast feeding. HRA examples include: about 460 lay members of research ethics committees and the Confidentiality Advisory Group; 97 members of the Public Involvement Network.	
Total number of rare disease patient results returned to GMCs (A)	GEL	As of 12 December 2019, 104,410 analyses have been returned to NHS GMCs – 73,812 of which are for rare disease.	C1; C15; C25; C42 (completed); C45
Total number of rare diseases patients recruited to 100,000 Genomes project (A)	GEL	As of 12 December 2019, the 100,000 Genomes Project has sequenced 119,286 genomes in total, with 83,747 of these for rare disease participants.	
Number of Masters in Genomic Medicine completed (A)	HEE	Figures from the last census in July 2019 show that the Genomics Education Programme have funded 1676 individuals from the NHS and PHE to undertake at least one module of the 21 modules on offer through the Genomic Medicine Masters framework. A total of 626 individuals have completed a full Master's qualification, with 237 people gaining a Post Graduate Certificate and another 51 individuals obtaining a Post Graduate Diploma.	
Total number of NHS and HE staff involved in 100,000 Genome Project	HEE/NHS England	Total number of NHS staff involved in the 100,000 Genomes Project is estimated to be 50,000. Within HEE those directly involved in the	

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	**Lead**	**Measurement from Jan '19 - Jan '20**	**Commitment(s) being measured**
(A)		100,000 Genomes Project via the GEP is 13	
Development of a secure electronic method to enable patients to self-register to NCARDRS (O; A) - Number of patients self-registered to NCARDRS. (A)	PHE	Electronic method for self-reporting was established and went live in December 2019.	C1; C7; C9; C29
Population coverage of NCARDRS across England. (O)	PHE	NCARDRS has 100% population coverage for conditions diagnosed in an antenatal or postnatal pathway, increasing coverage from 49% in 2017/18. NCARDRS have increased the number of conditions collected from 940 last year to 1,008.	
Clinicians treating rare diseases to have access to information collected by NCARDRS (O by 2020; A) – Number of access requests to data held in NCARDRS. (A; from 2020)	PHE	In total 28 data requests were made to NCARDRS including from clinicians.	
Number of response and advice requests to the Rare Diseases Policy Board; Forum and Rare Diseases UK. (A)	DHSC	In 2019 there were a total of three questions asked of the Rare Disease Policy Board via the forum platform. DHSC posted a total of 23 times, this includes responses to questions and information on minutes, agendas and events of the UK Rare Disease Policy Board.	
Number of providers endorsed as	DHSC/	There was a call from the European Commission to increase	C28; C1; C8;

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	**Lead**	**Measurement from Jan '19 - Jan '20**	**Commitment(s) being measured**
members/leads of European Reference Networks. (O)	NHS England	membership of ERNs. DHSC and NHS England ran an application process and endorsed 15 providers as members.	C49
Number of guidance documents developed by HRA/HTA on consent for sharing patient data/ tissue. (O)	HRA	HRA has published a report on attitudes to sharing of anonymised data. HRA has also published guidance on data access and research approvals for data-driven technology.	C40
Number of approaches/enquiries to data provider sites for access to data presented via 'Health Data Finder' (A)	NIHR	1,908 visitors to the Health Data Finder between September 2018 and September 2019.	C35
• Risk-proportionate regulation • Time line for overall approval including both ethical review and assessment for proportionate review studies • Number of amendments for adaptive trials. • Development of a streamlined process to local NHS permissions with a view to reducing timescales.	HRA	HRA have focussed efforts on supporting site set-up, working UK-wide wherever possible and a UK Local Information Pack has been launched, providing a single mechanism across the NHS. HRA have supported work led by NHS England and the NIHR Clinical Research Network on commercial costing and contracting, and non-commercial treatment costs. HRA Approval have now embedded arrangements where the action required of sites to enable the study to be initiated is set out as part of the HRA Approval. This ensures a consistent and risk-proportionate approach to site set-up.	C39; C41
MHRA to ensure risk proportionality	MHRA	MHRA continues to support risk proportionate regulation.	

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	**Lead**	**Measurement from Jan '19 - Jan '20**	**Commitment(s) being measured**
remain a principle in UK clinical trials, marketing authorisations and risk-based inspections after UK's exit from the EU. (O)		There have also been further blog posts on risk adapted approaches for example: • Neonatal pharmacokinetic clinical trial of ciprofloxacin in critical care part 1 • Neonatal pharmacokinetic clinical trial of ciprofloxacin in critical care part 2 • Short format development safety study update report for type A trials	
Monitor the impact of the Joint Statement on the Application of Good Clinical Practice (GCP) to Training for Researchers issued with other regulators, royal colleges and industry in October 2017. (O)	MHRA and HRA	The MHRA GCP Inspectorate have inputted to the NIHR GCP course revision and this work continues as input/review is required. See following links: • NIHR - Good clinical practice • NHS Research Scotland – Training and courses • MHRA Inspectorate – GCP training NIHR Clinical Research Network have provided adapted versions of training for different site staff. HRA now provides a range of learning resources through its website.	
Industry income and number of studies from contract and collaborative studies through the	NIHR	In 2018/19: industry income - £245.7 million (up 6% on 2017/18) number of industry studies – 4,412 (up 11% on 2017/18)	C45; C46; C47; C48; C49; C51

Measure as per the actions in the 2018 implementation plan (Q = quarterly, A = annually, O = ongoing)	**Lead**	**Measurement from Jan '19 - Jan '20**	**Commitment(s) being measured**
NIHR infrastructure. (A)			
Number of participants recruited through NIHR infrastructure. (A)	NIHR	Number of participants recruited to studies supported by the NIHR was 1,024,483 in 2018/19 (up 23% on 2017/18)	
Numbers of grants received / publications in peer reviewed journals. (A)	NIHR	Total publications by NIHR in 2018/19 was 14,242	
Funding for NIHR BioResource. (A)	NIHR	£37.5 million over 5 years from 1 April 2017 to 31 March 2022	
Number of researchers and clinicians links through GeCIPS. (A)	GEL	As of 25 November 2019, there are 3,589 GeCIP members.	
Number of industry partners taking part in the Discovery Forum. (A)	GEL	As of 12 December 2019, the Discovery Forum industry network has grown to 176 companies. Of these, 21 have subscribed to Full Membership, which provides access to the Main Programme dataset within our Research Environment.	
Progress on rare diseases to be considered by OSCHR in 2019.(O; A)	OSCHR	At its meeting in March 2019, the OSCHR Board discussed the extent of UK rare diseases research. Work is being led by the MRC which will be presented to the OSCHR Board for a further discussion on the co-ordination of rare disease research in June 2020.	C50

Annex 3: UK Rare Disease Forum Conference 2019 Agenda

Tuesday 29 October 2019 – City of Edinburgh Methodist Church, Edinburgh

AGENDA ITEM	TIME
Registration: Tea / Coffee	10:00 – 10:30
Welcome and Introduction • Dr Tracey Gillies, Chair, Rare Disease Strategic Oversight Group • Alastair Kent, Independent Consultant and Co-chair of the UK Rare Disease Policy Board	10:30 – 10:45
Theme 1 - Scientific Developments in Rare Disease including research • Dr Rachael Wood, Consultant in Public Health Medicine, NHS National Services Scotland, Information Services Division • Beverly Searle, CEO, Unique Presentation from Scotland on Establishing a Scottish Congenital Anomalies Register	10:45 – 11:00
Breakout Discussion on theme 1 - Looking at the challenges and opportunities brought by scientific developments Breakout groups will be asked to reflect on the CARDRISS presentation and to consider the wider challenges and opportunities that scientific developments can bring to share experience and best practice.	11:00 – 12:00
Networking Lunch	12:00 – 13:15
Theme 2 – Genomics – An opportunity to reflect on the development of genomic medicine and its relevance for rare diseases and the consider next steps Presentation from Genomics England – Department of Health & Social Care. Followed by updates from Wales, Northern Ireland and Scotland • Dr Richard Scott, Clinical Lead for Rare Disease, Genomics England • Professor Tim Aitman, Director of the Centre for Genomic and Experimental Medicine, Institute of Genetics and Molecular Medicine • Professor Zosia Miedzybrodzka, Professor of Medical genetics, University of Aberdeen, Scottish Lipid Forum Genetics lead Lead clinician, Scottish Genetics Laboratories Consortium • Dr Graham Shortland, Chair of the Welsh Rare Diseases Implementation Group • Dr Shane McKee, Consultant in Genetic Medicine & Principal Investigator of Northern Ireland Genomic Medicine Centre. Followed by Panel Session with the Genomics speakers and an opportunity for Q&A	13:15 – 14:00

AGENDA ITEM	TIME
Theme 3 – Co-ordination of Care for Rare Disease Patients • Dr Jayne Spink, CEO, Genetic Alliance UK • Patient Representative video from Keith Swankie • Professor Steve Morris, Chief Investigator, Co-ordinated Care of Rare Disease (CONCORD) Survey for Genetic Alliance UK A day in the life of a Rare Disease Patient - opportunity to hear from rare disease patient and their lived experience of co-ordinated care, including patient support reflections from the third sector (GAUK) Update on the UK Wide CONCORD Project.	14:00 – 14:15
Breakout Discussion on theme 3 – Looking at the challenges and opportunities in delivery of co-ordination of care for rare disease patients • Dr Karen Ritchie, Chair, Rare Disease Working Group, Awareness Raising Amongst Healthcare Professionals Following overview from Dr Ritchie, Breakout groups will be asked to reflect on the previous session and to consider the opportunities and challenges around delivering co-ordinated care and to share experience and best practice from both a Healthcare Professional and Rare Disease Patient viewpoint.	14:15 – 15:15
Tea/Coffee and Poster Stands	15:15 – 15:45
Summary and reflections – includes challenges going forward and opportunities for nations to work together • Dr Tracey Gillies, Chair, Rare Disease Strategic Oversight Group • Alistair Kent, Co-Chair, Genetic Alliance UK	15:45 – 16:00
Close	16.00

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