



Department
of Health &
Social Care

England Rare Diseases Action Plan 2022

Published February 2022

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Ministerial foreword

In January 2021 the 4 nations of the United Kingdom published the UK Rare Diseases Framework, outlining national priorities for improving the lives of those affected by rare conditions. One year on, in this first England Rare Diseases Action Plan, we report on progress made, and take a significant step forward in transforming the collective priorities of the UK Rare Diseases Framework into tangible and concrete action.

Rare conditions are those which affect fewer than one in every 2,000 people. There are an estimated 7,000 different conditions and, owing to their rarity, people living with rare diseases face specific challenges with the health and care system and with wider public services. The rare disease community includes those living with genetic and non-genetic conditions; those with a diagnosis and those who may never receive one; newborns, children, and adults. While individually rare, rare diseases are collectively common. 1 in 17 people are affected by a rare condition at some point in their lifetime: in the UK alone, this amounts to over 3.5 million people.

Despite the immense strain COVID-19 has placed on the health and care system, the actions described in this plan are far-reaching and diverse; from piloting new approaches for patients with undiagnosed rare conditions, to monitoring the uptake of drugs for rare diseases to promote equal access throughout the country, determining how best to include rare diseases in health profession education, and improving the way decisions are made on newborn screening for rare diseases.

These commitments have been developed collaboratively with our delivery partners across the health landscape and in close consultation with members of the rare disease community. To ensure delivery and accountability, each action lists an owner, desired outcomes, and crucially how we will measure and report on progress. An important step in addressing the challenges faced by the rare disease community, this action plan is the first in a series, with annual updates reporting progress made, and new initiatives. Over the course of this year, we will continue to work with delivery partners and the rare disease community to monitor progress and drive change and step up engagement with other organisations with crucial roles in supporting people living with rare disease.

While the framework and action plan represent the government's primary commitments to the rare disease community, it is also important to recognise the strengths of the UK in science and research and the promise of wider initiatives in addressing the challenges of rare disease. Government strategies including Genome UK, the Future of UK Clinical Research Delivery, and the Life Sciences Vision will all

support work to continue improving the lives of patients – from those affected by the most common conditions to the very rarest of diseases.

This action plan, developed in close collaboration with the rare disease community, continues to build upon existing UK strengths and addresses areas of weaknesses; translating ambition to progress. Thank you to all involved in the development of this plan and its future implementation – from officials working across the health and care system, to the clinicians and researchers, the dedicated patient organisations, and, most importantly, those personally affected by rare disease.

Maria Caulfield MP

Parliamentary Under Secretary of State (Minister for Patient Safety and Primary Care), Department of Health and Social Care

Rt Hon Sajid Javid MP

Secretary of State, Department of Health and Social Care

Executive summary

The UK government and devolved administrations published the UK Rare Diseases Framework in January 2021, setting out a shared vision for addressing health inequalities and improving the lives of people living with rare diseases across the UK. The framework outlined 4 key national priorities: helping patients get a final diagnosis faster; increasing awareness among healthcare professionals; better coordination of care; and improving access to specialist care, treatment and drugs.

To turn this vision into action, each of the 4 UK nations has committed to developing nation-specific action plans detailing how the priorities identified in the framework will be addressed. This is England's first Rare Diseases Action Plan, developed together with delivery partners across the health system, and in close consultation with the rare disease community. It sets out specific, measurable actions for the next year under each of the 4 priority areas, including the outcomes that we aim to achieve. Key commitments in this action plan include:

- improving how decisions are made on newborn screening for rare diseases
- designing an ethically approved research pilot using whole genome sequencing to screen for genetic conditions in healthy newborns
- piloting new approaches for diagnosis and care of patients with undiagnosed rare conditions
- determining how best to include rare diseases in UK health professional education and training frameworks
- developing a toolkit to increase the effectiveness of virtual consultations for patients with rare diseases
- supporting rapid access to drugs for patients with rare diseases in the NHS
- monitoring the overall uptake of drugs for patients with rare diseases and mapping this access across the country

By bringing together many key organisations within the health system to deliver this action plan, we have a unique opportunity to tackle the challenges facing those living with rare diseases. Over the course of the coming year, we will monitor the progress of these actions closely, seeking input from those living with rare diseases to ensure we are measuring the outcomes that matter most.

However, we know that there is more to do, particularly as the health system begins to recover from the effects of the coronavirus (COVID-19) pandemic. While delivery of this first action plan is underway, we will continue to explore future directions and develop new actions, informed by the needs of the diverse rare disease community.

Through this action plan we will take the first steps in England towards achieving our overarching vision – delivering improvements in diagnosis, awareness, treatment and care, and creating lasting positive change for those living with rare diseases.

Introduction

On 9 January 2021, the Department of Health and Social Care (DHSC) published the [UK Rare Diseases Framework](#), outlining a national vision for how the UK will improve the lives of those living with rare diseases. All 4 nations of the UK have committed to developing clear and tangible action plans to deliver on our collective framework. This publication is England's first Rare Diseases Action Plan; developed together with delivery partners across the health system and representatives of the rare disease community, to bring about specific and measurable improvements for people living with a rare disease.

It is currently estimated that there are over 7,000 rare diseases, with new conditions continually being identified as research advances. While around 80% of rare diseases have an identified genetic origin, they can also be caused by other factors such as disordered immunity, infections, allergies, deterioration of body tissues and organs, or disruption to development while in the womb.

Although rare diseases are individually rare, they are collectively common – with 1 in 17 people being affected by a rare disease at some point in their lifetime. In the UK this amounts to over 3.5 million people. It is important that the NHS and other services provide this large and diverse patient population with the best possible care.

Rare diseases can be both life-limiting and life-threatening, and disproportionately affect children. 75% of rare diseases affect children and more than 30% of children with a rare disease die before their fifth birthday. People living with rare diseases, and their families, often face a lifetime of complex care leading to a profound impact on their education, financial stability, physical mobility, and mental health. It is vitally important that their voices are heard and acted on when developing wider policy.

UK Rare Diseases Framework

Development of the UK Rare Diseases Framework was based [on the outcomes of the 'National Conversation on Rare Diseases'](#), launched by the government in 2019. The conversation gathered views across the rare disease community on the major challenges faced by people affected by rare conditions across the UK. An impressive 6,293 responses were received, which helped identify 4 high-level priority areas to bring about real change, forming the basis of the UK Rare Diseases Framework. The 4 priorities are: helping patients get a final diagnosis faster; increasing awareness of rare diseases among healthcare professionals; better coordination of care; and improving access to specialist care, treatments and drugs.

To turn these priorities into a reality, we recognise that significant action is needed across the health and social care system. In addition to the 4 priorities, the UK Rare Diseases Framework identifies 5 underpinning themes on which we will focus in support of the 4 priorities, to improve the lives of those living with rare diseases. These themes are patient voice; national and international collaboration; pioneering research; digital, data and technology; and wider policy alignment.

UK-wide implementation

The UK Rare Diseases Framework is a UK-wide document. However, each of the 4 UK nations has its own delivery or implementation group, responsible for drafting and monitoring nation-specific action plans. All 4 nations have committed to publishing their action plans by the end of 2022. Throughout development of the action plans, each nation is engaging with members of the rare disease community, to make sure the work is both relevant and fit for purpose. Once published, action plans will be reviewed regularly to measure progress, update actions, or add new ones.

Developing nation-specific action plans involves balancing the specific health needs of the individual UK nations with the government's commitment to health equity and avoiding disparities. To further help with implementation of the framework, two UK-wide boards have been created: the UK Rare Diseases Framework Board, providing high level coordination of rare disease policy and action plans across the 4 UK nations; and the UK Rare Diseases Forum, providing a way to engage a wide range of stakeholders in the rare disease community for advice and input (see Figure 1). The forum has two parts: a core membership which meets twice a year, and an online knowledge and collaboration platform for continual engagement with a broad range of stakeholders, which both feed into the strategic UK Rare Diseases Framework Board.

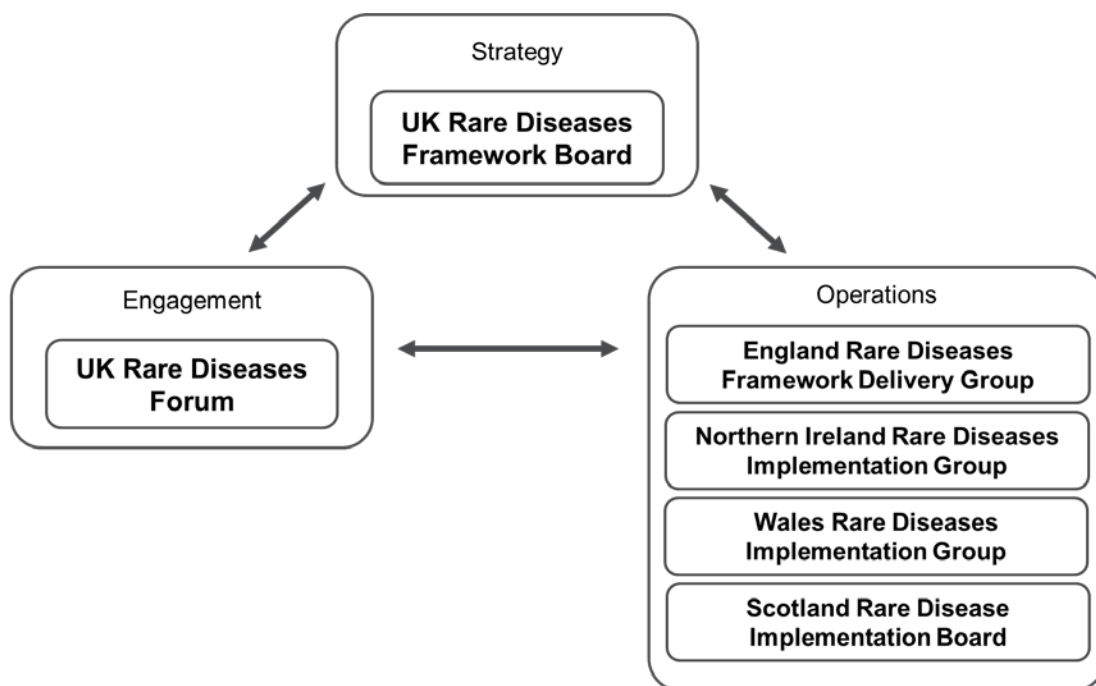


Figure 1. Governance structures for implementing the UK Rare Diseases Framework. Delivery or implementation groups are responsible for developing nation-specific action plans. The UK-wide UK Rare Diseases Framework Board provides strategic oversight and facilitates alignment of policy across the 4 UK nations. The UK Rare Diseases Forum, also UK-wide, provides a means of engagement with the community.

England action plan

The England Rare Diseases Framework Delivery Group develops, oversees, and coordinates delivery of England's action plans. It has brought together publicly-funded delivery partners across the health system including NHS England and NHS Improvement (NHSE/I); the National Institute for Health and Care Excellence (NICE); the Medicines and Healthcare products Regulatory Agency (MHRA); Health Education England (HEE); the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS, NHS Digital); Genomics England; the National Institute for Health Research (NIHR) and Medical Research Council, as major funders of rare diseases research; and representatives of rare disease patient and public voice and the clinician community. Over the course of 2021, the delivery group has met every 6 weeks to develop and agree on actions which have formed the basis of the plan. Following publication of this action plan, the delivery group will continue to meet to coordinate and report on delivery and develop actions for our second action plan in 2023.

As well as the publicly funded delivery partners (listed in [Annex B](#)), whose actions are described here, there are also many other organisations with crucial roles in supporting people living with rare diseases and bringing about much needed change. These organisations include charities, patient advocacy groups, philanthropically funded organisations, independent policy institutes and industry. This action plan sits within this wider system and will help to facilitate continued engagement, increased co-ordination and more joined up working with others.

Community engagement

In developing the UK Rare Disease Framework, and now this action plan, we have placed the needs of those living with rare diseases at the forefront. With the support of Genetic Alliance UK, we recruited representatives of patient and public voice to both the England Rare Diseases Framework Delivery Group and UK Rare Diseases Framework Board. We have used the UK Rare Diseases Forum online platform to engage continuously with a broad range of people from the rare diseases community, providing an opportunity for discussion and feedback, as well as a source of updates on progress and related initiatives. We have also held two community roundtables to seek input on the draft actions and action plan, again with participants recruited with support from Genetic Alliance UK. In November 2021 we launched a targeted online questionnaire to gather detailed feedback on draft actions from people and organisations across the rare disease community. The questionnaire was open for three weeks and received 92 responses from people living with a rare disease, their carers, and family members, rare disease charities, healthcare professionals, industry partners and researchers. We also held a workshop in partnership with [Breaking Down Barriers](#) (a network of over 50 organisations working together to improve the lives of families from diverse and marginalised communities) to better understand health inequalities experienced by people from diverse and marginalised communities affected by rare conditions.

Our delivery partners have also proactively engaged and sought extensive feedback from the rare diseases community as they have developed their actions. For example, NHSE/I hosted engagement sessions with more than 80 individuals representing over 50 different organisations, including patient charities, patient advocacy groups, medical royal colleges, and academics.

Further details of engagement activities can be found in [Annex C](#).

Progress to date

It is now a year since the UK Rare Diseases Framework was published. Alongside the work to produce this action plan, progress has already been made against the priorities of the framework. Some of this progress is summarised below, with further details provided in [Annex D](#).

Significant steps forward have been made in helping patients receive a diagnosis faster, with the [NHS Genomic Medicine Service](#) (GMS) carrying out over 600,000 genomic tests in England over the last year, many of which are for rare diseases. The roll-out of the whole genome sequencing clinical service began in November 2021. Seven NHS Genomic Medicine Service Alliances have been established to support the embedding of genomic testing in end-to-end clinical pathways. An initial national dialogue was held to explore public views on the implications of whole genome sequencing for newborns, and found the public was broadly supportive, providing appropriate safeguards and resources are in place. Additionally, the power of whole genome sequencing (WGS) to uncover new diagnoses for people living with rare diseases has been demonstrated through research such as the [100,000 Genomes Project](#), and [a study focusing on the use of WGS for investigating suspected mitochondrial disorders](#) – both of which resulted in improved clinical decision making for patients or their relatives.

Over the course of 2021, Health Education England (HEE) has continued to develop bespoke education resources to underpin the implementation of the NHS GMS and raise awareness of rare disease among healthcare professionals. Alongside this, work is ongoing to improve coordination of care across the health and social care system. This includes the NIHR-funded [CoOrdinated Care of Rare Diseases \(CONCORD\) study](#) which aims to understand how care for people living with rare disease and common chronic conditions is coordinated, and how they would like it to be coordinated.

On access to specialist care, treatment and drugs, [NICE recently announced a package of changes it will be making to its methods and processes for health technology evaluation](#). The changes will ensure its methods and processes are suited to new and emerging types of technology and provide more equitable access for those with severe diseases. Changes to methods and processes which are relevant to rare diseases are included in this action plan. In March 2021 we published '[Saving and Improving Lives: The Future of UK Clinical Research Delivery](#)' setting out our ambition to create a patient-centred, pro-innovation and digitally enabled clinical research environment. Implementing the vision will unleash the true potential of our clinical research environment to improve health, capitalise on our renowned research expertise, and make the UK one of the best places in the world

to design and deliver research. [An additional £340 million of funding has also been announced, for the Innovative Medicines Fund](#), which will provide early access to promising new medicines, including cutting-edge gene therapies (see Box 1 for further information). Further significant developments included NHSE/I agreeing a number of 'smart deals' to secure innovative medicines. These included three treatments for patients with spinal muscular atrophy (SMA): [nusinersen](#), [risdiplam](#) and the [gene therapy onasemnogene abeparvovec](#).

The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS, NHS Digital) has published studies on the impact of COVID-19 on the health of people with non-genetic rare diseases, helping to steer policy and leading to further pioneering research (see [Annex D](#) for further detail).

Current context

Progress in developing this action plan has taken place against the backdrop of the current COVID-19 pandemic, which has caused significant disruption to health and care services. The effects are ongoing, with the emergence of the Omicron variant and the need to support the vital increase in the vaccination programme having had a significant national impact on the NHS.

The impact of the pandemic has been particularly severe on the rare disease community, as highlighted by the ARDEnt report, '[Making the Unseen Seen: Rare disease and the lessons learned from the COVID-19 pandemic](#)', and a 2020 EURORDIS-Rare Disease Europe [Rare Barometer Survey](#). A focus on COVID-19 has meant that some routine and primary care services have been scaled back, leading to delays or cancellations in diagnostic testing, transfusions, surgeries, scans, and routine appointments. Similarly, safety considerations, redeployed staff and travel restrictions have caused additional barriers to rare disease research, where cohort sizes are already small. Resources have also been diverted to much-needed COVID-19 initiatives, including the development of both vaccines and therapeutics against severe disease. This has affected the operations of many other services and, while we recognise these need to be built back up, this will take time.

Plans are underway to put workforce and technology at the heart of long-term planning across the health service. It was recently announced that Health Education England (HEE), NHS Digital, NHSX and NHSE/I are to become one organisation. This will enable patients to benefit from the best care possible, thanks to a highly skilled workforce and faster digitalisation services. By transitioning these organisations into one, the government and the NHS are ensuring the health and social care sector is fully equipped to face the future and deliver for patients. While work will be needed to finalise logistics, the changes will ultimately better support the

recovery of the NHS, address waiting list backlogs and support hardworking staff, all while driving forward an ambitious agenda of digital transformation and progress.

Our commitment to improving the lives of those living with rare diseases remains as strong as ever, and we will look to learn lessons from the experiences from the COVID-19 pandemic. While ambitious, this action plan is therefore also realistic, recognising that it may take time to implement change within the current context. The actions this year build on the existing strengths of our delivery partners and in many cases, highlight work already underway. However, by publishing them as actions together in this plan we aim to increase transparency and visibility of progress for the rare diseases community. We know that it has not been possible to address all the community's concerns within this first action plan, and we will continue to engage with the rare disease community to determine how further progress can be made in future annual updates to the action plan.

Below, we set out our plans for the year ahead. Additionally, the [Future directions](#) section of this action plan lists further focus areas identified by the rare disease community, to which we will turn our attention over the course of the next year, as services begin to recover from the pandemic.

Addressing the priorities of the framework: actions for 2021 to 2022

The 4 priorities of the UK Rare Diseases Framework have been highlighted as major challenges by the rare disease community. Progress in these areas is vital to meet our commitment to improve the lives of those living with rare conditions. Here we describe actions under each of the 4 priority areas for the next year, including the outcomes that we aim to achieve. Each action is described below with further information provided in Table 1 in [Annex A](#). This table provides details on each action's owner alongside how progress will be measured. This will be reported publicly to aid transparency and accountability and make progress as visible as possible to the rare diseases community. As well as these specific actions, which we will measure and report on, the narrative text below also describes many supporting activities which will all contribute to making progress on the aims of the framework.

Funding for all the actions listed below is already committed, either through delivery partners' existing organisational budgets, or accounted for in the 2021 autumn budget and spending review. In this budget, the Chancellor announced a £5 billion investment over the next three years to increase health-related research and development. This includes funding to support Genomics England's research initiative, a national research pilot testing 100,000 newborns using whole genome sequencing to detect rare diseases with a genetic cause.

Priority 1: helping patients get a final diagnosis faster

Getting a rapid and accurate diagnosis is of vital importance for people living with rare diseases and their loved ones. An accurate and timely diagnosis can facilitate access to treatment and care, provide a possible prognosis, and offer options for family planning. It can also provide a means of connection with a supportive community and open up the possibility of involvement in research including clinical trials. In some cases, if a condition is diagnosed before the onset of symptoms, it may be possible to limit or even prevent harm.

Importantly, a correct diagnosis also ends what, for many, can be a lengthy 'diagnostic odyssey'. The complex nature of many rare conditions means that patients may undergo multiple referrals, inconclusive tests, and sometimes incorrect diagnoses before a final diagnosis is reached, during which time their condition may deteriorate, as well as having a negative impact on mental health. Delayed diagnosis may also mean missing the window of opportunity for certain treatments, such as gene therapies, resulting in poorer outcomes. The COVID-19 pandemic has further

exacerbated challenges in patients receiving a timely diagnosis. Alongside great personal cost, [research from 2018](#) estimated that over a 10-year period the 'diagnostic odyssey' for rare diseases has costs NHS England in excess of £3.4 billion.

Genomics offers enormous potential for increasing diagnosis of rare disease and several of the actions under priority one make use of advances in genomics. This is a strength in the UK and our 2020 strategy [Genome UK – the future of healthcare](#), sets out our ambition to create the most advanced genomic health system in the world. However, it is important to recognise that 20% of rare diseases do not have a known genetic basis, and many rare diseases remain undiagnosed. We have taken this into account throughout this action plan and, under this priority, [action 5](#) is specifically focussed on patients with undiagnosed rare conditions.

Action 1: improving how decisions are made on newborn screening for rare diseases

Newborn screening plays an important role in diagnosing rare diseases early, offering opportunities for treatment, management, and support - often before symptoms even develop. This is particularly important because 75% of all rare diseases affect children, accounting for about a third of infant mortality in the UK.

The UK National Screening Committee (UK NSC) advises Ministers and the NHS in all 4 UK countries on all aspects of screening. Using research evidence, pilot programmes, and economic evaluation, the Committee assesses the evidence for national screening programmes against a set of internationally recognised criteria, taking a range of different factors into account. Proposals to screen for new conditions are considered in an annual call for topics which runs between September and December each year. Currently 9 rare conditions are screened for in newborns through the NHS Newborn Blood Spot Screening Programme.

As part of England's Rare Diseases Action Plan, the Department of Health and Social Care commits to improving how decisions are made across the UK on newborn screening for rare diseases. In the year ahead this will be actioned through:

- a new UK NSC with a broader remit, revised terms of reference, and greater collaboration with researchers and stakeholders, including those with an interest in rare disease
- establishing a UK NSC Bloodspot Task Group to identify practical and innovative approaches to facilitate research and evidence which will inform evaluations of blood spot screening

Opportunities will also be taken to engage internationally and learn from, and contribute to, international best practice on screening. Work is underway to produce a paper which compares the UK NSC bloodspot screening policy processes and programme delivery to proposed [EURORDIS newborn screening criteria](#) for good practice, to identify any areas of good practice and areas for improvement. This will provide an understanding of how UK screening policy and practice compares with an important, patient-defined, checklist. Further details of UK NSC's international activities are provided in the [National and international collaboration](#) section.

The new UK NSC is set to commence work in summer 2022.

Action 2: whole genome sequencing to screen for genetic conditions in healthy newborns

Scientific advances mean that there are an increasing number of conditions where interventions are available to reduce or avoid harm, or improve long term outcomes, if the condition is detected early. Whole genome sequencing could significantly increase the diagnoses of genetic conditions not currently covered by the NHS Newborn Blood Spot Screening Programme.

Guided by the outputs of the public dialogue on whole genome sequencing in newborns published in July 2021, Genomics England and NHSE/I are leading a programme to explore the benefits, risks and broader implications of whole genome sequencing in newborns. This includes co-designing and running an ethically approved research pilot using whole genome sequencing to screen for rare genetic conditions in healthy newborns. New funding for this research pilot was announced in the October 2021 [Spending Review](#). The research pilot will:

- sequence the genomes of up to 100,000 newborns and follow their progress to better understand the clinical benefits and potential risks of newborn genomic screening, aiming to accelerate diagnoses and access to treatments for rare genetic conditions
- be designed through careful consultation with the rare disease community, healthcare professionals, the UK National Screening Committee, and other key stakeholders. For instance, through their NHS [Steering Group](#)
- include careful testing of how the whole pathway – from diagnosis, through genetic counselling and care – can be rolled out within the NHS, if there is proven benefit

- ensure robust processes are in place for consent, return of results, data use and access
- develop a framework to identify genes for targeted analysis, based on clinical and genomic evidence and wide consultation. Outputs from the consultation to date have led towards a focus on diseases which present in early childhood for the research pilot, where an outcome-changing action would be available if they were detected earlier
- improve understanding of the patient experience of genomic newborn screening
- provide evidence to support the UK NSC to make recommendations for future NHS screening programmes

Action 3: continuously develop the National Genomic Test Directory

For the approximately 80% of rare diseases with a genetic origin, the systematic application of genomic technologies has the potential to transform patients' lives. It can enable quicker diagnoses in both adults and children and match patients to the most effective medications and interventions. The NHS Genomic Medicine Service continues to support the implementation of the UK Rare Diseases Framework, through a national network of 7 NHS Genomics Laboratory Hubs, 7 NHS GMS Alliances; a National Genomic Test Directory (Test Directory); an NHS GMS Research Collaborative; and an integrated clinical genomics service.

The Genomic Test Directory sets out the genomic testing strategy, which is delivered through the NHS GMS. It currently includes 357 rare and inherited clinical indications, and is reviewed every year to make sure it reflects the latest scientific and technological developments, including new rare disease clinical indications. The NHS GMS continues to expand the range of testing technologies it offers, as it moves towards targeted and personalised care – including single gene tests, larger next generation sequencing panel tests to help diagnose based on broader phenotypes, exome sequencing and whole genome sequencing – to help improve diagnostic yield. In 2022 steps will be taken to continuously develop the National Genomic Test Directory by [adding new tests](#) for rare diseases where there is scientific and clinical evidence to do so.

Action 4: further develop the Genomics England clinical-research interface

Over the next year Genomics England commits to further developing its clinical-research interface so that when genomic researchers find information of relevance to an individual's health, it can be passed back to the NHS. This will support researchers and clinicians in collaborating to resolve complex or novel findings from genome data. This action builds on existing systems and processes in place for participants from the 100,000 Genomes Project and patients accessing the NHS Genomic Medicine Service, who have opted to make their deidentified data available to researchers from academia or industry in the National Genomic Research Library. The Library is a comprehensive database which provides access for approved researchers to de-identified genomic data, health data and samples. This action to further develop the clinical-research interface should enable more rare disease patients to receive a diagnosis, foster collaborations between researchers and clinicians, and accelerate new discoveries. The number of diagnoses returned to the NHS through the clinical-research interface will be publicly reported, with the goal of returning at least 100 diagnoses per year.

Action 5: pilot new approaches for patients with undiagnosed rare conditions

While advances in genomics offer powerful tools to increase diagnosis, it is critical to support those living with undiagnosed rare diseases, and the healthcare professionals caring for them, to reach a rapid diagnosis. This includes those whose conditions do not have a known genetic cause but who would, like all patients, benefit from a recommended care package and better signposting to existing services.

This year, NHSE/I will pilot new approaches for patients with undiagnosed rare conditions through a combination of outpatient appointments, inpatient stays, and assessment by multiple clinicians. Following consultation with rare disease patients and their families, these pilots are currently under design, but examples could include a holistic one-stop paediatric clinic or a more targeted adult neurology clinic, or the use of virtual expert multidisciplinary teams. All of these will benefit from input from clinicians, genetic counsellors, clinical scientists, healthcare professionals with specific expertise in the management of syndromes without a name ('SWAN' conditions), and potentially, psychological support, depending on the outcome of the intervention and patient need. Pilot approach(es) will be developed in April 2022, with sites selected during summer 2022.

Measuring changes in the 'diagnostic odyssey'

To understand whether these actions have been successful in leading to improvements in the time to diagnosis, particularly for those with non-genetic rare conditions, there is a need to develop an effective and efficient methodology to measure changes in the diagnostic odyssey over time. A recently published [report](#) has demonstrated the feasibility of studying the diagnostic odyssey for both genetic and non-genetic rare diseases in secondary care, using Hospital Episode Statistics (HES) data for three exemplar conditions (Tuberous Sclerosis Complex, Bardet Biedl syndrome and the non-genetic condition, ANCA-associated Vasculitis). The report also sets out a number of recommendations for future evaluations. Over the coming year, we will explore ways to build on these findings, to monitor the impact of interventions on the length of the diagnostic odyssey.

Rare disease research

Research is a key driver of innovation in rare disease diagnostics, and underpins several of the actions described above. Further detail of steps we will take to make sure that research is funded to improve our understanding of rare disease, and help patients get a final diagnosis faster, can be found in the [Pioneering research](#) section of this action plan. The [National and international collaboration](#) section also highlights that the small numbers of patients with individual rare diseases necessitates a collaborative approach to research across the UK, within Europe and globally.

Supporting those without a diagnosis

Receiving a diagnosis can unlock a wealth of opportunities for rare disease management and treatment. However, for some people living with extremely rare diseases, the complex and rare nature of their conditions may mean that they never receive a diagnosis, even after all the appropriate genomic and/or non-genomic diagnostic tests have been carried out and all relevant experts consulted. We are committed to ensuring that these people and their families also receive the support and care they need, and that the healthcare professionals who care for them are equipped to manage their condition, including signposting to sources of support within the rare diseases community, such as [SWAN UK](#).

Priority 2: increasing awareness among healthcare professionals

Raising awareness of rare diseases within the health system is crucial to improving the speed and accuracy of diagnoses, as well as ensuring patients receive the best possible clinical care, particularly in medical emergencies. With over 7,000 rare diseases, it is not possible for healthcare professionals to receive comprehensive training on every condition. It is therefore important that they are aware of rare diseases more broadly, and are alert to considering them. This includes providing training and resources to enable healthcare professionals to recognise rare diseases in patients and be aware of potential specialist treatment needs, as well as signposting to support and care pathways.

Action 6: develop an innovative digital educational resource

Alongside the steps we are taking to create the most advanced genomic health system in the world, healthcare professionals must be offered resources and training to extend their knowledge of genomics, particularly given its application in diagnosing certain rare diseases. Health Education England is developing an innovative digital educational resource, 'GeNotes', to help healthcare professionals make the right genomics decisions at each stage of a clinical pathway by providing concise 'just-in-time' clinical information to support patient management. Content is developed by clinicians who are members of expert working groups that cover different clinical specialties, for example oncology, paediatrics and foetal and women's health. Content will be reviewed on a regular basis through an established governance process and will be updated as required. Further details about this process and membership of the working groups can be found on the [HEE Genomics Education Programme website](#). In addition to linking to the National Genomic Test Directory, it will signpost to extended learning opportunities for clinicians. The resource is being built so that the educational content in GeNotes can be integrated into other digital platforms or websites that healthcare professionals already use, placing easily accessible information on rare diseases at their fingertips. GeNotes is currently in development and the beta phase product is being tested with healthcare professionals to ensure the content is relevant and the design is intuitive for users to navigate.

Action 7: determine how best to include rare diseases in UK health professional education and training frameworks

It is important that training for healthcare professionals across all relevant specialities, including general practice and those involved with emergency care,

includes information on how to engage, inform, involve and support the diverse rare disease population. To address this, Health Education England will determine how best to include rare diseases in UK health professional education and training frameworks. HEE will collaborate with professional organisations, curriculum developers, and other stakeholders (including people living with rare diseases, their families and carers, and charities active in this area such as Medics 4 Rare Diseases) to work out what information on rare diseases needs to be included in healthcare professionals' education and training, from undergraduate education through to continuing professional development. In the coming year, the first stage of this project will be undertaken, which will include the review of existing frameworks and curricula to identify where and when rare disease is mentioned. This will provide the baseline and inform how best to include rare diseases in UK health professional education and training frameworks. Findings will be compiled into a report which will include recommendations to address identified gaps for consideration by curriculum developers.

Action 8: extend the remit of the Genomics Education Programme to include non-genetic rare diseases

Health Education England will extend the remit of their Genomics Education Programme (GEP) to now include non-genetic rare diseases. Current mention of rare diseases within the GEP focuses on those with a genetic cause.

This new programme of work will extend the development of targeted resources for healthcare professionals to include rare diseases with a non-genetic cause, including the establishment of a rare disease education network and digital hub. The network will, in the first instance, provide a mechanism to link stakeholders and organisations with an interest in healthcare professional education in rare disease, and an opportunity through virtual meetings and other communication networks to share projects (for example Medics 4 Rare Diseases resources such as 'Rare Diseases 101) and best practice (for example, involving the rare disease community in co-production of resources). The digital hub will provide an online portal (or a one-stop-shop) with links to education and training resources developed by the members of the rare disease education network. The work of establishing the network and digital hub will be influenced by input from people living with rare conditions, their families, and carers. Bearing in mind that 20% of rare diseases do not have an identified genetic origin, HEE is taking an important step forward in including non-genetic rare diseases within its programme.

Action 9: publish high-quality epidemiological and research papers to increase the understanding of rare diseases

It is important that information used by healthcare professionals is accurate, relevant, and up to date. This can be challenging in rare diseases, where the best evidence available may be based on non-representative samples (like hospital-based studies), or draw conclusions based on small populations. The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) has been working in partnership with academics, clinicians, and patient groups to collect high quality data on rare diseases at a population-level. This work has built some of the largest datasets on rare diseases globally.

As part of England's action plan, NCARDRS has committed to work with existing and new partners to analyse these datasets and publish high-quality epidemiological and research papers, to increase the understanding of rare diseases and raise their profile. This will include papers looking at basic rare disease epidemiology, the impact of COVID-19 on people with certain rare diseases, and cancer-related risk factors or outcomes for people with some rare diseases.

In addition to raising awareness, improving the evidence base on rare disease has the potential for significant impact on quality of life, health economics and joined up care. NCARDRS aim to collaborate on, and publish, at least 6 papers describing novel findings or methods relevant to rare disease by the end of 2022, and will work with patient organisations and healthcare professionals to ensure visibility of these publications.

Priority 3: better coordination of care

Because of the chronic and complex nature of their conditions, many people living with rare diseases will require support from across the health and social care system. This may involve multiple healthcare professionals, covering a variety of different specialties. In some cases responsibility for coordinating appointments and services falls to individuals affected by the condition or a family member or carer, which can result in a significant care burden. An additional challenge is navigating the transition between paediatric and adult services, particularly in cases where no equivalent adult service is available. Improving coordination of care is essential to ensure care is effectively managed, the burden on patients and carers is minimised, and that healthcare professionals are working together to provide the right care at the right time.

While most health services are commissioned locally by clinical commissioning groups (CCGs, to be replaced by integrated care boards subject to the passage of

the Health and Care Bill), NHSE/I commissions about 150 specialised services for the population of England, and sometimes the whole of the UK, where the number of patients is very small. Specialised services support people with a range of rare and complex conditions. In line with its statutory duties, NHSE/I commissions specialised services through over 200 comprehensive service specifications, many of which include services that provide treatment and care for patients with rare diseases.

While planning of specialised services takes place at a national and regional level, NHSE/I works closely with local areas and providers in order to improve care coordination for patients who need specialised treatment. In some cases, NHSE/I commissions specialised services for patients with rare diseases through generic service specifications, for example, [paediatric neurology services](#). In other cases, NHSE/I commissions services for a group of rare diseases that need a similar approach to treatment and care, for example, [adult metabolic services](#). NHSE/I also commissions services directly for certain individual rare diseases, for example, [paediatric cystic fibrosis services](#).

[Highly specialised services](#) are a subset of specialised services which care for people with very rare conditions; usually no more than 500 patients per year. For this reason they are typically delivered nationally, through centres of excellence.

The [Rare Diseases Advisory Group \(RDAG\)](#) is responsible for making recommendations to NHSE/I and the devolved administrations on the development of services for people with rare diseases, and on highly specialised services. This includes how highly specialised services should be commissioned, and which expert centres should be nominated to deliver them. RDAG is updated regularly on progress to implement the UK Rare Diseases Framework.

Through the Health and Care Bill, integrated care boards (ICBs) become statutory organisations that will take on the commissioning functions of CCGs as well as delegated responsibility for some of NHSE/I's commissioning functions (see [Integrated care systems](#) section). This will put more power and autonomy in the hands of local systems, to plan and deliver seamless health and social care services. For all specialised services, NHSE/I will continue to have responsibility for developing and setting standards nationally, which local healthcare providers will be expected to follow. It is likely that highly specialised services will not be delegated to ICBs for commissioning, as the small number of patients and/or providers, and the complexity of the service would make them unsuitable. However, NHSE will work to involve ICBs in the design of those services so that their delivery can be integrated with wider pathways of care.

Rare disease collaborative networks (RDCNs) are part of NHSE/I's provision to support coordination of care for patients with rare diseases. There are currently thirteen RDCNs (listed in [Annex E](#)), each made up of providers (rare disease collaborative centres) who have an interest in a particular rare disease and are committed to working together to progress research, increase knowledge and improve patient experience and outcomes. They are based on the principle that, when it is practical, 'the knowledge moves rather than the patient', and coordinate care by operating national virtual multidisciplinary team meetings and in-person clinics. RDCNs also support the transition between child and adult services by working closely with paediatric services and running transition clinics. RDCNs are supported by the NHSE/I Highly Specialised Commissioning Team and independently set their own priorities and objectives. Proposals for new RDCNs are considered annually at the autumn meeting of the Rare Diseases Advisory Group. An example RDCN can be found in Box 1.

Box 1: Familial Pneumothorax Rare Disease Collaborative Network

A pneumothorax occurs when the lung deflates because of an air leak, causing pain and breathlessness. Primary Spontaneous Pneumothorax occurs in patients with no obvious underlying lung disease, with an incidence of 20 cases per 100,000 per year. In 10% of these cases, another family member has also suffered a pneumothorax, making the diagnosis one of Familial Pneumothorax. A variety of rare inherited syndromes account for many cases of Familial Pneumothorax.

The Familial Pneumothorax Rare Disease Collaborative Network was approved by NHSE/I's Highly Specialised Services in 2021 and has centres in Cambridge University Hospitals NHS Foundation Trust and Queen Elizabeth University Hospital, Glasgow. The network has formalised a route for genetic testing and counselling services, and offers a national online multi-disciplinary team, which meets to discuss patient cases and provide local teams with guidance around diagnosis and treatment, which is delivered locally. If necessary, patients can also attend clinics at the centres in person or by video link. By pooling resources and expertise the network can diagnose patients faster, deliver treatments more quickly, conduct research and work in partnership with other experts for the benefit of patients.

Care coordination should be flexible, keeping the patient at the heart of decision making, and recognising that different patient groups may have different needs. People living with rare diseases and their carers should have the support they need to navigate the health and social care system, and should be empowered to coordinate their own care should they wish to do so. To improve our understanding

of care coordination, the National Institute for Health Research (NIHR) funded the CONCORD ([CoOrdinated Care Of Rare Diseases](#)) study. This University College London (UCL)-led study, involving a consortium of partners including Genetic Alliance UK, gathered evidence on how care is currently coordinated, and sought to determine how care coordination might be centred around the needs and preferences of patients and families affected by rare diseases. Over the coming year we will partner with NHSE/I to explore how the economic case for different care coordination models proposed in the study could best be evaluated, in order to provide the evidence needed to operationalise improved coordination of care within the NHS. Further information about the CONCORD study and its landmark definition of coordination of care can be found in [Annex D](#). We are aware that uncoordinated care is not just an issue for people with rare diseases, similar challenges and treatment burdens are experienced by people living with multiple long-term conditions (MLTC) and their carers. Over the coming year we will also work with colleagues leading the NIHR Strategic Framework for MLTC to consider where programmes of research on MLTC can incorporate and benefit people living with rare diseases.

Better coordination of care for people with rare genetic conditions is also supported by ongoing work to further develop the underpinning NHS Genomic Medicine Service Alliances. This is helping to embed genomic medicine into mainstream patient care pathways in the NHS. The NHS GMS Alliances are developing networks across all other NHS providers and organisations (for example: primary care networks; integrated care systems; academic health science networks; and academia) across the country, to support standardisation and equal access to genomic medicine.

Below we set out the immediate, specific action we will take in the next year to address the coordination of care priority. We appreciate that there is more to do, and this is a priority which will need our particular attention over the coming year so that we can take further steps to address the challenges raised by the rare diseases community. This includes looking at how lessons learnt from specialised and highly specialised services may be applied to care for people living with rare diseases more broadly.

Action 10: develop a toolkit for virtual consultations

There are many potential benefits of using technology and new digital tools to support care coordination, enabling patients to access services remotely and allowing specialists from across the health system to share information and discuss the creation of tailored care plans. The COVID-19 pandemic has catalysed the wider adoption of telemedicine, with a dramatic shift from face-to-face appointments to

virtual consultations. This can be a particular advantage for some rare disease patients who may live a long way from centres specialising in their condition, who may not need a face-to-face appointment on every occasion or who may find travel difficult. The adoption of digital approaches has the potential to improve coordination of care by allowing rare disease patients to access the care of multiple specialists without the need to travel long distances. Building on learning from COVID-19 and findings from consultation with the rare diseases community, NHSE/I will develop a toolkit for virtual consultations, to increase the effectiveness of videoconference and telephone clinic calls in services for patients with complex, multi-system rare diseases. The toolkit will:

- support shared learning about which approaches are most effective
- improve the outcomes of virtual clinics for patients and families/carers by ensuring consistency of approach and agreeing mutual expectations
- support clinical teams in discussing issues that need to be considered in the planning and delivery of virtual clinics
- ensure that both patients and their data are safe and follow agreed governance processes
- facilitate remote clinics across different providers

This will be balanced against the possibility of digital exclusion, to make sure that any changes improve rather than exacerbate, health inequalities. The choice of face-to-face versus digital consultation will usually be made based on the most clinically appropriate option, while taking into account individual patient/family circumstances. Services are developing expertise in appropriately stratifying patients and constantly reviewing and evolving their practice.

The toolkit will be published in spring 2022, and its uptake monitored through annual clinical meetings and reported back to the England Rare Diseases Framework Delivery Group.

A holistic approach to care and support

Despite ongoing advances in medical science, the vast majority of rare diseases currently have no effective, disease-modifying treatment. It is therefore also important that individuals and families living with rare conditions have access to timely advice, aids and equipment, and support in managing both symptoms and the wider impact of the disease.

Improving coordination of care for rare diseases goes beyond treatment, requiring holistic consideration of the support needs of individuals and families across a wide range of public services. Over the course of the coming year, we will look at ways in which the provision of advice for rare disease patients can be improved, including clearer signposting to existing sources of support, and how we can coordinate with other government departments to ensure an integrated package of care.

Recognising that many people living with rare diseases face additional physical challenges, we will also seek to align with other policy areas which address the complex needs of people living with rare diseases (such as the [National Disability Strategy](#)), further details of which can be found in the [Wider policy alignment](#) section.

Mental health care and rare diseases

From waiting for a diagnosis to undertaking multiple avenues of care, individuals and families living with the realities of a rare disease may find the burden affecting their mental health and wellbeing. In some cases, the impact of this has been exacerbated by the COVID-19 pandemic, which has affected the lives of those more vulnerable in society, as well as causing disruption to routine care services. Groups who had the highest risk of mental ill-health before the pandemic, including those living with pre-existing conditions, seem to have been worst affected.

To address mental health challenges more broadly, we're investing an additional £2.3 billion per year by 2023 to 2024 to expand and transform mental health services. We are also reforming the Mental Health Act to give people greater control over their treatment and help ensure they receive the dignity and respect they deserve.

We are tackling the impact of the pandemic on mental health through our [COVID-19 Mental Health and Wellbeing Recovery Action Plan](#) – detailing how we plan to prevent, mitigate and respond to the mental health impacts of the pandemic during 2021 to 2022. The plan is backed by an additional £500 million funding to expand mental health services, accelerate the key commitments in the NHS Long Term Plan, and provide support to those that need it most. Additionally, in 2020, NHSE/I published their [Advancing Mental Health Equalities Strategy](#) which committed to supporting local health systems to better address inequalities in access, experience and outcomes of mental healthcare. The government's [Better Health – Every Mind Matters campaign](#) also supports people to take action to look after their mental health and wellbeing and help support others such as family and friends. The campaign includes expert advice and practical tips to help individuals look after their mental health and wellbeing.

There is always more to do, and so we'll be publishing a long-term Mental Health Strategy looking at what action can be taken to help everyone live healthier, happier lives. A discussion paper will be published soon, calling on the public, charities, businesses, and the mental health sector to share their views and help shape the strategy.

We recognise that people living with rare diseases and their families often have very specific needs for mental health support and that this needs to be well coordinated with their wider health and social care. We are committed to exploring this further in future action plans.

Integrated care systems

Health and care needs for individuals with rare diseases are complex, requiring collaboration across both the health and care systems. To support more integrated care across the system, the government introduced [integrated care systems](#) (ICSs) in 2018 – partnerships between the organisations that meet health and care needs across an area, to coordinate services and to plan in a way that improves population health and reduces inequalities between different groups. [ICSs provide an opportunity to further align the design, development and provision of services – including specialised services – with linked care pathways](#), where it supports patient care, while maintaining consistent national standards and access policies across the board.

Taking this further, the Health and Care Bill proposes to build on the work of existing non-statutory integrated care systems (ICSs) by putting integrated care boards (ICBs) on a statutory footing to replace CCGs as local commissioners, and requiring the creation of integrated care partnerships in each local system area. This proposes to give ICBs clear responsibilities, empowering them to better join up health and care, improve population health and reduce health inequalities. Giving ICBs responsibility for commissioning as many of the services that are accessed by their population as possible is a key enabler for integrating care and improving population health. It gives the flexibility to join up key pathways of care, leading to better outcomes and experiences for patients, and less bureaucracy and duplication for clinicians and other staff.

Currently NHSE/I commissions all specialised services (the 149 'prescribed' services as set out in regulations). The Health and Care Bill includes powers which, if passed, will enable NHSE/I to delegate its functions to other NHS bodies, individually or jointly. NHSE/I is considering how it can use new powers in respect of delegating commissioning responsibility for some specialised services to ICBs individually and

jointly. This would be with a view to realising the benefits of more integrated pathways, joined up care and population health management approach for specialised services and their patients where appropriate.

For all specialised services, NHSE/I will remain accountable for their commissioning, and so continue to set consistent national service standards, and evidence-based access policies, which should be consistently applied across the country. For those services for which commissioning responsibility is delegated, NHSE/I will carry out assurance of the commissioning activity.

Highly specialised services are characterised by small numbers of patients being treated by a small number of providers, using complex treatments. Therefore, delegating commissioning responsibility to ICBs is unlikely to be the appropriate route to realising the benefits of more integrated care. This will better be achieved through involving ICBs in the design of pathways and the development of pathways, and working with local systems to deliver care in a more joined up way.

Primary and secondary healthcare providers will need to work with specialist centres to join up pathways and provide integrated interfaces for patients.

Development and implementation of sustainable, meaningful metrics to measure the impact of these changes on rare disease care will require collaboration across delivery partners, including NHSE/I and NHS Digital (NCARDRS), with input from rare disease stakeholders. These metrics should be used as part of assuring commissioning activity.

Priority 4: improved access to specialist care, treatment and drugs

Many rare diseases do not have established treatments, but where they do exist, they can be life-changing and lifesaving, significantly improving prognoses and an individual's quality of life. Providing access to safe, high-quality specialist care and treatments presents challenges, with some patients needing to travel significant distances to access specialist centres. The small numbers of patients affected by each rare condition mean that the scale of clinical trials typically used to assess the safety and efficacy of medicines may not be possible. Assessment of, and access to, rare disease medicines can also require additional consideration from health technology assessment bodies, due to limited and uncertain data.

The UK is a world leader in science and technology, with a world-class research infrastructure, and an increasing number of innovative drugs and treatments being developed for rare diseases. Ensuring rapid, safe and equitable access to these

treatments as they become available will not only improve the lives of those living with rare diseases, but will foster an environment that will attract substantial investment in the high-value life sciences products of the future and promote continued innovation in the field. There are clear challenges with promoting equitable access, with multiple treatments being developed for some rare conditions, while others do not receive the same levels of research, investment, or attention.

There is a great deal of wider ongoing activity under this priority to improve access to specialist care, treatment and drugs. We begin by describing some of this activity, and the implications for rare diseases, before moving on to describe the specific and measurable actions we will put in place and monitor as part of this action plan.

Promoting access to specialist treatments

Within the UK, a number of schemes exist to promote research and facilitate early access to novel and high-cost treatments (see Box 2). Many of these have the potential to improve access for rare disease patients. Although promoting the development of, and access to, new medicines is vitally important, treatments for rare diseases may also result from identifying new uses for existing medicines. The Medicines Repurposing Programme, hosted by NHSE/I, provides support and, where necessary, public funds, to facilitate repurposing of priority medicines, including those which may be relevant to rare diseases (see Box 3).

Box 2: schemes facilitating early access to rare disease treatments

In July 2021 NHSE/I announced that £340 million of funding per year will be allocated to the new Innovative Medicines Fund (IMF) to provide early access to promising and innovative new drugs that have been granted marketing authorisation. Building on the success of the Cancer Drugs Fund, the IMF will support patients, including those with rare genetic diseases, to get early access to the most innovative and effective new treatments, where further data is needed to support a NICE recommendation on routine funding. Detailed proposals for the Fund were recently subject to a public consultation process, which closed on 11 February 2022.

The Early Access to Medicines Scheme (EAMS), operated by the Medicines and Healthcare products Regulatory Agency (MHRA), aims to give patients with life threatening or seriously debilitating conditions access to medicines that do not yet have a marketing authorisation when there is a clear unmet medical need. This means that patients with rare diseases may gain access to certain medicines before they become available as a licenced treatment option. Under the scheme, the MHRA will give a scientific opinion on the benefit/risk balance of the medicine, based on the data available when the EAMS submission is made. The scientific opinion lasts for 1

year and can be renewed. The scheme is voluntary and the scientific opinion from the MHRA does not replace the normal licensing procedures for medicines.

Medicines supplied to patients with rare diseases under the scheme are still subject to robust safety and monitoring procedures, including a pharmacovigilance system and risk management plan. A drug registry collects relevant data to ensure that any safety concerns can be identified and addressed, and that treatment is taking place in line with the EAMS treatment protocols.

The Innovative Licensing and Access Pathway (ILAP) provides a unique framework for enhanced collaboration between the 4 ILAP permanent partners: the MHRA, NICE, the Scottish Medicines Consortium (SMC) and the All Wales Therapeutic and Toxicology Centre (AWTTC). By supporting expedited, efficient and innovative approaches to product development and patient access, ILAP allows the MHRA and its partner agencies to support the path to market of innovative and novel treatments, while ensuring there are no compromises in assessing the safety and efficacy of the treatments. ILAP's 'innovation passport' designation is the gateway to the pathway and includes a rare disease component among the criteria. The decision on whether to issue an innovation passport is made between the partners and includes input from the ILAP Patient and Public Reference Group which includes rare disease representation.

Access to advice and support for innovators, including those preparing treatments for rare diseases, is available through the MHRA Innovation Office, which provides free and confidential expert regulatory information, advice and guidance to organisations including academia, not for profits and industry. The Office is open to all innovative queries - particularly those that challenge the current regulatory framework – offering developers of rare disease medicines an additional level of visible support. Where new products may create regulatory challenges, this service can provide the opportunity for the MHRA to work with the innovator, offering advice and guidance that clarifies regulatory requirements before formal scientific advice meetings. Of particular relevance to rare diseases, this support is available to manufacturers developing innovative medicines, where technologies or materials are being used for the first time, or where products like gene, cell therapy, or nanomedicines are being developed.

The effective use and development of all of these programmes will help enable patients with rare diseases to access treatments more quickly, with the same degree of confidence. This will require coordination, transparency and partnerships to ensure patients, patient organisations, healthcare professionals and innovators are empowered to navigate the regulatory landscape

Box 3: repurposing medicines

The Medicines Repurposing Programme aims to identify and develop opportunities to repurpose out of patent medicines - meaning medicines used in ways not included in the original licence. Medicines are prioritised and then provided with tailored support, potentially including building further clinical evidence, facilitating licensing, and/or enabling more equitable access. The programme, which began formally in March 2021, is a multi-agency initiative jointly sponsored by NHSE/I, the NIHR, the MHRA, the DHSC, and NICE. A small programme team is hosted by NHSE/I. The first pilot medicine has been adopted into the programme and work on licensing is underway. Key documents, including how to propose a medicine to the programme, will be published during 2022.

Embedding genomics into clinical care through the NHS Genomic Medicine Service will contribute towards improved access to specialist treatments, with more accurate and insightful diagnostics laying the foundations for personalised medicine and facilitating access to clinical trials of new treatments. Under the [National Disease Registration Service Directions](#), NCARDRS has been directed by Secretary of State to collect genomic data to support rare disease registration and already collects, curates and quality assures data from the NHS GMS for a range of rare diseases into its standardised registry. Genomics England is also engaging with industry partners on how the National Genomic Research Library, the database allowing access to approved researchers to de-identified data, can best meet requirements for development of therapeutics and support for rare disease clinical trials, via the Discovery Forum. The Discovery Forum provides a platform for collaboration and engagement between Genomics England, industry partners, academia, the NHS and the wider UK genomics landscape to join up the continuum of research. Companies have come together within the Discovery Forum to work in a pre-competitive environment with access to a selection of whole genome sequences.

Improving clinical research delivery

The safety and efficacy of new medicines are tested in clinical trials. For rare disease medicines there are special considerations, and innovative approaches will be needed to address challenges, including low patient numbers and difficulties patient may face in accessing specialist centres. By working in partnership across the NHS, regulators, research funders, industry, medical research charities, academia, and

government, we can create a clinical research ecosystem which is more efficient, more resilient, and more effective than ever before. This will empower patients, families and carers to explore research opportunities and to make informed decisions about participating in the research that is of relevance to them.

As referenced in the [Progress to date](#) section above, the [Vision for the Future of UK Clinical Research Delivery](#), published in March 2021, sets out the government's ambition for delivering innovative, patient-centred clinical research. A [Phase 1 Implementation Plan](#) was published in June 2021, outlining goals for 2021 to 2022. From expediting ethical approval and study set-up, to fresh investment to digitise clinical research delivery, this plan aimed to increase the UK's capacity and capability to deliver cutting-edge clinical research, with the goal of bringing more research and greater investment to the UK. We are continuing work on the Phase 2 Implementation Plan, which we will publish later this year, and will include actions to support clinical research delivery for rare diseases. During the course of this year we will also convene a workshop to explore the particular challenges facing rare disease clinical research delivery, including barriers to entry and innovative approaches to trial design

Action 11: support rapid access to drugs for patients with rare diseases in the NHS

To ensure that the benefits of the access schemes described in Box 2 can be realised and delivered to patients, NHSE/I has committed to a number of actions to support rapid access to drugs for patients with rare diseases. This includes mapping these schemes to promote understanding of their place in the evaluation pathway, using horizon-scanning and engaging with partners such as NICE and industry (for example through [Office for Market Access](#) meetings) to anticipate which drugs will be available when, and identifying at an early stage any challenges in delivering them to patients. This will allow commissioning processes to be adapted to ensure that NHS services are prepared to deliver drugs as soon as they become available. Mapping will be finalised in summer 2022.

Action 12: develop a strategic approach for gene therapies and other advanced therapy medicinal products

With the expected number of advanced therapy medicinal products (ATMPs) coming to market increasing significantly, there is a need to consider how such therapies are

delivered, adopted and evaluated. NHSE/I will develop a strategic approach for gene therapies and other ATMPs by summer 2022, which will be used to set out the NHSE/I commissioning position to ensure clarity for the pharmaceutical industry, providers, and patients. NHSE/I's initial focus will be on understanding its internal strategic approach, following which there will be engagement with key stakeholders as necessary, including with all companies developing ATMPs. This will include understanding the service impact on the NHS of their treatment, feeding into action eleven. This work aligns with NHSE/I and NICE's consultation on the Innovative Medicines Fund. NHSE/I continues to work closely with colleagues in the devolved administrations on aligning capacity and capability for delivering ATMPs across the UK.

Nucleic acid therapies

Nucleic acid therapies are a class of ATMPs, which involve the delivery of synthetic DNA and RNA (nucleic acids) into cells. Harnessing the cell's natural processes, nucleic acid therapies work to upregulate or downregulate protein production or replace faulty genes completely. As such, they have the potential to effectively prevent and treat a wide range of complex diseases, including rare genetic conditions. Examples include RNA drugs (such as Patisiran, used in treating hereditary transthyretin amyloidosis) and gene therapies (such as onasemnogene abeparvovec, for spinal muscular atrophy).

The UK is a world leader in nucleic acid technology research, and government continues to invest significantly in infrastructure to support the development of these pioneering new therapies through [UK Research and Innovation](#). This includes a total of £18 million, in partnership with LifeArc, to develop a national network of cutting edge '[Gene Therapy Innovation Hubs](#)'; and the creation of a [Nucleic Acid Therapy Accelerator](#) to support interdisciplinary research to solve technical barriers to nucleic acid drug development and delivery (with a total investment of £30 million over 4 years).

Through Innovate UK, government also funds the [Cell and Gene Therapy Catapult](#), a not-for-profit organisation that supports innovation by providing access to expert technical capabilities, equipment and other resources required to take innovation ideas from concept to reality. The Catapult works collaboratively with academia, industry and health care providers to develop new technology and innovation to advance cell and gene therapies. The Catapult also coordinates the [Advanced Therapy Treatment Centre Network](#). The network has been funded by Innovate UK in a four-year, £30M programme to make cell and gene therapies more accessible to patients in the UK. The network does this through training, introduction of new tools

and techniques and preparing clinical centres to process and administer these sensitive treatments.

A new Department of Health and Social Care-funded Clinical Biotechnology Centre at NHS Blood & Transplant's base in Filton, North Bristol, was opened in October 2021. The Centre will specialise in the production of plasmid DNA and viral vectors – advanced products required for early phase clinical trials of experimental gene therapy medicines for rare and other genetic diseases. Collectively these initiatives have the potential to transform care for millions of patients, including those with rare and life-threatening genetic diseases.

Building on advances in therapeutics during the COVID-19 pandemic, government is committed to supporting the continuing development of nucleic acid technology, such as the development of new mRNA platforms that could allow rapid development of new therapies for multiple diseases, including many rare conditions.

Action 13: capitalise on the changes made to NICE's methods and processes to ensure that NICE continues to support the rapid adoption of effective new treatments for NHS patients with rare diseases

The UK aspires to be a world leader for development, testing, access and uptake of new and innovative treatments and technologies. The National Institute for Health and Care Excellence (NICE) is the independent body responsible for providing evidence-based guidance for the NHS on whether medicines represent a clinically and cost-effective use of NHS resources, ensuring that NHS funds are spent in a way which provides the most health benefit for society.

NICE assesses the majority of medicines through its standard technology appraisal programme and also operates a separate highly specialised technologies (HST) programme for a small number of medicines for very rare diseases. This programme is a deliberate departure from the standard approach, recognising these very rare, severe diseases often have limited treatment options, as the uniqueness of the condition creates challenges for research, and difficulties in generating a robust evidence base to bring the product to market and secure access for patients. NICE ensures patients are an active part of the appraisal process from the start of any topic evaluation. The patient contribution plays a part in supporting the committee to understand the condition, as well as the impact it has on patients and families. NICE now appraises all new medicines, and as a result of new commercial flexibilities, is able to recommend the vast majority of new medicines.

NICE has implemented its updated methods and processes for all topics starting after 1 February 2022. NICE gave particular consideration to the suitability of its methods and processes for treatments for rare diseases, as part of a review which took into account a wide range of stakeholder views through a series of consultations.

A number of the changes adopted by NICE will support timely patient access to innovative medicines for patients with rare diseases, through the introduction of additional flexibilities in the appraisal process. For example, NICE has introduced a new severity modifier for committees to consider the severity of the disease or condition under consideration when making recommendations. This reflects evidence that society values more highly health benefits for people with very severe conditions. Many rare conditions are also severe, and committees will be able to give additional weight to the treatment benefits for these conditions. Where there is uncertain evidence in relation to a medicine, a particular issue for those for rare diseases where the population is small, NICE will now adopt a more flexible, and proportional attitude towards evidence uncertainty within its decision making.

In addition, NICE are adopting process changes to help improve participation of patients and clinical experts, introducing a summary of information for patients, as well as more flexibility to adapt consultation timelines for each appraisal, to support more efficient timely access. NICE has also refined the criteria used for routing a topic to the HST programme. This will achieve greater clarity and predictability for stakeholders around when a topic meets the HST criteria. NICE will now implement these changes to make its methods and processes fairer, faster and more consistent – supporting timely patient access to new cost-effective treatments on the NHS, including those for rare diseases. NICE will also be monitoring the effects of the new methods to assess their impact and has committed to undertaking modular updates to the methods as and when new evidence becomes available, and this will be reported to the England Rare Diseases Framework Delivery Group. NICE will continue to provide support to the life sciences industry, including companies developing therapies for rare diseases, further details of which are provided in [Annex E](#).

Action 14: monitor overall uptake of drugs for patients with rare diseases and map geographical access to those drugs

The actions described above all aim to improve access to specialist care, treatment and drugs. However, there can be considerable variability in whether, and how easily, rare disease patients are actually able to get access to them. NHSE/I and NHS Digital (NCARDRS) are developing analytical approaches to monitor overall uptake of drugs for patients with rare diseases and map geographical access to

those drugs. This will improve understanding of the uptake and impact of NHSE/I's high-cost drug commissioning policies and relevant NICE technology appraisals at the population level. It will also provide insight into whether our ambitions of equity of access to treatment for rare diseases, regardless of geography (or whether care is at a specialised centre or not), have been met.

The action involves measuring the overall number of people who are accessing a drug that has been recommended by NICE or commissioned directly by NHSE/I and comparing this to the number of people who would have been expected to access the drug. In addition, NHSE/I will identify if there is a difference between the number of patients accessing the treatment and the number which would be expected for each region. The number of people expected to receive the drug will be measured against the figures set out in NICE guidance, which will have been informed by stakeholder engagement (clinical and non-clinical) in the preparation of the guidance, or from other data sources such as NCARDRS. For most services this exercise will be run annually, with results published in the highly specialised services annual report, and should lead to improvements in uptake and geographical access. Any services identified as outliers will be followed up and a plan developed outlining mitigation measures to improve access.

In parallel, NHSE/I and NHS Digital (NCARDRS) will develop plans to share data and resources to support exemplar project(s) to put access to high-cost drug data into context of the wider rare disease population. By utilising NCARDRS' unique position to collect patient level population-based data on everyone with a rare disease, NCARDRS can compare data on patients who are receiving high-cost treatments to those that are not. This will generate enhanced, actionable, data on geographical variation and potential access inequality in the use of high-cost drug treatments for rare diseases.

Underpinning themes

The UK Rare Diseases Framework sets out 5 underpinning themes where we need to drive progress to create a strong foundation for delivery across all 4 priorities of the framework. These themes are patient voice; national and international collaboration; pioneering research; digital, data and technology; and wider policy alignment. In the framework we also commit to considering health inequalities in developing national action plans. Here, in England's action plan, we include health equity as a sixth underpinning theme. Maintaining momentum across these 6 themes is vital to our work to improve the lives of those living with rare diseases.

Patient voice

People living with rare conditions, their families, carers, and the organisations that support them have a wealth of knowledge and lived experiences to share - it is crucial that their voices are heard. We are committed to putting the voice of the rare disease community at the heart of policy making and to collaborating closely with those affected by rare disease.

With this in mind, we have consulted with the patient community at every level of the development of England's Rare Diseases Action Plan. With the support of Genetic Alliance UK, we recruited representatives of patient and public voice to both the UK Rare Diseases Framework Board and the England Rare Diseases Framework Delivery Group. Delivery partners have also been asked to provide evidence of consultation with the community in both developing and implementing the actions which form the basis of this plan. Examples of this include:

- NHSE/I's engagement sessions with more than 80 individuals representing over 50 different organisations, including patient charities, patient advocacy groups, medical Royal Colleges, industry and academics to inform the development of their actions
- Genomics England's highly engaged Participant Panel who meet regularly to provide advice on research participant priorities, and also sit on key committees including the Ethics Advisory Committee, and the Access Review Committee which oversees data access processes for the National Genomic Research Library
- Health Education England's commitment to establishing a patient and public involvement group as part of its Genomics Education Programme strategy. This group will advise on different aspects of the programme including the HEE

specific rare disease actions, as well as providing a vehicle for individuals with lived experience of genomics and rare disease to input into the resources they develop

The findings from reports published by others in this space have also been considered in the development of the action plan, including the recent Public Policy Projects report, ['Implementing the UK Rare Diseases Framework'](#); ARDEnt's report, ['Making the Unseen Seen: Rare disease and the lessons learned from the COVID-19 pandemic'](#); Alexion's report, ['Reforming Rare Diseases'](#), and Genetic Alliance's ['Rare Experience 2020'](#) patient survey. These reports and others emphasise concerns within the rare diseases community which are echoed within the priorities of the UK Rare Diseases Framework.

During development of this action plan two community roundtables and an online questionnaire were used to capture feedback on the draft action plan from a range of stakeholders, including those living with rare diseases. We additionally partnered with Breaking Down Barriers, (a network of over 50 organisations working together to improve the lives of families from diverse and marginalised communities) to host a workshop specifically aimed at understanding health inequalities in the context of the framework. [Annex C](#) contains further detail of these roundtables and workshops.

The UK Rare Diseases Forum online platform has been used as a key route for continual engagement with the rare disease community, promoting transparency through public posting of minutes and papers from board meetings; a quarterly newsletter with updates from the 4 nations; discussion boards around each framework priority; and a repository for policy and community papers. A summary of issues and suggestions raised by users on the platform have been fed back directly to the delivery group and framework board, who in turn can ask the online platform to consider and provide feedback on specific topics. Now that the action plan has been published, we will continue to seek feedback via the online platform and the stakeholder forum as the actions are implemented.

We will also seek community input regarding the best measures of success when evaluating the outcomes of the action plan, in order to ensure the lives of those living with rare diseases continue to improve under the framework.

National and international collaboration

The small numbers of patients with individual rare diseases make collaboration essential, both for the support of patient care and the delivery of robust research. We are committed to continuing collaboration with the rare diseases community across

the world, including patients, healthcare professionals, researchers, and industry, to share knowledge and ideas to improve outcomes.

We continue to work closely with our counterparts in the devolved administrations to ensure close alignment of the rare diseases action plans each of the 4 nations is developing. One important area of collaboration is on national registries for congenital anomalies and rare diseases. Registries already exist in England and Wales, with a registry under development in Scotland. Work is also underway in Northern Ireland towards developing a rare disease registry. All 4 nations of the UK have committed to working together to make sure that registry activities align so that we can begin to achieve truly national disease registration. This will mean that we will have some of the largest population-based rare disease cohorts in the world and allow us to compare activity and outcomes across nations. Over the next year the national registries will establish a formal work schedule to produce a plan for how to achieve standardisation of the minimum core dataset and inclusion criteria, coding and routine analysis, as well as prioritising efforts regarding development of new methods to support rare disease registration and output. This will also include taking into account stakeholder input on priorities and acceptable practice, meeting fair processing obligations, and including opt out systems for individuals.

We will also continue to build upon and explore new opportunities for international collaboration including through the current European Joint Programme for Rare Disease (EJP RD); and the proposed European Partnership on Rare Disease (which is expected to succeed the EJP RD). It will be important to assess what assets and programmes of relevance to rare disease research, the UK might use to contribute to the goals of this new Partnership, while also investigating the possibility of leading new research actions. We will engage in policy developments emerging from the recent UN Resolution 'addressing the challenges of persons living with a rare disease and their families'. As set out in the National Disease Registration Service Direction we will ensure the UK can continue to contribute to [Orphanet](#) (so that UK data is included in the database) while also making use of this unique global information portal and its nomenclature for coding Rare Disease. Although the UK is no longer able to participate as a member of the European Reference Networks (ERNs), many UK clinicians and patient advocacy groups continue to collaborate effectively with ERNs across Europe.

We will also look to further align with international stakeholder engagement exercises such as the [Rare 2030 foresight study](#), which gathered the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations, under the leadership of EURORDIS-Rare Disease Europe.

The World Health Organisation is currently designing the establishment of a Global Network for Rare Diseases (GNRD). This network will pool resources and connect centres of excellence around the world, to improve diagnosis and care for people living with a rare disease. Over the next year we will engage with WHO to support their work on setting up GNRD, and determine how centres of excellence in England could best engage with and support this network.

At a national level, NHSE/I has developed and implemented a process for recognising rare disease collaborative networks (RDCNs). RDCNs are made up of providers (rare disease collaborative centres) who have an interest in a particular rare disease and are committed to working together to progress research, increase knowledge and improve patient experience and outcomes. There are currently thirteen RDCNs in place (listed in [Annex E](#)). While the networks are coordinated by NHSE/I, providers can be from across the UK, and NHSE/I welcomes both national and international collaboration with RDCNs, including with ERNs to ensure the best standards of care for patients. NCARDS is currently working with the RDCN for Congenital Thoracic Malformations to explore how disease registration can be utilised to support the delivery of national UK-wide clinical care for rare conditions by these emerging networks.

International collaboration is also vital for screening policy. The UK National Screening Committee (NSC) has links with policy making bodies in many countries. Further to this, the NSC's discussion about newborn screening for the rare condition severe combined immunodeficiency (SCID) was informed by contact with New Zealand, the US, and the Netherlands, the learning of which is being transferred and used to inform the evaluation of SCID screening in the UK.

UK screening programmes are linked to European networks to further facilitate improved international engagement. The programme's laboratory adviser is currently President of the International Society of Neonatal Screening. This has facilitated links with German contacts in the discussion about whole genome sequencing in the newborn screening programme. The UK NSC will also continue to work with Genomics England on whole genome sequencing and this will bring opportunities to broaden international relationships with initiatives such as [Screen4Care](#).

Pioneering research

Scientific advances have underpinned many breakthroughs in the field of rare diseases, including the development of innovative treatments. As a global leader in science with a world-class research infrastructure and health system, we must continue to utilise these resources to benefit those affected by rare conditions. The government is committed to supporting ground-breaking, innovative research. In the

October 2021 budget we announced over £5 billion of funding over the next three years for health-related research and development (R&D), which included the largest ever uplift in health R&D.

Many of the actions outlined under the 4 priorities of the UK Rare Diseases Framework either support or are supported by research. Example actions include the development of Genomics England's clinical-research interface, the newborns whole genome sequencing research pilot, raising awareness of rare diseases through the publication of epidemiological and research papers, and monitoring the uptake of high-cost drugs. Given the small numbers of patients living with individual rare diseases, the ongoing work described in the [National and international collaboration](#) section will also be key - connecting researchers with relevant expertise and increasing the pool of potential participants, to enable high-quality research with maximum patient benefit.

The UK has particular strengths in rare disease research. Alongside charities, industry, and other organisations, the government primarily funds research into rare diseases via the National Institute for Health Research (NIHR) and UK Research and Innovation (UKRI). One prominent initiative is the rare diseases component of the NIHR BioResource, which works in over 50 disease areas to link genetic information to clinical characteristics in order to provide greater understanding of disease mechanisms for the development of new treatments and diagnostics. As of December 2021, NIHR BioResource has recruited over 21,230 patients with rare diseases from 50 NHS trusts in England. All participants are genetically characterised and have given consent to be recalled for clinical studies including trials for new treatments. Over the next five years we will provide £40 million of new funding to the NIHR BioResource.

The NIHR has also invested £816 million to support the infrastructure of 20 Biomedical Research Centres (BRCs) around the country. BRCs enable effective collaboration between world-leading universities and NHS organisations, bringing together academics and clinicians to translate laboratory-based scientific breakthroughs into potential new treatments, diagnostics, and medical technologies. The centres undertake themed research across a range of disease and therapeutic areas, including rare diseases, genomics, stem cell therapy and regenerative medicine. NIHR also supports the delivery of research funded by medical research charities and the life sciences industry through NIHR Clinical Research Facilities (CRFs) - purpose-built facilities in NHS hospitals where researchers can deliver early-phase and complex studies - and the NIHR Clinical Research Network (CRN) which supports the set up and delivery of clinical research in the NHS and in other health and care settings.

UKRI's Medical Research Council (MRC) also has a strong focus on rare diseases. The MRC's Population and Systems Medicine Board supports the UK Rare Diseases Framework, encouraging the scientific community to come forward with their best ideas aimed at addressing research questions related to rare conditions. UKRI funds world-leading institutes such as the MRC Laboratory of Medical Biology, MRC London Institute of Medical Sciences, the Francis Crick Institute and Health Data Research UK. National leadership and capacity is provided by assets such as world-leading cohort studies (e.g. UK Biobank and Born in Bradford) and MRC units and centres at the cutting-edge of human genetics, mitochondrial biology, metabolic diseases, and biostatistics. For businesses, the Innovate UK-supported Catapult network provides a gateway to supercharge innovation in the discovery and development of medicines, including cell and gene therapies.

Action 15: map the rare disease research landscape to identify gaps and priorities for future funding

While some rare diseases attract considerable research interest, others are less well studied and understood. With an estimated 7,000 rare diseases it is important to better understand unmet need, engage with the rare disease community and elicit expert opinion to support prioritisation in addressing gaps. To support pioneering research and translate outcomes into frontline clinical care, during the next year we will work with the major funders of rare disease research, including research charities, to map the rare disease research landscape to identify gaps and priorities for future funding. This will involve mapping research against all of the framework's priorities and across all disciplines, including social and economic research. We plan to publish the findings of this work in early 2023 and to feed the outcomes into next year's action plan. We will also seek input on the particular challenges facing rare diseases clinical research delivery, which will feed into ongoing development of policy and strategies in this area. This will be embedded within the Clinical Research Delivery Phase 2 Implementation Plan, which will be published later this year.

Digital, data and technology

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

In September 2021 we published our draft strategy – [Data saves lives: reshaping health and social care with data](#). The strategy highlighted our vision to deliver truly

patient centred care, by giving people better access to their personal health and care data, and a better understanding of how it is used. This is vital for giving patients the peace of mind that health and care staff have all the relevant information about them at their fingertips. This vision will support patients to have digital access to their information (such as test results, medications, procedures, and care plans) from across all parts of the system; systems to easily manage appointments and refill medications; confidence that staff have the most up-to-date information, regardless of the care setting; and increased transparency about how data is protected and used. This vision will better support coordination of care throughout the patient journey including for the rare disease community.

To effectively support patient care and development of new treatments it is crucial that data systems are interoperable, and that data can be easily and securely shared. Understanding how data standards, tools and systems can best work together will allow us to use data science more effectively to advance rare disease diagnostics, treatment, care, research and social support.

NHS DigiTrials, the health data research hub for clinical trials, was established in 2019 to reduce the cost and complexity of developing new treatments and create more opportunities for patient participation in research. Developed in close collaboration with patient and public representatives, the service enables clinical trials to use NHS Digital data efficiently, improving the assessment of trial feasibility and supporting improved planning and delivery of clinical trials in the UK. NHS DigiTrials will help NHS patients across England participate in clinical trials of new treatments, generating results which will influence care for a wide range of conditions, including rare diseases, as well as supporting research.

Data is a cross-cutting theme of the government's strategy for genomic healthcare, Genome UK, with a commitment in its first implementation plan for NHSX to develop the case for investing in infrastructure to support the safe and efficient sharing of genomic data to improve healthcare and research. This work aims to ensure that the UK's existing genomic datasets, including those holding consented data from rare disease patients, are interoperable with each other. We will ensure that the UK's rare disease registries are considered as part of this work. This federated approach would be supported by implementing standards set by the international Global Alliance for Genomics and Health (GA4GH), which the UK continues to fund through the NIHR, MRC and Wellcome. Commitments on this will also be included in forthcoming second-phase second Genome UK implementation plans, due to be published later this year.

Rare disease registries, such as England's NCARDRS, play an important role in helping researchers, clinicians, patients, and service commissioners increase their

understanding of rare diseases. The data collected and held by NCARDRS describes the distribution of rare diseases at a population level, enabling rare disease epidemiology and the assessment of geographical variation in medicine uptake and specialist care access. It can be used to identify potential health inequalities and support clinical research at a national level, while upholding the principles of data autonomy and fair processing obligations, by communicating patients' rights to exercise the national data and NCARDRS opt-outs.

NCARDRS also has a system for patient self-registration, and can carry out collection and analysis of patient self-reported data, and support other stakeholders in doing so. This has been recently demonstrated by the [MELODY](#) study, which collects extensive self-reported data from people with rare autoimmune rheumatic disease (further detail in [Annex D](#)). This allows people and families living with rare diseases to contribute their own data and observations, enriching the phenotypic data that are available for research.

In the coming year we will partner with NCARDRS to host a workshop to gather stakeholder feedback on the existing rare disease registration service. We will explore what more can be done to ensure that, as the lead delivery partner on population-level rare disease data, the service NCARDRS offers meets the need of the rare disease community in delivering the ambitions of the action plan.

Drawing on lessons from the COVID-19 pandemic, which included an increased adoption of telemedicine, NHSE/I plans to create a digital toolkit to support virtual consultations, as set out under [action 10](#) above. This has the potential to significantly reduce the burden on people living with complex, multi-system rare diseases by enabling patients to see more than one specialist simultaneously and alleviating the need to travel to different appointments across the country. As with all digital tools, use of the toolkit will need to be balanced with the need in some specialties for ongoing in-person consultations, and with patients' preferences and ability to access the required technology.

Digital and online resources, such as those described above (under priority two) being developed by Health Education England, provide a valuable tool for healthcare professionals in understanding how best to care for a patient with a rare disease. The resources under development are being linked to existing digital platforms used across the NHS and include training materials, and point-of-care resources providing relevant, concise, and widely accessible information to support patient management.

In terms of using technology for diagnostic applications, the UKNSC is overseeing an in-service evaluation of polymerase chain reaction (PCR)-based screening for severe combined immunodeficiency (SCID). The introduction of PCR technology in the UK labs screening for SCID may make it logistically less complicated to screen

for other conditions, such as spinal muscular atrophy (SMA), provided they meet the UK NSC criteria. The UK NSC secretariat and NICE are in contact to understand how the large study of SMA screening test accuracy might, depending on its outcome, inform SMA screening. The use of artificial intelligence (AI) in bloodspot screening is also a potential topic of interest for the UK NSC and will be factored into the Committee's horizon scanning work.

Wider policy alignment

In implementing the UK Rare Diseases Framework, we will continue to work to ensure that the needs of rare disease patients are recognised in wider policy development. This will include close alignment with the [NHS Long Term Plan](#); [Genome UK](#), the government's UK-wide strategy for genomic healthcare; the [Life Sciences Vision](#); the National Institute for Health Research (NIHR) [Best Research for Best Health: The Next Chapter](#); [Saving and Improving Lives: The Future of UK Clinical Research Delivery](#); and other relevant strategies and policies. Officials with wider responsibility for each of these strategies are members of our governance structures.

Improving the lives of those living with rare diseases goes beyond healthcare, encompassing a wide range of potential interventions including housing adjustments, social care, financial aid, mental health support and special educational needs support. As described under priority three above, there are policy initiatives with particular importance for coordination of care with which we continue to align. These include initiatives on mental health and integrated care systems. Some people living with rare diseases have additional physical or learning needs, and we will continue to explore ways in which these can be supported through policy, such as the 2021 [National Disability Strategy](#), and recent [NICE recommendations regarding people with learning disabilities](#).

With the increasing use of genomics in healthcare, workforce planning is critical. In alignment with the NHS Long Term Plan and delivery of the Genomic Medicine Service, Health Education England (HEE) is committed to educating and training the current and future workforce, reflected in this action plan through the training of future healthcare professionals on rare disease (see [action 7](#)). HEE, working with NHSE/I, will undertake periodical workforce data collection to aid understanding of workforce numbers and required configuration in the key speciality areas that focus on the care for people with both genetic and non-genetic rare diseases, in health and social care.

Health equity

Some people living with rare diseases may face additional barriers to accessing services and support, beyond the immediate challenges of their condition. The principles of the framework commit the 4 nations to “ensure any impacts on health inequalities are considered when developing action plans”. This includes involving a diverse range of voices at every level of policy development.

As part of the development of England’s action plan, we have sought to better understand the health inequalities experienced by people from diverse and marginalised communities who are affected by a rare condition, and to explore how health inequalities could be addressed under each of the framework priorities. The Breaking Down Barriers workshop and stakeholder publications such as the [“Whose Voice is it Anyway?” meeting report](#) (summarising the findings of an NHSE/I engagement session hosted by RareQoL and Medics 4 Rare Diseases) highlighted a number of common themes, including the need for:

- a holistic approach to care and support, which considers the needs of the whole family, both at the point of diagnosis and over the longer term
- accessible resources, taking into account people’s lived experiences, and the challenges associated with communicating complex medical terms across cultural and language barriers
- developing and maintaining trust in healthcare professionals

With this in mind, Health Education England is committed to ensuring that the resources and promotional materials it develops reflect the diversity of the UK population, both in terms of design and in the use of inclusive language. HEE’s competency frameworks include information on cultural awareness, and healthcare professionals can access a suite of online resources to help them support patients and their families through the Genomics Education Programme. HEE also plans to enhance the involvement of the rare diseases community in the Genomics Education Programme, by including individuals in the development of resources where appropriate.

To ensure that the benefits of genomic healthcare are relevant to all communities, Genomics England is working on a ‘Diverse Data’ initiative to increase representation of minority groups in genomic research programmes. The initiative aims to tackle health inequalities by recruiting at least 15% of people from ethnic minority backgrounds to take part in research programmes, thereby reducing the historical bias towards populations of European ancestry.

Certain rare conditions, such as sickle cell disease, severe combined immunodeficiency (SCID) and tyrosinaemia, disproportionately affect minority ethnic groups or communities, and it is important that these are not overlooked. An in-service evaluation of newborn screening for SCID has begun and a consultation on newborn screening for tyrosinaemia by the UK NSC is currently underway. Barriers to entry for clinical trials, including the need for services to be delivered locally, will be considered as part of the workshop on clinical research delivery mentioned above.

Action 16: reduce health inequalities in NHS highly specialised services

NHSE/I has committed to look at ways to build on existing best practice such as the use of Equality and Health Inequalities Impact Assessments to further reduce health inequalities in their highly specialised services (HSS), through improved data collection, data sharing (e.g. with NCARDRS), training, and sharing best practice. This includes considering health inequalities at HSS annual clinical meetings, in service development and commissioning decisions, and in provider selection processes.

In addition, [action 14](#), described above, will monitor overall uptake of drugs for patients with rare diseases and assess geographical variation in the use of high-cost drug treatments for rare diseases using population-based data in collaboration with NCARDRS. This will be important in identifying and quantifying potential health inequalities in terms of access to treatments.

Monitoring and evaluation

The 2021 UK Rare Diseases Framework commits to working closely with the rare diseases community to ensure the commitments developed are actionable, measurable, and regularly reviewed.

All of the individual actions within this action plan are underpinned by a [logic model](#), setting out the problem the action addresses, a clear organisational owner for the action, the outputs and outcomes that will be delivered, and the metrics which will be used to measure progress and effect of each outcome. Following publication of this action plan, the delivery partners which make up the England Rare Diseases Framework Delivery Group will continue to meet regularly to report on progress on these actions and identify any barriers to implementation which need to be resolved. We will also continue to work together across the 4 nations of the UK to align policy and share, and learn from, best practice.

A central objective of this action plan is to improve transparency and to make sure that progress is visible to the rare disease community. The second England Rare Diseases Action Plan will be published at the start of 2023, and will report on progress against the actions set out here as well as proposing updated and new actions.

As well as being able to measure progress against individual actions, it is also important to be able to see whether the measures we are introducing are bringing about real change for people living with rare diseases. It can be challenging to capture the impact of individual actions on the overall patient experience, which despite progress may also be influenced by wider external factors, as we have recently seen during the COVID-19 pandemic. Studies such as the recently proposed [mixed-methods evaluation of the NHS Genomic Medicine Service for paediatric rare diseases](#) provide a valuable template, gathering views from those designing, implementing and using the service to better understand its outcomes and impact.

One way of measuring change is through NCARDRS, which has been directed by the Secretary of State for Health and Social Care to collect data “to help the NHS in England, researchers, charities, people with congenital anomalies and rare diseases, and the public understand the prevalence of congenital anomaly and rare disease in England and to support wider understanding and treatment of these conditions”. We will utilise the expertise and infrastructure within NCARDRS (NHS Digital) to understand the impact of relevant actions using patient level data.

Our aim is to improve the lives of people living with rare diseases and, in consultation with the rare disease community, following the publication of this action plan, we will also develop high-level metrics to support assessment of progress against each of the framework's priorities. This is likely to include specifically designed surveys to create baseline data against which to measure change. In so doing we aim to find robust approaches to address the following questions:

- do rare disease patients experience more rapid diagnosis?
- has awareness increased among healthcare professionals?
- is care better coordinated?
- is there improved access to care, treatment, and drugs?

As well as being able to report on the progress of each action, we aim to be able to measure progress in what matters to people living with rare diseases – improvements to the quality of care that they receive.

Future directions

This is the first Rare Diseases Action Plan for England, setting out specific, measurable commitments to improve the lives of those living with rare diseases, which will be delivered over the course of 2022 to 2023. Although ambitious in their scope, these commitments are set against the backdrop of a health system which is continuing to manage the impact of new SARS-CoV-2 variants. While we remain committed to improving the lives of those living with rare diseases, it will not be possible to address every issue within this first year. The actions we have put forward do, however, create strong foundations on which we can build in future years.

During the coming year, the England Rare Diseases Framework Delivery Group will monitor and report on progress against the current actions. Progress will be reported in meeting minutes with a more detailed report in the second England Rare Diseases Action Plan at the start of 2023. At the same time, we will continue to engage with stakeholders through the UK Rare Diseases Forum and on-line platform to gather feedback on implementation of actions. As described above we will work with the rare disease community to develop an approach for monitoring and evaluation.

We are aware that the rare diseases community has raised concerns which have not been addressed, or only partially addressed, in this action plan, which we will need to turn our attention to over the coming year. The England Rare Diseases Framework Delivery Group will continue to meet every 8 weeks and, as well as reporting on progress, will develop new actions to address outstanding community concerns. These will be informed by delivery partners' own engagement activities, feedback from the UK Rare Diseases Forum and on-line platform and workshops (such as those described above on NCARDS and clinical research delivery). Areas where we will focus include:

- making sure future actions provide additional, proportionate support for those with non-genetic conditions
- exploring ways of providing wider support and coordinated care for people living with rare diseases, including those who remain undiagnosed, for example through social care provision, mental health support and special education support
- taking steps to improve access to advice and supportive resources for rare disease patients

- supporting people living with rare conditions and their families during the transition from paediatric to adult health and care services
- improving equitable access to specialists and post-diagnosis care
- strengthening rare disease networks and multidisciplinary teams
- ensuring care is given according to best practice guidelines
- continuing to improve data collection on rare diseases by NCARDRS, including the ongoing expansion of existing data feeds from Genomic Laboratory Hubs to include the entire repertoire of rare disease National Genomic Test Directory results
- working across all delivery partners to improve access to rare disease data and reduce data silos, harnessing the power of data to assist diagnosis, research and decision-making
- ensuring as many people with rare diseases as possible have the opportunity to be entered into good quality research studies
- continue to work with regulators to ensure rapid, safe and equitable access to innovative therapies, including new technologies such as ATMPs
- continuing to collaborate across the UK and abroad, learning from examples of best practice, and pooling resources to secure the best outcomes for small patient populations

Recognising that there are many organisations with crucial roles in supporting people living with rare diseases beyond the publicly funded delivery partners listed in this action plan, we will look to partner with patient organisations and charities already performing valuable work under the 4 priorities of the framework. Most importantly, we will ensure, through ongoing dialogue, that people with rare diseases remain at the heart of the decision-making process.

Annex A: summary of actions

Table 1. Summary of actions

Action	Lead organisation(s)	Outputs	Outcomes	Action-specific monitoring and evaluation	Framework priority
1. Improving how decisions are made on newborn screening for rare diseases	DHSC	<p>Having already completed stakeholder engagement, a new UK NSC terms of reference and remit</p> <p>UK NSC Bloodspot Task Group terms of reference</p> <p>UK NSC Bloodspot Task Group products:</p>	<p>UK NSC structures and processes enabled to more fully support the research and evaluation effort in newborn screening / rare diseases</p> <p>UK NSC methodological guidance available to inform the research effort in newborn screening / rare diseases</p>	<p>UK NSC terms of reference publicised in spring 2022</p> <p>UK NSC bloodspot task group terms of reference publicised in spring 2022</p> <p>First new UK NSC meeting held in summer 2022 with research subgroup established, formal</p>	1

(1) baseline comparison between UK and EURORDIS key principles for newborn screening

(2) manuscript advising on the methodological principles for screening test accuracy study designs in rare disease settings, such as newborn blood spot conditions. Expected to be published on gov.uk and to be submitted to a peer review journal

(3) paper on technical

horizon scanning process defined, access to modelling capacity considered

First UK NSC bloodspot task group meeting held in spring 2022 with outputs defined, timescales set and intention to publish outputs stated.

		<p>and procedural considerations for modelling exercises around newborn screening to bring consistency to UK NSC modelling exercises</p> <p>(4) paper on use of registries in the newborn screening evaluation process</p>			
<p>2. Whole genome sequencing to screen for genetic conditions in healthy newborns - designing an ethically approved research pilot</p>	<p>Genomics England and NHSE/I</p>	<p>Pilot ready to implement at a small number of NHS trusts.</p> <p>Results pathway mapped for return of findings to NHS clinicians and</p>	<p>Clinicians across all relevant medical specialties have been engaged.</p> <p>Detailed plans are in place to capture patient experience of genomic newborn</p>	<p>Number of NHS pathways in place to support families with positive results.</p> <p>Number of engagement interactions completed related to</p>	<p>1, 3 and 4</p>

patients.

Mapping of NHS Pathways in place to support babies with positive results.

Co-designed principles have been developed to underpin the candidate list of conditions to be looked for in the analysis.

Modelling using existing data to determine yield of different analysis approaches to genomic newborn screening is

screening.

Evidence for commissioning decisions on future genomic screening has been defined.

pilot implementation and research planning.

		finalised/available.			
3. Continuously develop the National Genomic Test Directory - including rollout of whole genome sequencing (WGS) that will play an important role in diagnosis of rare diseases.	NHSE/I	<p>Phase 2 and 3 clinical indications for WGS will be launched in 2021 to 2022.</p> <p>An annual process for updating the Test Directory will have been implemented in Q4 2021 to 2022.</p>	<p>Patients in the NHS in England will be able to access a world leading NHS GMS, offering the full repertoire of genomic testing technologies from single gene testing to WGS.</p> <p>The NHS GMS will support equitable access to genomics across the NHS in England and provide standardised care across the population.</p> <p>The NHS GMS will continue to implement new technologies as the testing strategy</p>	<p>Patient Level Contract Monitoring (PLCM) data will show the genomic testing strategy is being delivered across England, with increased activity as the range of conditions on the Test Directory is expanded, and developments in technology are introduced in the NHS GMS.</p> <p>All 7 NHS Genomic Laboratory Hubs (GLHs) will be delivering the testing set out in the updated Test Directory, with regional variations</p>	1 and 4

			develops.	<p>monitored on a quarterly basis through performance data submitted to NHSE/I. (A subset of these data may be made publicly available, if clinically appropriate and in adherence with laws and data protection standards)</p> <p>All 7 NHS GLHs will have met the exit criteria for live clinical testing of whole genome sequencing (WGS).</p>	
4. Further develop the Genomics England clinical research interface - increase the	Genomics England	NHS Genomics Laboratory Hub able to report new diagnostic results when returned under Diagnostic Discovery	<p>Patients receive diagnoses</p> <p>Analysis pipeline gains new test control</p>	<p>Number of potential diagnoses returned to the NHS; aiming to return >100 diagnoses per year, the exact number will</p>	1 and 4

<p>number of diagnoses from genome data, and provide evidence to support the NHS Genomic Medicine Service in developing its diagnostic Test Directory.</p>		<p>process</p> <p>Researchers and clinicians communicate about complex results</p>	<p>samples</p> <p>Researchers and clinicians can collaborate on new discoveries</p>	<p>be publicly reported</p> <p>Number of papers published</p>	
<p>5. Pilot new approaches for patients with undiagnosed rare conditions</p>	<p>NHSE/I</p>	<p>Development of pilot approach(es) in April 2022</p> <p>Selection of sites during summer 2022</p>	<p>An evaluated approach to testing new approaches to diagnosing individuals with rare diseases</p> <p>Faster diagnosis and improved care for patients with undiagnosed conditions</p> <p>Design and outcomes of the pilots and</p>	<p>Number of clinics piloted</p>	<p>1</p>

			evaluations published, to help monitor impact and disseminate learnings		
6. Develop an innovative digital educational resource ('GeNotes') - providing healthcare professionals with relevant and concise information to support patient management, linking to the NHS Genomic Test Directories, and signposting to extended learning opportunities.	Health Education England	<p>Phase 1: User testing and publication of a minimal viable product including resources for paediatricians, oncologists and GPs. Completed April 2022.</p> <p>Phase 2: Scale up content production for other specialities. April 2022 onwards with input/co-development with the rare disease community and Genomics Education Programme Public Patient Involvement</p>	<p>A central locale that brings together relevant clinical/scientific information and signposts to applicable guidelines and other supplementary clinical and scientific information.</p> <p>A more informed NHS workforce.</p> <p>An educational platform that can integrate with NHS and other digital systems via a functional Application</p>	<p>Impact evaluation will form phase 3 of this action. Full details are still to be determined, but are likely to include:</p> <ul style="list-style-type: none"> - User experience (to include data from private and public beta phases) - Web analytics including API adoption, for instance how many NHS digital systems are 'talking with' and therefore using GeNotes content. - Impact metrics, for example self- 	2

		<p>(GEP PPI) group where appropriate.</p> <p>Phase 3: Evaluation of use and effectiveness of the resource. From October 2022.</p>	<p>Programming Interface (API), which allows two digital applications to talk to each other, thereby allowing NHS digital platforms to utilise content from GeNotes within their own system.</p>	<p>evaluation by users on impact to practice.</p>	
<p>7. Determine how best to include rare diseases in UK health professional education and training frameworks -</p> <p>to ensure rare disease competencies and learning outcomes are embedded in NHS education and training</p>	<p>Health Education England</p>	<p>Review current UK health professional education and training frameworks (to include curricula, proficiency standards, etc.) to determine rare disease content. The review process will take place over the 2022 to 2023 financial year.</p> <p>Identification of rare disease competency needed for each</p>	<p>Awareness of which training programmes lack sufficient rare disease content.</p> <p>Increased amount of rare disease content included in education frameworks (will require additional actions in 2023 to 2024).</p>	<p>Percentage of education frameworks that include sufficient rare disease content. Change measured by repeating the review over time.</p>	<p>2</p>

<p>frameworks across all relevant specialities including general practice training and those involved with emergency care.</p>		<p>education and training framework document, using existing generic curricula and competencies. Working with professional organisations, curriculum developers and other stakeholders (including patients/family members/carers identified through the GEP PPI group).</p> <p>Gap analysis to identify deficits in the inclusion of rare diseases in the frameworks.</p>			
<p>8. Extend the remit of the Genomics</p>	<p>Health Education</p>	<p>Established links and agreed way of working or</p>	<p>Joined-up approach within HEE in regard to</p>	<p>Establish links with fifty percent of relevant HEE</p>	<p>1 and 2</p>

<p>Education Programme</p> <p>to include non-genetic rare diseases</p>	<p>England</p>	<p>programme of work with key HEE programmes (for instance urgent and emergency care).</p> <p>Established and maintained links with integrated care systems (ICS).</p> <p>Rare disease education network to establish collaborations and share best practice. Membership of the network to include industry, third sector and research organisations, as well as other NHS organisations.</p>	<p>rare disease projects.</p> <p>Receive intelligence on workforce issues to inform workforce planning and new ways of working.</p> <p>Collaborative working and minimising duplication of effort.</p> <p>One-stop shop for clinicians to access rare disease education and training materials.</p>	<p>programmes, for example Innovation and Transformation, Technology Enhanced Learning and agree joint programmes of work.</p> <p>Percentage of ICSs in England with established links created through regional HEE networks and HEE transformation leads.</p> <p>Number of members signed up to the rare disease education network.</p> <p>Web analytics of rare disease hub and</p>
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		Published rare disease hub webpage and resource library.		resource library.	
9. Publish high-quality epidemiological and research papers to increase the understanding of rare diseases - including papers looking at basic rare disease epidemiology, impact of COVID-19 on people with some rare diseases and cancer-related risk factors or outcomes for people with some rare diseases.	NHS Digital	<p>NCARDRS will collaborate on and publish at least 6 papers describing novel findings or methods relevant to rare disease by the end of December 2022.</p> <p>NCARDRS will work with patient organisations to ensure visibility in their communities.</p> <p>Findings disseminated through presentations at conferences and other relevant</p>	Increased knowledge and raised profile of rare diseases through the publication of high-quality epidemiology and research.	Impact will be measured using Altmetric scores (a measure of the dissemination of a peer-reviewed paper via platforms like news outlets, social media, blogs).	2

		events/platforms.			
10. Develop a toolkit for virtual consultations - improving use of videoconference and telephone clinic calls in services for patients with complex, multi-system rare diseases.	NHSE/I	Toolkit published in spring 2022.	Shared learning resources Improved outcomes of virtual clinics for patients and families/carers Sharing of issues that clinical teams need to consider in planning and delivery of virtual clinics	Uptake of the toolkit within the Highly Specialised Portfolio will be monitored through annual clinical meetings. A survey will be developed for the services to determine whether patients and healthcare professionals found the toolkit helpful.	3
11. Support rapid access to drugs for patients with rare diseases in	NHSE/I	Map of programmes that promote access to drugs	Rapid access to clinically effective and cost-effective drugs for patients with rare	High percentage of drugs available at anticipated date of delivery	4

<p>the NHS - assessing the complexity of the service in which the drugs will be used, by mapping available access initiatives, identifying drugs and delivery challenges through horizon scanning, and listing drugs that have been identified for access.</p>	<p>Produce drug/service 'preparedness template'</p> <p>Commissioning process developed to support rapid access to drugs</p> <p>Report on whether or not there is access at the point of anticipated delivery, looking at: (a) if a drug is intended to be available on a certain date, is that the case, (b) is the uptake overall as expected and (c) is the geographical spread as expected</p>	<p>diseases</p> <p>Improved understanding by the system of requirements to enable rapid access to take place</p>	<p>Access initiatives mapped and narrative on progress provided</p> <p>Commissioning processes to support rapid access to drugs developed</p> <p>Ongoing list of relevant drugs developed and maintained</p> <p>Measurement taken of whether or not there is access at the point of anticipated delivery and why there may be</p>	
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		Monitoring of access via Blueteq data		deviation from this. Findings summarised in England Rare Diseases Framework Delivery Group minutes/ 2023 action plan	
12. Develop a strategic approach for gene therapies and other advanced therapy medicinal products (ATMPs) - based on horizon scanning by NHSE/I.	NHSE/I	An NHSE/I strategic approach for gene therapies	Clarity for the pharmaceutical industry, providers and patients around NHSE/I's strategic approach for gene therapies System prepared to deliver potential ATMPs if/when these are approved by NICE.	Development of a strategic approach by summer 2022, and narrative on progress provided	4

<p>13. Capitalise on the changes made to NICE's methods and processes to ensure that NICE continues to support the rapid adoption of effective new treatments for NHS patients with rare diseases - implementing NICE's new methods and processes to support access to new treatments for rare disease patients.</p>	<p>NICE</p>	<p>Replace the end-of-life criteria with a severity modifier</p> <p>Accept a greater degree of uncertainty when evidence generation is difficult, including rare diseases</p> <p>Adopting process changes to help improve participation of patients and clinical experts, introducing a summary of information for patients as well as more flexibility to adapt consultation timelines for each appraisal to support more efficient timely</p>	<p>More therapies made available to rare disease patients more rapidly</p> <p>Time efficient decisions made on topic routing</p>	<p>Number of medicines for rare diseases receiving a positive NICE recommendation</p>	<p>4</p>
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		access.			
		Adopt refined highly specialised topic routing criteria			
14. Monitor overall uptake of drugs for patients with rare diseases and map geographical access to those drugs	NHSE/I and NHS Digital	Standard operating procedure for undertaking systematic component of variation (NHSE/I) Agree data flows, deliverables and cross-organisation resources to support exemplar equity of access project(s) based on population-based, patient-level data drawn from high-cost medicines data.	Better understanding of the population uptake and impact of NHSE/I's high-cost drug commissioning policies and relevant NICE Technology Appraisals.	Annual report on update of drugs, including overall uptake and geographical equity using standard coefficient of variation. Plan of action agreed with any services where inequitable access identified. Evidence that this knowledge has informed future	4

				<p>commissioning policy wording / NICE guidance</p> <p>Findings summarised in England Rare Diseases Framework Delivery Group minutes/ 2023 action plan</p>	
15. Map the rare disease research landscape to identify gaps and priorities for future funding	DHSC and MRC	Publicly available paper describing the rare disease research landscape, gaps, priorities, and levers for change	<p>Improved understanding of the rare disease research landscape. Better understanding of gaps, priorities, and levers for change.</p> <p>A better joined-up rare disease research community.</p>	<p>Project plan fully defined by spring 2022</p> <p>Rare disease funding landscape mapped using existing databases by autumn 2022</p> <p>Workshops held on</p>	1, 2, 3, and 4

			Identification of opportunities for improved rare disease research addressing the priorities of the UK Rare Diseases Framework.	gaps and priorities by end 2022 Paper published early 2023	
16. Reduce Health Inequalities in NHS highly specialised services (HSS) - including considering health inequalities at HSS annual clinical meetings, in service development and commissioning decisions, and in provider selection processes.	NHSE/I	Discussion of health inequalities with all services – April 2022 to March 2023 Repeat of geographic access exercise – September 2023 Explore how consideration of health inequalities can be incorporated into future HSS procurements –	Increased understanding of health inequalities in highly specialised services resulting in more equal access and improvements for rare disease patients	Discussion about health inequalities at all HSS clinical meetings Paper about the outcomes of geographical access exercise to the NHS Rare Diseases Advisory Group A log of how health inequalities have been considered in	4

September 2023

procurements

Log of which Highly Specialised Commissioning Team have undertaken training

Appropriate metrics before and after the intervention will be recorded for each service

Findings summarised in England Rare Diseases Framework Delivery Group minutes/ 2023 action plan

Annex B: list of delivery partners

Department of Health & Social Care

The Department of Health & Social Care (DHSC) supports UK ministers in leading the nation's health and social care to help people live more independent, healthier lives for longer. Within DHSC the Rare Diseases Policy Team has worked alongside the Screening Policy and Research Capacity & Growth teams to develop specific actions for this first England Rare Diseases Action Plan.

Genomics England

Genomics England is a subsidiary limited company owned by the Department of Health & Social Care. It was established to deliver the 100,000 Genomes Project and is now working with the NHS to further develop and embed genomic healthcare and research in Britain. It works with patients, doctors, scientists, government and industry to improve genomic testing, and help researchers access the health data and technology they need to make new medical discoveries and create more effective, targeted medicines.

Health Education England

Health Education England (HEE) is part of the NHS and works with partners to plan, recruit, educate and train the NHS workforce. HEE provides national strategic leadership and policy development, national and international interventions, and tailored solutions regionally and locally.

Medical Research Council

The Medical Research Council (MRC) is part of UK Research and Innovation, a non-departmental public body sponsored by the Department for Business, Energy and Industrial Strategy (BEIS). MRC funds research at the forefront of science to prevent illness, develop therapies and improve human health. MRC support research across the biomedical spectrum, from fundamental laboratory-based science to clinical trials, and in all major disease areas.

Medicines and Healthcare products Regulatory Agency

The Medicines & Healthcare products Regulatory Agency (MHRA) safeguards public health in the UK through the licensing & enforcement of medicinal products for human use,

and enforcement of the laws relating to medical devices. The MHRA also regulates clinical trials of medicines & medical devices.

National Institute for Health and Care Excellence

The National Institute for Health and Care Excellence's (NICE) role is to improve outcomes for people using the NHS and other public health and social care services. It supports practitioners, managers, and commissioners by producing evidence-based guidance and advice; developing quality standards and performance; and providing a range of information services.

National Institute for Health Research

The NIHR is the largest funder of health and care research in England, spending over £1 billion from the Department of Health and Social Care on research every year. Its mission is to improve the health and wealth of the nation through research. It provides the people, facilities and technology that enable research to thrive and influence policy and practice. It also provides the research infrastructure and training to underpin the work of other research funders, research charities and industry.

NHS England and NHS Improvement

NHS England leads the NHS, sets strategic direction for the NHS through the NHS Long Term Plan, and funds key priorities for improvement. NHS Improvement is responsible for overseeing and supporting NHS foundation trusts, NHS trusts and some independent providers to ensure patients receive consistently safe, high quality, and responsive care within local integrated care systems. Since 1 April 2019 NHS England and NHS Improvement have come together as a single organisation.

NHS Digital

NHS Digital is the national information and technology partner for the health and care system. Its systems and information help doctors, nurses and other health care professionals improve efficiency and make care safer. It provides information and data to the health service; creates and maintains the technological infrastructure that keeps the health service running; and develops information standards that improve the way different parts of the system communicate.

NHS Digital is responsible for the management of the National Disease Registration Service (NDRS), which consists of the National Congenital Anomaly and Rare Disease

Registration Service and the National Cancer Registration and Analysis Service. NHS Digital operates the NDRS under the [National Disease Registries Directions \(2021\)](#), which provide it with legal permission to collect patient data to use it to protect the health of the population.

Annex C: list of engagement activities

UK Rare Diseases Forum meetings

The UK Rare Diseases Forum holds formal, twice yearly meetings with a core membership to discuss papers and updates released ahead of the UK Rare Diseases Framework Board meetings. These meetings are chaired by Alastair Kent OBE – previously co-chair of the UK Rare Disease Policy Board and previously Chief Executive of Genetic Alliance UK. Membership includes senior representatives with experience in the rare diseases field including patient organisations, industry representatives, research-active bodies, clinical representatives, and digital and data representatives. A full [list of organisations represented is published online](#).

Meetings are for UK-wide discussion but also feature nation-specific updates. Forum meetings have offered the opportunity for feedback on early-stage actions for the England Rare Diseases Action Plan, and on a late-stage draft of the document.

UK Rare Diseases Forum online platform

The UK Rare Diseases Forum online platform provides continuous engagement with a wide range of stakeholders around the delivery of the UK Rare Diseases Framework.

The platform hosts an active discussion space for community members and a repository for the upload of governance structure papers and meeting minutes. It also hosts a community newsletter providing national policy updates, news items, and upcoming events. A summary of the activity on the online platform is presented for discussion at meetings of the UK Rare Diseases Framework Board, UK Rare Diseases Forum, and England Rare Diseases Framework Delivery Group.

Community roundtables

Two community roundtables were held to seek community input on the action plan in a smaller group context.

The first roundtable was held on 11th October 2021. Participants were recruited in partnership with Genetic Alliance UK, with a particular emphasis on including those whose voices are seldom heard. The roundtable provided an opportunity for delivery partners and members of the rare diseases community to discuss the draft actions intended to form the basis of the England Rare Diseases Action Plan. Delivery partners presented their draft

actions, fielded questions, and gathered feedback. Participants also discussed the overall coverage of framework priorities and suggestions for further work.

The second roundtable was held on 16th December 2021 and brought together those with lived experience of rare disease as well as a range of healthcare professionals (including clinicians, nurses and genetic counsellors). Participants provided feedback on the first draft of the England Rare Diseases Action Plan, with discussion on each of the framework priorities and underpinning themes.

England Rare Diseases Action Plan questionnaire

In November 2021, a targeted online questionnaire was launched to gather detailed feedback on draft actions from people and organisations across the rare disease community. The questionnaire was open for three weeks and received 92 responses from people living with a rare disease, their carers, family members, charities for patients with rare diseases, healthcare professionals, industry partners and researchers. The questionnaire asked respondents questions on each of the draft actions, gauging whether the community felt it a) would contribute towards implementing the UK Rare Diseases Framework and b) was sufficiently ambitious. The questionnaire also asked whether the collection of actions listed under each priority addressed that priority as a whole. Respondents were able to provide free text comments on each of the actions and priorities.

Health inequalities workshop

A community workshop was held to focus specifically on health inequalities experienced by people from diverse and marginalised communities who are also affected by a rare condition. This was hosted in partnership with Breaking Down Barriers, a network of over 50 organisations working together to improve the lives of families from diverse and marginalised communities. Participants shared their lived experiences on issues relating to each of the framework priorities, with discussion questions including “what are your experiences of receiving or waiting for a diagnosis?”, “how much do you feel healthcare professionals understand about you and your condition?”, “what is your experience of care coordination?” and “is access to specialist care, treatments and drugs equitable?”. Participants further discussed the larger question of how to create a fairer system which meets the needs of a diverse community.

Annex D: progress to date

Priority 1: helping patients get a final diagnosis faster

Clinical diagnosis of rare diseases

The NHS Genomic Medicine Service (GMS) was launched in England in October 2018, to ensure the NHS has the infrastructure required to provide access to a high-quality, equitable genomic medicine service. The NHS GMS carries out over 600,000 genomic tests in England every year for common and rare diseases, inherited diseases, pharmacogenomics, and cancer. The testing that is available is set out in the [National Genomic Test Directory](#), from single gene tests to whole genome sequencing, and currently includes 357 rare disease clinical indications.

In December 2020, 7 NHS Genomic Medicine Service Alliances were established to support the mainstreaming and embedding of genomic medicine. In addition to their core work, each NHS GMS Alliance has also led a national transformation project in 2021/22, several of which impact on rare disease research and care. Over the course of 2021, each NHS GMS Alliance has also undertaken several local transformation projects focused on patient needs in their particular geography. For example, the North West GMS Alliance are undertaking a project evaluating a new genomic testing technology to better diagnose and profile rare diseases. This focus on transformation activity will continue in 2022/23.

In November 2020, the first NHS Genomic Laboratory Hub (GLH) started clinical testing of whole genome sequencing (WGS). All 7 NHS GLHs are now involved in live clinical testing of WGS, and significant progress has been made with overall samples increasing month on month. The capacity of the service is being continually expanded, with new rare disease conditions becoming eligible for whole genome sequencing, and new clinical indications beginning to be implemented across all 7 NHS Genomic Laboratory Hubs.

Rare disease research

A [significant new scientific study, published in the New England Journal of Medicine in November 2021](#), showed that whole genome sequencing can uncover new diagnoses for people across the broadest range of rare diseases investigated to date and could deliver enormous benefits across the NHS. The pilot study of rare undiagnosed diseases involved analysing the genes of 4,660 people from 2,183 families – all of whom were early participants in the 100,000 Genomes Project. The study, led by Genomics England and Queen Mary University of London, and supported by the National Institute for Health Research (NIHR) BioResource, found that using WGS led to a new diagnosis for 25% of the participants. Of these new diagnoses, 14% were found in regions of the genome that

would be missed by other conventional methods, including other types of non-whole genomic tests. Many of the participants had gone through years of appointments, without getting any answers. By having their whole genome sequenced, diagnoses were uncovered that would not have previously been detectable allowing patients to receive more focused clinical care.

Another [scientific study published in November 2021](#) similarly demonstrated the power of whole genome sequencing to provide a diagnosis to people living with a rare disease, looking specifically at those with a suspected mitochondrial disease. Published in The BMJ, researchers investigated the genomes of 345 patients with an unexplained multisystem progressive disorder who were participants in the 100,000 Genomes Project, and had already undergone extensive investigations for mitochondrial disease. A definite or probable genetic diagnosis was identified in 31% of families involved in the study, of which some allowed for improved clinical decision making or specific treatment pathways. Importantly, most of the new diagnoses (63%) were non-mitochondrial, meaning they would likely have been missed if a targeted approach, rather than whole genome sequencing, was taken.

The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS), in partnership with the patient charity Histio UK, academics and clinicians, published a [paper describing a robust method for identifying people diagnosed with the rare diseases haemophagocytic lymphohistiocytosis \(HLH\)](#) from routinely collected data. This means that England now has the largest population-based cohort of people with HLH globally, and further work can be done to achieve a greater understanding of this rare and often fatal disease, including risk factors that lead to its onset. It also means that this method may be suitable for modification to support work on other rare diseases.

Newborn screening for rare disease

The diagnosis of rare disease through the 100,000 Genomes Project and the NHS GMS uses whole genome sequencing to improve the diagnostics of patients who are already ill or symptomatic. One of the priorities outlined in the UK strategy for genomics – [Genome UK](#) – is to explore the potential of preventing disease through the expansion of genomic screening in early life. It has been estimated that over 3,000 babies a year could benefit from lifesaving or life-changing interventions if whole genome sequencing is used to support diagnosis. By identifying such diseases at the earliest possible stage, clinicians have the greatest chance to treat before the disease can cause significant harm, improving the likelihood that the child will survive with a higher quality of life over the long-term.

A national dialogue was performed to explore public views on the implications of whole genome sequencing for newborn screening in 2021. The dialogue, commissioned by the UK National Screening Committee, NHS England and NHS Improvement (NHSE/I), and

Genomics England, involved over 130 members of the public from across the UK with a range of backgrounds, including individuals with genetic conditions, new and expectant parents, ethnic minorities, and young adults. The [report was published](#) in July 2021 and found that the public was broadly supportive, providing appropriate safeguards and resources are in place.

Priority 2: increasing awareness of rare diseases among healthcare professionals

Upskilling the NHS workforce

Over the course of 2021, Health Education England's (HEE) Genomics Education Programme has continued to develop bespoke educational resources to underpin the implementation of the NHS GMS. This includes two competency frameworks outlining the expected knowledge, skills, and behaviours NHS staff need to facilitate genomic testing and the return of genomic test results; a suite of 5 online courses to underpin these competencies; and a genomics advisors competency framework to support the development of genomic expertise at local, regional, and national level. HEE has also continued to raise awareness of rare diseases through the [Genomics Education Programme's weekly blog](#), with more than 50% of the weekly blogs published in 2021 directly related to rare conditions.

Understanding the impact of the COVID-19 pandemic on people with rare diseases

In collaboration with academic partners, NCARDRS [published whole-population studies](#) into the impact of the COVID-19 pandemic on people with a range of rare, late onset non-genetic conditions (autoimmune rheumatic diseases) highlighting their increased risks of COVID-19 infection, hospitalisation and death compared to the general population. These results have informed policy and research initiatives to address the ongoing risks of severe outcome from COVID-19 in people who might remain vulnerable despite vaccination. It has also led to pioneering rare disease research with the launch of the MRC funded [MELODY study](#) (Mass evaluation of lateral flow immunoassays for the detection of SARS-CoV-2 antibody responses in immunosuppressed people), which is investigating vulnerability to COVID-19 infection following vaccination in a large cohort of people with rare diseases.

Priority 3: better coordination of care

Research to support coordination of care

In the UK, coordinated care for patients with rare conditions is provided in different ways. Some people are cared for through specialist centres, some have care coordinators, multi-disciplinary teams, care plans and/or residential clinics where patients can access a range of services during one hospital visit. For others care is not well coordinated and the burden of coordinating care can fall on the individual or their families. To improve coordination of care it is vital to understand patient experiences and what current and best practice look like.

To this end, NIHR funded the [Coordinated Care Of Rare Diseases](#) (CONCORD) study to investigate how services for people with rare diseases are coordinated in the UK, and how people living with rare diseases, and healthcare professionals who treat rare diseases, would like them to be coordinated.

Following an extensive literature review and consultation with the rare diseases community, [the study published a landmark definition of coordination of care in rare diseases](#):

coordination of care involves working together across multiple components and processes of care to enable everyone involved in a patient's care [...] to avoid duplication and achieve shared outcomes, throughout a person's whole life, across all parts of the health and care system...

coordination needs to be ...family-centred, holistic (including a patient's medical, psychosocial, educational and vocational needs), evidence-based, with equal access to coordinated care irrespective of diagnosis, patient circumstances and geographical location.

CONCORD also explored the extent and impact of care coordination (or lack thereof), through extensive interviews with individuals affected by rare conditions (including undiagnosed conditions), and family members who had a caring role. [The overall findings, published in 2021](#), emphasised the importance of flexible care, which is capable of meeting patients'/carers' individual needs throughout their rare disease journey. The study listed a number of recommendations from participants, including being able to access professional support in coordinating care; adapting the location, scheduling, and services available at clinics and appointments; and improving communication using technology, care plans, accessible contact points, and multi-disciplinary team working. The findings showed that care coordination for people with rare conditions could be improved, while recognising that there are additional challenges which can limit coordination, including

difficulties diagnosing rare disease and the small numbers of people diagnosed with each individual condition.

Building on these findings, CONCORD has produced a [taxonomy of care coordination](#), outlining different ways in which care can be coordinated for people living with rare conditions, as well as developing [hypothetical models of care coordination](#), exploring which types of coordination may be appropriate in different situations (including those with a specialist centre and those without).

Priority 4: improving access to specialist care, treatment, and drugs

Rare disease registration

NCARDRS records those people with congenital abnormalities and rare diseases across the whole of England. As a result, NCARDRS can provide data to support high quality clinical practice, empower patients and carers and monitor the frequency and distribution of conditions. It can also support research, inform planning and evaluation of health and social care services, including screening. NCARDRS continues to make improvements to its service and published its [congenital anomaly statistics 2019 report](#) in September 2021.

Also in 2021, NHSE/I put in place an arrangement to share high-cost medicines data with NCARDRS. This will help determine if there is equitable access to drugs for patients with rare diseases and can support future NHSE/I commissioning decisions. This agreement is already in place for cancer indications to support the National Cancer Registration and Analysis Service (NCRAS) and has proved to be exceptionally efficient and valuable.

Patient-centred research

In March 2021 we published [Saving and Improving Lives: The Future of UK Clinical Research Delivery](#), setting out our ambition to create a patient-centred, pro-innovation, and digitally enabled clinical research environment. Implementing this vision will unleash the potential of our clinical research environment to improve health, capitalise on our renowned research expertise, and make the UK one of the best places in the world to design and deliver research.

The vision will be delivered through the cross-sector Recovery, Resilience and Growth Programme and with the full involvement of stakeholders, including patients and the public, the NHS, industry, medical research charities, academia, research regulators, research funders including the National Institute for Health Research (NIHR), and the Devolved Administrations. A [Phase 1 Implementation Plan](#) was published in June 2021, setting out what we will deliver during financial year 2021 to 2022, and work is ongoing to

inform the development of the Phase 2 Implementation Plan, including the particular needs for rare disease clinical research delivery.

Access to the most effective treatments available

The National Institute for Health and Care Excellence (NICE) is now among the fastest health technology assessment bodies in the world. Despite the pressures of COVID-19, the average time from marketing authorisation to first NICE output in 2020/21 was 4 months for non-cancer topics and less than 1 month for cancer topics.

NICE recently concluded a review of its methods and processes for health technology evaluation. The changes made through the review will ensure NICE's methods remain world leading and that they are suited to new and emerging types of technology, and will make its processes fairer, faster and more consistent. NICE is now implementing a number of changes that will benefit rare disease patients (see [action 13](#)).

Annex E: rare disease collaborative networks

Rare disease collaborative networks (RDCNs) are an important part of NHS England and NHS Improvement’s (NHSE/I) provision to support patients with rare diseases. RDCNs are made up of providers (rare disease collaborative centres) who have an interest in a particular rare disease and are committed to working together to progress research, increase knowledge and improve patient experience and outcomes. Examples of priority areas of existing RDCNs include:

- raising awareness of the rare disease
- improving co-ordination of care
- sharing of expertise and best practice
- establishing a disease registry to improve the understanding of the epidemiology of the rare disease
- research on treatment options and diagnostics
- establishing a support network for patients and families, including co-ordinated transition from paediatric to adult services

Thirteen RDCNs have been established to date across a range of specialties and disease groups:

Table 2. List of current rare disease collaborative networks

Rare disease collaborative network	Designated provider(s)	Description
CDKL5	University Hospitals Bristol and Weston NHS Foundation Trust	CDKL5 is a rare genetic neurodevelopmental condition which affects children. It is characterised by seizures and developmental challenges.
National Refractory	Lead: Sheffield Teaching Hospitals	Refractory Coeliac Disease is a complex autoimmune disorder which

Rare disease collaborative network	Designated provider(s)	Description
Coeliac Disease Network	NHS Foundation Trust Royal Brompton Hospital (Guy's & St Thomas' NHS Foundation Trust)	predominantly affects adults. It is characterised by persistence or recurrence of symptoms despite a gluten free diet and is associated with an increased risk of lymphoma.
National Rare Cystic Lung Disease	Lead: Royal Brompton Hospital (Guy's & St Thomas' NHS Foundation Trust) Nottingham University Hospitals NHS Trust	Cystic lung disease is an umbrella term for a group of conditions which present with multiple lung cysts. Although each condition can present similarly to another, management differs between each type.
Hereditary Haemorrhagic Telangiectasia (HHT)	Lead: Imperial College Healthcare NHS Trust Bronglais District General Hospital, Aberystwyth King's College Hospital NHS Foundation Trust Royal Free London NHS Foundation Trust Western General Hospital, Edinburgh, NHS Lothian	HHT is a genetic disorder which affects the development of the blood vessels and presents at any age. It commonly first presents with bleeding but causes blood vessels to develop abnormally and can result in widespread complications.
Congenital Thoracic Malformations Network	Royal Brompton Hospital (Guy's & St Thomas' NHS Foundation Trust)	Congenital thoracic malformations are a broad group of abnormalities that result in underdevelopment of the chest or lungs. The abnormalities are often first identified before birth.
Familial	Lead: Cambridge University Hospitals	A genetic condition resulting in air accumulating within the chest but

Rare disease collaborative network	Designated provider(s)	Description
Pneumothorax	<p>NHS Foundation Trust</p> <p>Queen Elizabeth University Hospital, NHS Greater Glasgow & Clyde</p>	<p>outside of the lungs which impacts in breathing. It can affect all ages.</p>
Hereditary gastrointestinal polyposis syndromes	<p>Lead: St Mark's Hospital (London North West University Healthcare NHS Trust)</p> <p>University Hospitals Birmingham NHS Foundation Trust</p> <p>Manchester University NHS Foundation Trust</p> <p>Royal Infirmary of Edinburgh, NHS Lothian</p>	<p>A genetic condition resulting in the form of gastrointestinal polyps, often in children and adolescents, leading to a high risk of intestinal cancer.</p>
UK Hyperoxaluria RDCN	<p>Lead: Royal Free London NHS Foundation Trust</p> <p>Birmingham Women's and Children's NHS Foundation Trust</p> <p>Great Ormond Street Hospital for Children NHS Foundation Trust</p> <p>University Hospitals</p>	<p>Hyperoxalurias are a group of diseases (genetic and acquired) which impair the ability of the body to remove oxalate. Accumulation of oxalate can result in a number of complications which include kidney stones, bone fractures, reduced growth and anaemia.</p>

Rare disease collaborative network	Designated provider(s)	Description
	Birmingham NHS Foundation Trust	
Juvenile Myasthenia Gravis network	Oxford University Hospitals NHS Foundation Trust	Juvenile Myasthenia Gravis (JMG) is a rare disorder with an estimated incidence of 1.5/million person years in the UK. JMG is defined as MG in patients 18 years and younger but 20% of cases are under 10 when diagnosed.
Segmental overgrowth and vascular malformations	<p>Lead: Nottingham University Hospitals NHS Trust</p> <p>United Hospitals of Derby and Burton NHS Foundation Trust,</p> <p>St George's University Hospital NHS Foundation Trust</p> <p>Birmingham Women's and Children's NHS Foundation Trust</p> <p>Manchester University NHS Foundation Trust</p> <p>Royal Free London NHS Foundation Trust</p>	Segmental overgrowth and vascular malformations are group of rare disorders characterised by asymmetric overgrowth of tissues.
Paroxysmal motor disorders	St George's University Hospitals NHS	Paroxysmal motor disorders (PMDs) include a number of very rare

Rare disease collaborative network	Designated provider(s)	Description
RDCN	Foundation Trust	neurological diseases that affect the ability to move. Nearly all of the PMDs are rare genetic conditions.
Paediatric and primary lymphoedema	<p>Lead: St George's University Hospitals NHS Foundation Trust</p> <p>Nottingham University Hospitals NHS Trust</p> <p>United Hospitals of Derby and Burton NHS Foundation Trust</p>	<p>Patients with primary lymphoedema have developmental or functional anomalies of the lymphatic system. They may present with lymphoedema affecting one or multiple body segments or associated with problems affecting other organs or systems.</p>
Mosaic Disorders	Great Ormond Street Hospital for Children NHS Foundation Trust	Mosaic disorders are a group of rare disorders which present with extensive birthmarks and are caused by a genetic mutation which affects the developing baby during pregnancy.

Annex F: NICE support to the life sciences industry and health system

The National Institute for Health and Care Excellence (NICE) provides a variety of support to the life sciences industry and the health system to help enable and speed up the opportunity of securing access to new treatments. It helps companies at all stages of product development and commercialisation to get expert advice which helps to get new and innovative products adopted quickly. These include:

- standard/express scientific advice, a service which helps companies developing technologies that target rare conditions with their clinical and economic evidence generation strategy
- Preliminary Independent Model Advice (PRIMA) which provides expert independent advice to improve the quality of health economic models for companies working in rare conditions
- Medtech Early Assessment (META) Tool/Medtech Advice is a gap analysis/advice service for developers of medical devices, diagnostics and digital health technologies (including those that target rare conditions)
- the NICE Office for Market Access (OMA) which provides support with engagement between life science companies and the health system to help companies to make informed decisions on how to get products to patients faster. This enables the NHS to adopt drugs, devices and diagnostics quickly and foster innovation and growth in the UK's life science industries

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