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Department of Health & Social Care

Policy paper

England Rare Diseases Action Plan 2023: annexes

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Applies to England

Contents

This publication is available at https://www.gov.uk/government/publications/england-rare-diseases-action-plan-2023/england-rare-diseases-action-plan-2023-annexes

Annex A: progress against actions in England’s 2022 Rare Diseases Action Plan

Priority 1: helping patients get a final diagnosis faster

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

Owner: DHSC

Original 2022 outputs:

- having already completed stakeholder engagement, a new UK NSC terms of reference and remit
- UK NSC Bloodspot Task Group terms of reference
- UK NSC Bloodspot Task Group products:
- baseline comparison between UK and EURORDIS key principles for newborn screening
- manuscript advising on the methodological principles for screening test accuracy study designs in rare disease settings
- paper on technical and procedural considerations for modelling exercises around newborn screening
- paper on use of registries in the newborn screening evaluation process

Progress report:

- the UK NSC terms of reference and new remit was established in 2022
- terms of reference established for the UK NSC Bloodspot Task Group in 2022
- draft manuscript on baseline comparison between UK and EURORDIS discussed at November 2022 meeting of Bloodspot Task Group
- report on the methodological principles for screening test accuracy is in progress
- commissioning briefs are in progress for research into technical and procedural considerations for modelling exercises, and use of registries

Action 1 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- the UK NSC will continue work to improve the evidence available to them in evidence reviews
- draft manuscript on baseline comparison between UK and EURORDIS completed by spring/summer 2023

- draft report on methodological principles for screening test accuracy prepared for discussion with UK NSC and Netherlands Health Council in spring/summer 2023
- commissioning briefs ready for procurement in 2023

Action 2: whole genome sequencing (WGS) to screen for genetic conditions in healthy newborns – designing an ethically approved research study

Owner: Genomics England, NHSE

Original 2022 outputs:

- research study ready to implement at a small number of NHS trusts
- results pathway mapped for return of findings to NHS clinicians and 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 finalised and available

Progress report:

- research study due to begin during 2023 – all necessary systems, processes, education and training will be embedded before recruitment commences
- more than 150 clinicians have been engaged in preparation for mapping of results and healthcare pathways – this will be completed during 2023
- four principles for gene selection have been finalised following an extensive engagement process, including an online survey that received over 400 responses. Assessment of genes against these principles is due to be completed in mid 2023
- in 2022, data from 14,785 genomes from the National Genomic Research Library (NGRL) was used to model and test different approaches to the technical details or the analysis pipeline. Consultation on the outputs of this modelling with NHS and other genomics experts is currently underway

Action 2 status (ongoing, extended or concluded): extended

Metrics and milestones for 2023:

- optimal method for taking samples from newborns for the purposes of WGS
- data from the Baby and Mum Samples Study analysed and reported through Genomics England website (results anticipated in the first half of 2023)
- a list of genes and variants to be included in the screening panel, finalised and published over the next year

- participant information materials and website developed with approval by a Research Ethics Committee
- training for healthcare professionals involved in the study with training delivered at at least 3 NHS sites
- Research Ethics Committee-approved research study rolled out in a small number of NHS trusts
- evaluation strategy developed with processes established to capture data that informs the value of the programme, including with respect to health economic outcomes

Action 3: continuously develop the National Genomic Test Directory, including rollout of WGS, which will play an important role in diagnosis of rare diseases.

Owner: NHSE

Original 2022 outputs:

- phase 2 and 3 clinical indications for whole genome sequencing will be launched in 2021 to 2022
- an annual process for updating the Test Directory will have been implemented in Q4 2021 to 2022

Progress report:

- the Test Directory is regularly updated to reflect scientific and technological developments, including new clinical indications for rare disease and WGS
- in October 2022, testing for the rare lung condition Pulmonary Alveolar Microlithiasis was added to the directory
- 31 rare conditions are now included within the eligibility criteria for WGS, meaning access for more patients

Action 3 status (ongoing, extended or concluded): concluded

Metrics and milestones for 2023:

- there will be future updates to the Test Directory

Action 4: further develop the Genomics England clinical research interface – increase the number of diagnoses from genome data, and provide evidence to support the NHS Genomic Medicine Service in developing its diagnostic Test Directory

Owner: Genomics England

Original 2022 outputs:

- NHS Genomics Laboratory Hub able to report new diagnostic results when returned under diagnostic discovery process
- researchers and clinicians communicate about complex results

Progress report:

- 1000 diagnoses have been returned to the NHS during 2022, against an original target of 100

Action 4 status (ongoing, extended or concluded): concluded

Metrics and milestones for 2023:

- the clinical research interface is now embedded within Genomics England and within the NHS Genomic Medicine Service. Diagnoses continue to be made and returned to the NHS on a monthly basis for participants of the NGRL
- it is anticipated that numbers will continue to go up year on year as the number of participants and researchers within the NGRL increases

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

Owner: NHSE

Original 2022 outputs:

- development of pilot approach(es) in April 2022
- selection of sites during summer 2022

Progress report:

- the syndrome without a name (SWAN) pilot has been developed, aiming to reduce time to diagnosis for patients with undiagnosed rare diseases
- the proposed model covers all ages and aims to provide good geographical coverage

Action 5 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- the model is being discussed through NHSE governance and finance structures. If relevant funding is agreed, the SWAN pilot will be implemented in 2023

Priority 2: increasing awareness of rare diseases among healthcare professionals

Action 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

Owner: Health Education England

Original 2022 outputs:

- phase 1: user testing and publication of a minimal viable product, including resources for paediatricians, oncologists and GPs, completed by April 2022
- phase 2: scale up content production for other specialities from April 2022 onwards, with input and co-development with the rare disease community and GEP Public Patient Involvement (PPI) group, where appropriate
- phase 3: evaluation of use and effectiveness of the resource from October 2022

Progress report:

- private beta phase testing for oncology, paediatrics and general practice has been successfully completed
- GeNotes scored 91 out of 100 on the system usability scale assessment, with 93% of testers likely or very likely to use GeNotes in future
- the public beta platform for GeNotes was launched in June 2022 with oncology, and has achieved more than 36,800 page views and over 15,000 users as at 30 January 2023
- expansion of the Genomics Education Programme continues to support further and broader content created, and a Patient Advisory Group (Patient Advisors for Genomic Education – PAGE) has been established
- continued evaluation for the use and effectiveness of the resource is ongoing

Action 6 status (ongoing, extended or concluded): extended

Metrics and milestones for 2023:

- evaluation of resources will continue through 2023. Upcoming improvements include:
- advanced search and better filtering
- improved ‘In the Clinic’ specialty landing pages
- enhancements to Knowledge Hub
- next phase: pilot to syndicate GeNotes to primary care systems and exploring demand for a GeNotes app
- further specialities are planned to launch in spring/summer 2023, including: foetal and women’s health, cardiology, primary care, pharmacogenomics, paediatrics and endocrinology
- scale up content production for specialities from April 2023 onwards and resource being allocated to new specialities, for example neurology,

nephrology and haemato-oncology

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

Owner: Health Education England

Original 2022 outputs:

- review current UK health professional education and training frameworks (to include curricula, proficiency standards and so on) to determine rare disease content. The review process will take place over the 2022 to 2023 financial year
- identification of rare disease competency needed for each education and training framework document, using existing generic curricula and competencies, and working with professional organisations, curriculum developers and other stakeholders (including patients, family members or carers identified through the GEP PPI group)
- gap analysis to identify deficits in the inclusion of rare diseases in the frameworks

Progress report:

- since the publication of the 2022 Action Plan, HEE has adapted its approach to the development of healthcare competencies, resulting in a change to how it evaluates the impact and measures the inclusion of genomics and rare disease in healthcare workforce education and training
- HEE continues to work collaboratively with the Academy of Royal Colleges to evaluate medical curricula and ensure an increase in genomic and rare disease content. Whilst successful (particularly in paediatric, obstetric and oncology curricula), this has not been quantified and recorded as a percentage rise
- HEE’s current (additional) approach to rare disease competency frameworks is 3-fold:
- to develop role- or profession-specific frameworks through the introduction of the clinical pathway initiative
- competency frameworks for the leaders of genomic medicine (genomic advisors)
- nursing- and midwifery-specific competency frameworks
- HEE is evaluating uptake, acceptability and efficacy using a case study method using quantitative scoring and qualitative thematic analysis
- through its clinical pathway initiative, HEE is identifying patient touchpoints, competencies required and education and training interventions, to establish gaps in its frameworks

Action 7 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- the GEP will undertake a desktop review of educational curricula, standards of proficiency and frameworks with a view to incorporating teaching opportunities using rare disease examples (for example, the inclusion of genomics in the Nursing and Midwifery Council standards with underpinning teaching materials developed by the GEP included in the nurse educator toolkit)
- there will be ongoing qualitative and quantitative monitoring and evaluation of the Rare Disease massive open online course (MOOC)
- the GEP will also continually monitor the content of the course to ensure it is up to date with the ever-evolving genomics environment
- the PAGE will meet twice a year, and actions and impact will be measured against the group's objectives
- the GEP will develop a further 2 to 5 clinical pathway initiatives (CPIs) to facilitate the integration of genomic medicine through the alignment of patient pathways, workforce development and education and/or training requirements, relating to rare diseases
- based on the findings of the CPI competencies and gap analysis, the GEP will also develop further tier 1 and 2 GeNotes resources relating to rare disease

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

Owner: Health Education England

Original 2022 outputs:

- established links and agreed way of working or programme of work with key HEE programmes (for instance, urgent and emergency care)
- established and maintained links with ICSs
- rare disease education network to establish collaborations and share best practice, with membership including industry, third sector and research organisations, as well as other NHS organisations
- published rare disease hub webpage and resource library

Progress report:

- HEE has established a network of HEE Regional Genomics leads, who are linking into the Genomic Laboratory Hubs and Genomic Medicine Service Alliances system through connection with regional education and training leads
- HEE has established links with other HEE departments, NHS England Genomics Unit, Royal Colleges
- HEE is working in partnership with the Association of the British Pharmaceutical Industry (ABPI) who will be supporting and providing expert input, where relevant, into the development of GeNotes and other GEP resource

Action 8 status (ongoing, extended or concluded): extended

Metrics and milestones for 2023:

- the GEP and ABPI will work together to incorporate greater understanding of future advances in medicines, including rare disease
- the HEE Regional Genomic leads will continue to meet 6 times a year to ensure connection to regional education and training leads
- ‘month of genomics’ activity held in collaboration with the Royal College of Obstetricians and Gynaecologists (RCOG), Royal College of Paediatrics and Child Health (RCPCH) and Royal College of Midwives (RCM) to explore genetic and non-genetic rare diseases
- the GEP will partner with Medics for Rare Diseases (M4RD) to enhance the development of networks and resources for the rare disease hub, including those relating to non-genetic rare diseases

Action 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

Owner: NHS Digital

Original 2022 outputs:

- 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
- NCARDRS will work with patient organisations to ensure visibility in their communities
- findings disseminated through presentations at conferences and other relevant events and platforms

Progress report:

- NCARDRS has worked in partnership to contribute to 10 papers on non-cancer rare disease in 2022, which have been published, accepted for publication or are under peer review
- findings have been conveyed to patient groups, including Rare Autoimmune Rheumatic Diseases Alliance (RAIRDA), Wilson Disease Support Group and the Lily Foundation; and shared on the UK Rare Diseases Forum
- findings have been presented at national and international conferences and events, including the 4th American Society of Haematology Annual Meeting, the Royal College of Physicians annual conference, the International Clearinghouse for Birth Defects Surveillance and Research Conference, and the Mitochondrial Medicine Conference

- in December 2022, a stakeholder event was held with DHSC on opportunities and challenges of rare disease registration

Action 9 status (ongoing, extended or concluded): concluded

Metrics and milestones for 2023:

- NCARDRS will report on the Almetric scores on the 2022 papers in 2023 (where appropriate) as part of ongoing work under action 22 (see annex B)

Priority 3: better coordination of care

Action 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

Owner: NHSE

Original 2022 outputs:

- toolkit published in spring 2022

Progress report:

- toolkit has been developed and is awaiting publication

Action 10 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- NHS England will make its toolkit available to all highly specialised services clinical leads

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

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

Owner: NHSE

Original 2022 outputs:

- map of programmes that promote access to drugs

- produce drug and service ‘preparedness template’
- commissioning process developed to support rapid access to drugs
- report on whether or not there is access at the point of anticipated delivery, looking at:
- if a drug is intended to be available on a certain date, is that the case?
- is the uptake overall as expected?
- is the geographical spread as expected?
- monitoring of access through Blueteq data

Progress report:

- a map of programmes has been developed
- NHS England is actively using the latest version of the ‘preparedness template’ with companies

Action 11 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- NHS England will produce an annual report by the end of the 2022 to 2023 financial year on access to drugs for patients with rare diseases at the point of delivery. This report will also include an analysis of overall uptake and geographical equity for those drugs recommended in the NICE Highly Specialised Technology Programme
- NHS England is actively working with pharmaceutical companies and providers to assess the commissioning model needed for drugs to be delivered, so that access can be provided at the point of delivery

Action 12: develop a strategic approach for gene therapies and other advanced therapy medicinal products (ATMPs), based on horizon scanning by NHSE

Owner: NHSE

Original 2022 outputs:

- an NHSE and NHSI strategic approach for gene therapies

Progress report:

- development of the strategic approach has begun but is not yet finalised
- NHSE has engaged with stakeholders to establish a structured approach to commissioning for ATMPs
- using horizon scanning, NHSE has also instigated structured proactive engagement with individual ATMP manufacturers 12 to 24 months prior to marketing authorisation

Action 12 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- development and implementation of the strategic approach will continue

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, implementing NICE's new methods and processes to support access to new treatments for rare disease patients

Owner: NICE

Original 2022 outputs:

- replace the end-of-life criteria with a severity modifier
- accept a greater degree of uncertainty when evidence generation is difficult, including rare diseases
- 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
- adopt refined highly specialised topic routing criteria

Progress report:

- end-of-life criteria was replaced with a severity modifier in a methods update from February 2022
- NICE accepts a higher degree of uncertainty when evidence generation is difficult, including rare diseases in a methods update in February 2022
- NICE has introduced the use of the summary of information for patients (SIP) – a form developed by NICE which companies complete to help support individual patient experts participate in NICE technology appraisals
- a refined highly specialised topic routing criteria was introduced in February 2022

Action 13 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- all actions have been implemented as business as usual for all new treatments starting evaluations with NICE
- due to length of the medicines evaluation process and number of rare disease topics using older methods or processes, the analysis of the impact of changes (for example, percentage of positive NICE recommendations

made following old compared with new methods and processes for rare diseases) will be available in 2023 at the earliest

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

Owner: NHSE and NHS Digital

Original 2022 outputs:

- standard operating procedure for undertaking systematic component of variation (NHSE)
- 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

Progress report:

- the standard operating procedure has been developed
- equity of access project is underway. Information is being gathered from a number of data sources, such as Blueteq, and locally held NHS England systems.

Action 14 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- an annual report will be produced, and plans agreed as a way forward if access is deemed not equitable

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

Owner: DHSC and MRC

Original 2022 outputs:

- publicly available paper describing the rare disease research landscape, gaps, priorities and levers for change

Progress report:

- the project group (MRC, NIHR and DHSC) developed a search protocol to identify rare disease research in the portfolios of the 2 major public funders (MRC and NIHR) over the past 5 years
- search results have been validated and analysed, and a report detailing the results is being prepared
- the outputs of this action have been delayed as data analysis has proved more challenging than anticipated

Action 15 status (ongoing, extended or concluded): ongoing

Metrics and milestones for 2023:

- paper published by mid 2023
- continued work with industry and the charity sector to map the rare disease research funded by these organisations
- workshop held to identify ongoing priorities and next steps following paper publication

Action 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

Owner: NHSE

Original 2022 outputs:

- 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 – September 2023

Progress report:

- health inequalities are on the agenda for all the Annual Clinical Meetings for highly specialised services
- a paper on geographical access was presented to the Rare Diseases Advisory Group and a standard operating procedure for undertaking the process has been developed. The exercise will be repeated every 3 to 4 years
- a log of how health inequalities have been addressed in recent procurements has been developed
- NHS England has developed a draft framework and resource pack to help highly specialised commissioning teams understand and address health inequalities in procurement, which will be piloted with a few services

Action 16 status (ongoing, extended or concluded): concluded

Annex B: summary of actions for 2023 to 2024

This annex includes details on each action. As in 2022, each action is underpinned by a logic model, setting out the problem the action addresses, a

clear organisational owner, the outputs and outcomes that will be delivered, and the metrics used to measure progress, which will be reported on publicly.

The logic models also describe assumptions which underpin delivery of the actions, such as workforce stability, and external factors which may impact delivery, such as wider challenges to the healthcare system and competing organisational priorities.

Priority 1: helping patients get a final diagnosis faster

Action 17: commission research on how best to measure the diagnostic odyssey

Lead organisation(s): DHSC

Outputs:

- funding call launched
- research commissioned
- metrics developed to measure diagnostic odyssey

Outcomes:

- establish a baseline time to diagnosis
- assess effects of policy interventions on length of diagnostic odyssey
- basis for working with the NHS to identify and address challenges in delivering diagnoses faster
- identification of potential areas of health disparities

Action-specific monitoring and evaluation:

- funding call launched in early 2023
- outcomes of the 2-stage application process by autumn 2023, with research commencing as soon as possible after contracting
- the outcome of the call, the successful research proposal, and progress made will be reported in the 2024 action plan

Action 18: increased data-sharing for patient benefit to improve our understanding of equity of access to genomic testing and support interpretation of genomic test results

Lead organisations: Genomics England, NCARDRS and NHSE

Outputs:

- development of analytic plan for patients receiving tests against patients potentially eligible to receive a test, to investigate testing patterns and identify inequalities
- investigation of routes to share diagnostic variant data

- engagement with patient organisations to ensure patient benefit

Outcomes:

- the regulatory framework and technical pathway for sharing data within the healthcare ecosystem will be trialled as a proof of concept
- the role of central data at this level of granularity in monitoring equity of access will be assessed

Action-specific monitoring and evaluation:

By February 2024:

- one round of WGS data transfer from GEL to NCARDRS has taken place
- analysis plan in place at NCARDRS
- feasibility of transfer of non-WGS data from NHSE to NCARDRS assessed
- workshop held with patient organisations

Priority 2: increasing awareness of rare diseases among healthcare professionals

Action 19: publishing and implementing specific strategies for increasing awareness of rare diseases in the nursing and midwifery, pharmacy and primary care workforce

Lead organisation(s): Health Education England (this enhances and is in addition to HEE's wider work to ensure all healthcare professionals are aware of rare disease)

Outputs:

- GEP pharmacy lead in place to oversee education and training needs, including rare diseases
- genomics competency framework for nurses launched to address education and training (similar initiatives for midwives)
- rare disease case studies featured in the development of the nursing educator's toolkit
- GEP primary care lead contributing to Royal College of General Practitioners (RCGP) curricula review, RCGP genomics toolkit, primary care GeNotes and workforce development to include rare disease
- nursing and midwifery roundtable to help shape future education and training for the professionals including rare disease

Outcomes:

- improved understanding of rare diseases amongst the nursing and midwifery, pharmacy and primary care workforce

- a strategy outlining the approach to supporting educational and training needs for the pharmacy workforce will be published in early 2023. This work will encompass core concepts around genomics, enabling pharmacy teams to understand the application of genomics in healthcare, such as in the area of rare disease
- nursing and midwifery educational resources feature content on rare disease
- primary care education and training provision to include genomics featuring rare disease

Action-specific monitoring and evaluation:

- updated curricula to feature content on genomics and rare diseases
- continued engagement with stakeholders and relevant professional bodies to evaluate the uptake and impact of resources against baseline surveys of workforce in midwifery, pharmacy and primary care

Priority 3: better coordination of care

Action 20: commission research to provide the evidence needed to operationalise better coordination of care in the NHS

Lead organisation(s): DHSC

Outputs:

- funding call launched
- research commissioned
- evaluation of care coordination approaches complete

Outcomes:

- identification of the most cost-effective, tractable and impactful approaches to improving care coordination for people living with rare diseases
- evidence generated to support prioritisation of care coordination for possible future implementation within the NHS
- development of effective, evidence-based future policy, including facilitating policy alignment across wider government

Action-specific monitoring and evaluation:

- funding call launched in early 2023
- outcomes of the 2-stage application process by autumn 2023, with research commencing as soon as possible after contracting
- the outcome of the call, the successful research proposal, and progress made will be reported in the 2024 action plan

Action 21: include the definition of coordination of care in all new and revised services specifications for patients with rare diseases, and ensure the priorities of the UK Rare Diseases Framework are embedded across NHSE highly specialised services

Lead organisation(s): NHSE

Outputs:

- paper setting out requirements for any service specification involving patients with rare diseases to be formalised at the NHSE Specialised Commissioning Service Specification Tracking meeting
- new and revised service specifications include definition of care coordination
- priorities of UK Rare Diseases Framework discussed and reported on at HSS annual clinical meetings

Outcomes:

- increased awareness of framework priorities among commissioners and service providers
- HSS aligned with UK Rare Diseases Framework priorities, including coordination of care
- more joined up care (including access to mental health support) across HSS

Action-specific monitoring and evaluation:

- from 2023, all new and revised service specifications include definition of care coordination
- discussion of priorities of UK Rare Diseases Framework included as an agenda item at all HSS annual clinical meetings in 2023
- progress against framework priorities reported in 2024 action plan

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

Action 22: improved “findability” of people living with rare diseases using NCARDRS

Lead organisation(s): NCARDRS

Outputs:

- submission and/or publication of papers or reports describing methods to identify people with some rare diseases, including assessments on whether the methods might be applied to other diseases
- reporting requirements for specialised services commissioning specifications
- submission and/or publication of papers on descriptive epidemiology of some rare diseases

- public dashboard that includes information about rare disease data collected by NCARDRS, including which diseases, how it is collected and prevalence and incidence figures, where available
- external NCARDRS rare disease data dictionary
- web-based patient self-reporting system

Outcomes:

- improved national rare disease data for England, using peer-reviewed techniques that are potentially reproducible in the other countries and/or can be applied to other rare diseases
- improved understanding of rare diseases in England
- increased registration of people with rare disease to NCARDRS by NHSE specialised services
- increased transparency of the data that NCARDRS collects
- the NCARDRS rare disease data dictionary will make clear to external stakeholders which data items may be available and/or what analysis might be undertaken for different diseases
- patients will be able to register themselves with the national register, so they are included in national data for their disease, ensuring “findability”

Action-specific monitoring and evaluation:

- number specialised services sharing data with NCARDRS
- number of rare diseases with national (England) data including prevalence and incidence figures published on NDRS website
- number of patients who have self-registered through the new system

Action 23: continue to improve the understanding of the impact of NHS England’s specialised services commissioning activities on rare disease patients and act on this information

Lead organisation(s): NHSE

Outputs:

- PIAs considered in the development of all relevant commissioning policies
- revised RDAG terms of reference for the committee to consider all policy propositions for patients with rare diseases by spring 2023
- routine requests for Equality and Health Inequalities Impact Assessments (EHIA) included in provider selection exercises – from Spring 2023
- examples of new PROMs to be used in service specifications for patients with rare diseases

Outcomes:

- a continued and further developed process for ensuring that NHS England commissioning activities consider the impact of their activities on patients with

rare diseases

Action-specific monitoring and evaluation:

- number of PIAs considered
- number of policy propositions considered by RDAG
- confirmation that all provider selection exercises for patients with rare diseases have included a request from bidders for an EHIA, and examples of how providers have addressed issues raised in the EHIA policy propositions
- examples of best practice in PROMs

Action 24: establish a Highly Specialised Services Programme Board and strengthen the role of NHS England in commissioning wider services for patients with rare diseases

Lead organisation(s): NHSE

Outputs:

- Highly Specialised Services Programme Board established to provide assurance that services for patients with rare diseases continue to have a high profile and are of high quality
- extended clinical membership of the Rare Diseases Advisory Group (RDAG)
- clear terms of reference for both groups to provide clarity of roles and decision-making

Outcomes:

- role of NHSE in commissioning HSS and wider services for patients with rare diseases strengthened
- clinical advice and clinical leadership role of RDAG strengthened
- a continued focus, in light of the implementation of the Health and Care Act 2022, on highly specialised services and the Rare Disease Framework

Action-specific monitoring and evaluation:

- Highly Specialised Services Programme Board established by June 2023
- terms of reference of Rare Diseases Advisory Group (RDAG) revised by June 2023
- new clinical members recruited to RDAG and given an induction

Action 25: review the effectiveness of EAMS, ILAP and the IMF in supporting access to treatments for people living with rare diseases

Lead organisation(s): NHSE, NICE, MHRA

Outputs:

- report on number of applications and medicines made available through the schemes, which are treatments for rare diseases

Outcomes:

- improved understanding of the effectiveness of these schemes for improving and supporting access to rare disease medicines

Action-specific monitoring and evaluation:

- proportion of applications which are rare disease medicines reported in 2024 action plan
- number of rare diseases medicines progressing through the schemes reported in 2024 action plan

Action 26: registration of national data for exemplar rare genetic conditions which cause an inherited predisposition to cancer

Lead organisation(s): NCARDRS

Outputs:

- outputs using the national data to possibly include publications, dashboards and data sharing of aggregate variant level data with relevant stakeholders including CanVAR-UK, DECIPHER and GEL

Outcomes:

- improved understanding of these diseases (including cancer risk), which will support better coordination of care, access to new treatments, and better outcomes for those that have them

Action-specific monitoring and evaluation:

- number of conditions with a predisposition to cancer registered

Action 27: improving the Be Part of Research platform for people living with rare diseases

Lead organisation(s): DHSC, NIHR

Outputs:

- further development of the Be Part of Research platform will be informed by feedback from the rare diseases community
- the rare disease community will be represented on Be Part of Research's stakeholder engagement group

Outcomes:

- the Be Part of Research platform will effectively meet the needs of the rare disease community

- awareness of clinical research participation opportunities will be increased within the rare disease community

Action-specific monitoring and evaluation:

- the rare disease community will be invited to take part in private beta phase testing of the Be Part of Research platform user interface by autumn 2023
- outcomes of consultations with the rare disease community on the most effective way to categorise rare diseases studies on the Be Part of Research platform will be reported in the 2024 action plan

Action 28: develop a plan to include rare diseases in NHSE's Core20PLUS5 Framework

Lead organisation(s): DHSC, NHSE

Outputs:

- evidence collated to support the inclusion of people living with rare diseases in the 'PLUS' target population cohort
- people living with rare diseases are highlighted as a population to be identified by integrated care systems (ICSs) in the 'PLUS' target population in the 'Core20PLUS5' Framework to improve health inequalities
- the Core20PLUS5 Framework is applied to people living with rare diseases, to detail steps that could be taken within NHSE to address health inequalities for people living with rare diseases

Outcomes:

- people living with rare diseases recognised by NHSE as a population group that are likely to experience poorer than average access, experience and/or outcomes in healthcare services
- increased awareness of the inequalities in access, experience and outcomes faced by people living with rare diseases
- ICSs work to reduce health inequalities faced by people living with rare diseases. People living with rare diseases recognised by NHSE as a population group that are likely to experience poorer than average access, experience and/or outcomes in healthcare services
- increased awareness of the inequalities in access, experience and outcomes faced by people living with rare diseases
- ICSs work to reduce health inequalities faced by people living with rare diseases

Action-specific monitoring and evaluation:

- plan for how work will be carried out developed by autumn 2023
- collation of evidence to support the inclusion of rare diseases in the 'PLUS' category to begin by winter 2023

- report of the work done to support the inclusion of people living with rare diseases in the ‘PLUS’ target population included in the 2024 England Rare Diseases Action Plan

Action 29: commission portfolio level evaluation of England’s Rare Diseases Action Plans with input from the rare disease community on design of metrics

Lead organisation(s): DHSC

Outputs:

- funding call launched
- research commissioned
- metrics developed

Outcomes:

- evaluation of the influence and outcomes of the framework using metrics co-developed with rare disease community
- identification of potential areas of disparity
- development of effective, evidence-based future policy

Action-specific monitoring and evaluation:

- funding call launched in early 2023
- outcomes of the two-stage application process by autumn 2023, with research commencing as soon as possible after contracting
- the outcome of the call, the successful research proposal, and progress made will be reported in the 2024 action plan

Annex C: further detail on progress in 2022

Promoting access to specialist treatments

The Early Access to Medicines Scheme (EAMS), led by the Medicines and Healthcare products Regulatory Authority (MHRA), promotes access to medicines that patients with life threatening or debilitating conditions would be otherwise unable to access. A legal basis for EAMS came into force in April 2022 through the Health and Care Act. This included provision for supporting the collection of real-world data (RWD) during the EAMS scientific opinion period, with the goal to streamline this evidence generation activity. This could be particularly useful for those developing medicines in rare diseases, where additional data sources can help offset uncertainties of the smaller clinical development programme.

The Innovative Licensing and Access Pathway (ILAP) supports the path to market of innovative and novel treatments, including those for rare diseases,

while ensuring there are no compromises in assessing the safety and efficacy of the treatments. In March 2022, the ILAP Health Technology Assessment access forum tool was announced. The tool aims to help:

- developers understand where the new medicine may potentially fit in the care pathway
- service delivery implications of introducing the new medicine to the NHS
- high-level health technology assessment challenges
- the application of processes in practice
- commercial options to support the new medicines value proposition and minimise financial burden on the NHS

In July 2022, the Innovation Accelerator (IA) was announced and provides innovators and developers of innovative products access to MHRA scientific expertise, regulatory guidance and enhanced advice and signposting. The IA engages with many groups working on rare diseases including the Association of Medical Research Charities, the Cell and Gene Therapy Catapult, and the AGORA Initiative (Access to Gene therapies fOr RAre diseases) that aims to establish sustainable regulatory pathways for patients to access medicines for rare conditions.

Pioneering research

Case study: NIHR BioResource Rare Diseases RNA Phenotyping Project

The Rare Diseases RNA Phenotyping Project (https://bioresource.nihr.ac.uk/using-our-bioresource/our-cohorts/rare-diseases-bioresource/rare-diseases-rna-phenotyping-project/) was set up following the success of the whole-genome sequencing study from the Rare Diseases BioResource that contributed to the 100,000 Genomes project.

The project involves collection of a blood sample from people with various rare diseases, which is then used to generate a large epigenetic dataset on each patient, including RNA, whole-genome and single-cell sequencing, proteomics, and deep phenotyping. This dataset will provide a comprehensive genetic and clinical picture across multiple rare disease areas that will facilitate research to improve understanding of the causes, diagnosis and treatment of several rare diseases.

17 different rare disease areas (https://bioresource.nihr.ac.uk/studies/?speciality=&studytype=&researchertype=&tag=Rare%2BDiseases%2BRNA%2BPhenotyping&location=) are now involved in the RNA Phenotyping Project, including bleeding, thrombotic and platelet disorders, inherited neurological disorders, stem cell and myeloid disorders and giant cell arteritis. Approximately 1,000 participants will take part in the initial phase of the project.

A new satellite laboratory at the BioResource Centre in Leeds has also been set up so that biological samples from the north of England can reach a laboratory more rapidly for processing before the RNA profile of the cell changes. Data from this project will be shared through the NIHR BioResource.

Annex D: supporting activities under the 4 priorities of the UK Rare Diseases Framework

Priority 1: helping patients get a final diagnosis faster

Advances in genomic medicine

For the approximately 80% of rare diseases with a genetic origin, genomic testing through the NHS Genomic Medicine Service (GMS) is a key part of the diagnostic process. Launched in 2018, the GMS enables the NHS to harness the power of genomic technology and science to improve health. Currently over 600,000 genomic tests are carried out in England every year, for conditions including rare diseases.

Published in autumn 2022, Accelerating genomic medicine in the NHS (https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/) sets out NHSE's strategy for embedding genomics in the NHS. It details how the NHS GMS will evolve over the next 5 years to deliver a high-quality, equitable and affordable service, with priorities for improving services for the rare disease community. This includes plans to increase the capacity and capability of the workforce, including clinical geneticists and genomic counsellors.

In December 2022, the Office for Life Sciences published Genome UK: 2022 to 2025 implementation plan for England (https://www.gov.uk/government/publications/genome-uk-2022-to-2025-implementation-plan-for-england/genome-uk-2022-to-2025-implementation-plan-for-england) with a range of specific actions that genomics delivery partners in England will take to implement the commitments in Genome UK (https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare).

Alongside continuing roll out of whole genome sequencing in the GMS, the plan includes £105 million of government funding for Genomics England to lead the landmark research programme, in partnership with the NHS, to study the effectiveness of using whole genome sequencing to find and treat rare genetic diseases in newborns (see also section 1, priority 1).

These publications come at a pivotal moment for genomics in the NHS, as technological, clinical and scientific developments, together with increased affordability, have led to an acceleration in the opportunities to improve patient

and population health using genomic medicine. One such advance is the publication in June 2022 of recommendations for updating existing standards for determining the disease-causing potential of genomic variants (https://www.genomicsengland.co.uk/news/updated-global-guidelines-proposed). Building on findings from the 100,000 Genomes (https://www.genomicsengland.co.uk/initiatives/100000-genomes-project) cohort, the recommendations are a starting point for standardising and refining the characterisation of so-called 'variants of unknown significance', helping to improve the diagnostic yield of whole genome sequencing tests.

A good diagnosis

Getting a final diagnosis faster is an important step in ensuring people living with rare diseases are able to access the right treatment and care. But as Genetic Alliance UK's 2022 'Good Diagnosis' report emphasises (https://geneticalliance.org.uk/gauk-news/news/good-diagnosis/), reducing the time to diagnosis is only one element of improving the diagnostic experience. While a lengthy diagnostic odyssey can have significant implications for the physical and mental health of people living with rare conditions and their families, a good diagnosis can only be achieved if people with rare conditions have access to information and support, throughout their diagnostic journey and beyond. This includes mental health support at the point of diagnosis and beyond – a fact highlighted by recently published work on the 'Emotional Odyssey' by Rare Diseases Research Partners (RDRP).

Particular consideration should be given to the specific impact of delivering and receiving a rare disease diagnosis in childhood, on both the individual and their family, as outlined in the guidelines on genetic testing in childhood (https://www.rcplondon.ac.uk/projects/outputs/genetic-testing-childhood) published jointly by the Royal College of Physicians, Royal College of Pathologists and British Society for Genetic Medicine in November 2022.

It is also important to consider the implications of predictive testing, where this is available, recognising that the time frame to diagnosis in people without symptoms should be in their control, and some may never wish to receive a 'predictive diagnosis'.

Priority 2: increasing awareness of rare diseases among healthcare professionals

Building on their actions outlined in the 2022 action plan, Health Education England (HEE) has also continued to work closely with genomic education experts, Royal Colleges, professional bodies and charities to create rare disease-relevant educational tools and resources for the healthcare workforce. This includes ongoing work to evaluate the use and effectiveness of GeNotes, as outlined in section 1, and the Clinical Pathway Initiative (CPI) (https://www.genomicseducation.hee.nhs.uk/the-clinical-pathway-initiative/), a collaboration between NHSE, HEE and the Academy of Medical Royal Colleges

to facilitate the integration of genomic medicine through the alignment of different patient pathways, workforce development and education and training requirements. A number of pathways involving rare diseases have been prioritised as part of an initial CPI pilot.

From January 2023 HEE will also be working with Medics 4 Rare Diseases, to enhance the development of networks and resources for their rare disease hub, including those relating to non-genetic rare diseases. Activity will include connecting the Genomics Education Programme (GEP) to the rare disease advocacy community; enhancing patients', family members' and carers' involvement in the GEP; building relationships with cross-sector stakeholders; and promoting GEP resources at rare community events and on M4RD platforms.

HEE has also developed an easily accessible, 2-week rare diseases massive open online course (MOOC) (https://www.genomicseducation.hee.nhs.uk/news/genomics-in-the-nhs-a-clinicians-guide-to-genomic-testing-for-rare-disease/), aligned to genomic rare disease patient pathways, aimed at healthcare professionals working with people and families with rare diseases. The MOOC, launched in November 2022, is designed to support clinicians to request genomic tests and feedback results, and is hosted on the FutureLearn platform. Data on engagement of healthcare professionals with the MOOC will be gathered and shared with England's Rare Diseases Framework Delivery Group.

HEE is in the early stages of developing a genomic advisor framework, identifying the knowledge, skills and behaviours that healthcare professionals need to spearhead the integration of genomic medicine across healthcare specialties. This will also include developing a network of experts who can facilitate optimal access to genomic testing and support people living with rare diseases through genomic testing and into their onward pathway of care, including signposting to appropriate psychological support. All of HEE's resources are being developed in collaboration with the Genomics Education Programme Patient Advisors for Genomic Education.

A 'month of genomics' is also planned for 2023, with HEE collaborating with Royal Colleges to raise awareness and promote engagement across specific speciality areas and professions in genomics. Initially working with the RCOG, the RCOM and the RCPCH, a series of activities is planned between January and March 2023, to include vlogs, webinars and events, as well as contributions to a range of GEP education and training resources.

Because people living with rare conditions often require support beyond healthcare, we will continue to work with other government departments to increase awareness of rare diseases and ensure they are taken into account in broader policy, as outlined in the 'wider policy alignment' section below.

The role of NHS Genomic Medicine Service Alliances in supporting workforce development

To further support the embedding of genomic testing and implementation of the NHS Long Term Plan commitments, 7 NHS Genomic Medicine Service (GMS) Alliances were established in December 2020. An NHS GMS Alliance is a collective made up of a small number of NHS providers working in partnership to support the strategic systematic embedding of genomic medicine for a given population. They drive this embedding across all providers within their geography from primary and community care to secondary and tertiary care. The infrastructure includes multidisciplinary clinical leadership to secure input from a range of professions, including clinical leads for different areas such as cancer, nursing, midwifery, allied healthcare professionals, and pharmacy.

The NHS GMS Alliances support the Nursing and Midwifery Collaborative, a pioneering project that aims to empower nurses and midwives to design or implement an agreed approach or framework into their practice, to drive an increase in genomic testing.

In February 2022, NHS North-East and Yorkshire GMS Alliance and NHS North-West GMS Alliance hosted ‘From niche to necessity’, a webinar for nurses, midwives and health visitors. The webinar raised awareness of the latest genomic developments across the north of England and demonstrated the importance and benefits of genomics in clinical practice.

The NHS GMS Alliances each have a pharmacy lead within their infrastructure to ensure the strategic delivery of education and training opportunities to support the pharmacy workforce to realise the benefits of genomics for medicines optimisation and precision medicine initiatives.

Priority 3: better coordination of care

Rare disease collaborative networks

Rare disease collaborative networks (RDCNs) are an important part of NHS England’s provision to improve care, and support patients with rare diseases. The current 13 rare disease collaborative networks (https://www.england.nhs.uk/commissioning/spec-services/highly-spec-services/rare-disease-collaborative-networks/) continue to demonstrate the breadth of work that has been undertaken by the centres, with an emphasis on collaboration and clarifying pathways for patients. Five additional RDCNs have also recently been approved, covering PTEN Hamartoma Tumour syndrome, rare bone conditions, arteriopathy and aortopathy, Bloom’s syndrome and Tuberous sclerosis complex.

Quality standards for rare diseases

NICE guidance and quality standards (https://www.nice.org.uk/about/what-we-do/into-practice/resources-help-put-guidance-into-practice/how-guidance-standards-help-you) are important tools in developing and delivering high-quality, evidence-based care. Over the past year, a group of UK Rare Diseases Forum members has met

regularly to discuss the possibility of developing quality standards for rare diseases. In December 2022, this group was formally recognised as the first forum Independent Advisory Group (IAG) (see the section 'Patient voice', below). We have committed to taking the recommendations produced by the group to our delivery partners, and to feeding back how these recommendations have been considered.

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

A proportionate approach to NICE technology appraisals

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 that provides the most health benefit for society. The range and complexity of technologies that NICE now reviews are greater than ever. But not all technologies require the same type of evaluation.

NICE is exploring new approaches for taking a proportionate approach to technology appraisals (https://www.nice.org.uk/about/what-we-do/proportionate-approach-to-technology-appraisals), including technologies for rare diseases. A pilot using this approach for Vutrisiran, for treating hereditary transthyretin-related amyloidosis, resulted in the publication of final draft guidance 20 weeks (60%) faster than the normal process. NICE will continue to explore and roll out these processes in 2023.

UK Clinical Research Recovery Resilience and Growth

The UK Clinical Research Recovery Resilience and Growth (RRG) programme is delivering the Vision for the Future of UK Clinical Research Delivery (https://www.gov.uk/government/publications/the-future-of-uk-clinical-research-delivery), which sets out the government's ambition for delivering innovative, people-centred clinical research. A phase 1 implementation plan (https://www.gov.uk/government/publications/the-future-of-uk-clinical-research-delivery-2021-to-2022-implementation-plan) was published in June 2021, and the phase 2 implementation plan (https://www.gov.uk/government/publications/the-future-of-uk-clinical-research-delivery-2022-to-2025-implementation-plan/the-future-of-clinical-research-delivery-2022-to-2025-implementation-plan) in June 2022.

In the phase 2 implementation plan we committed to maintaining a rich and balanced portfolio of studies in rare and common diseases, ranging from complex, intensive studies in small, highly targeted populations to pragmatic population health research in large cohorts, and included a section on rare diseases on our RRG microsite (https://sites.google.com/nihr.ac.uk/thefutureofukclinicalresearch/home/the-future-of-uk-

clinical-research-delivery/programme-updates/aligning-our-research-programmes-and-processes-with-the-need#h.fgjmpx6gme0) to provide further information.

The ongoing effects of the COVID-19 pandemic continues to impact on clinical research delivery in the NHS, with increased workload pressure on the NHS R&D workforce, the challenges of recovery of wider NHS services and changes to care pathways. We need to return to a situation in which new studies are able to be delivered within planned timescales. We are taking steps to address this with the aim of 80% of all open studies on the National Institute for Health and Care Research (NIHR) Clinical Research Network portfolio being delivered to time and target by June 2023.

As we continue work to reset the portfolio of clinical research, we are looking to preserve a diverse portfolio of studies that are deliverable. We recognise that for complex or rare diseases a study can be both resource-intensive and practicable. We will continue to ensure studies across a wide range of conditions, settings and levels of complexity remain on the portfolio and can be progressed. Further information is available at the Research Reset section of the RRG microsite. (https://sites.google.com/nihr.ac.uk/thefutureofukclinicalresearch/home?authuser=0)

Supporting clinical research in the NHS Genomic Medicine Service

The recently published NHS genomics strategy Accelerating genomic medicine in the NHS (https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/) includes a number of commitments to support clinical research in genomics, of particular relevance to people living with genetic rare diseases. These include:

- aligning clinical trial targets with standard of care NHS testing
- expanding the use of NHS generated genomic data to support approved research
- working with patients, the public and key partners to evolve the patient choice framework
- putting in place mechanisms to enable the consent to, and collation of, NHS genomic sequencing data for research and innovation purposes at a national and regional level

The NHS Genomic Medicine Service (GMS) Research Collaborative was established as a partnership between NHS England, the NHS GMS, Genomics England and the NIHR to facilitate and fulfil the research mission. The NHS GMS Research Collaborative aims to make it easier for researchers in academia, the NHS and the life sciences industry to conduct interactive genomic research and validate new genomic technologies, diagnostics and treatments to drive improvements for patients and the NHS.

Supporting activities under underpinning themes

Patient voice

Case study: patient-led research – Ring 20 Research and Support UK and the NIHR BioResource

Ring Chromosome 20 Syndrome (r(20)) is a very rare condition where one copy of chromosome 20 has formed a ring rather than the typical linear structure. Typically, this results in early childhood onset of severe epilepsy and is associated with learning difficulties. It is believed that r(20) is sometimes under- or mis-diagnosed.

In collaboration with Ring20 Research and Support UK, the NIHR BioResource has collected DNA samples for the UNRAVEL study, a joint patient-led project. Whole genome sequencing has been performed on these samples for further understanding of ring chromosome syndromes.

Through continued collaboration with Ring 20 Research and Support UK, a r(20) patient cohort will be established within the Rare Diseases BioResource in 2023. Similar models, used by the NIHR BioResource for more than 50 rare diseases, have been successful with other charity partners, such as the Cystic Fibrosis Trust.

Digital, data and technology

The European Platform on Rare Disease Registration (https://eu-rd-platform.jrc.ec.europa.eu/_en), developed by the European Commission's Joint Research Centre, aims to overcome the fragmentation of rare disease registries across Europe by making rare disease registry data discoverable, searchable and findable.

The platform facilitates collaboration between registries, maximises the value of each registry's information and enables extended use of registries' data. The platform also enables pseudonymisation and data-linkage between registries, to enable encrypted and secure data transfer. Alignment, interoperability and sharing of data between registries at an international level will result in the most effective use of data arising from the rare disease community.

Wider policy alignment

The Minister for Disabled People announced plans for a new Disability Action Plan (DAP) in December 2022. The new DAP will set out the practical actions government will take over the next 2 years to improve disabled people's lives. The policies in the DAP will be developed and consulted on over the course of 2023, so that disabled people, disabled people's organisations and other interested parties, including people living with rare diseases, can have their say.

In March 2022, the Department for Education announced high needs provision capital allocations (https://www.gov.uk/government/publications/high-needs-provision-capital-allocations) amounting to over £1.4 billion of new investment. This funding

forms part of the £2.6 billion the government is investing in special educational needs and disabilities support between 2022 and 2025, and represents a significant investment in new high needs provision. It will support local authorities to deliver new places in mainstream and special schools, as well as other specialist settings, and will also be used to improve the suitability and accessibility of existing buildings.

The Department for Work and Pensions expects to publish a Health and Disability White Paper in the coming months. It will respond to key areas set out in the 2021 Health and Disability Green Paper (https://www.gov.uk/government/consultations/shaping-future-support-the-health-and-disability-green-paper/shaping-future-support-the-health-and-disability-green-paper), including improving services provided to disabled people, and how government can support more disabled people to start, stay and succeed in work.

These new announcements are supported by existing initiatives, such as the Disabled Facilities Grant (DFG) (https://www.gov.uk/disabled-facilities-grants), a locally administered grant that contributes to the cost of home adaptations, including stair-lifts, heating systems and assistive technology, for eligible people to help them live safely and independently at home.

The Down Syndrome Act 2022 (https://www.legislation.gov.uk/ukpga/2022/18/enacted) became law in April 2022. The act aims to ensure that health, social care, education and other local authority services take account of the specific needs of people with Down syndrome when commissioning or providing services. The government made a commitment as the Down Syndrome Bill was progressing through parliament to consider whether the support needs of people with Down syndrome and people with other genetic conditions are similar.

In July 2022, DHSC launched a national call for evidence to inform the development of draft guidance on the act for local authorities, which will then be subject to a full public consultation. The call for evidence sought the views of people with Down syndrome, parents and carers, professionals and people with a learning disability and other genetic conditions. The information and evidence received through the call for evidence will inform the development of the Down Syndrome Act guidance.

We will continue to work closely with other government departments to ensure that the voices of people and families living with rare diseases are heard as policy is developed. We will also continue to raise awareness within the rare diseases community of opportunities to input into wider policy.

Annex E: commissioning of rare disease services in NHS England

The 2022 Health and Care Act (https://www.legislation.gov.uk/ukpga/2022/31/contents/enacted) marks an important step in the government's ambitious health and care agenda. It sets up systems

to reform how health and adult social care work together, tackle long waiting lists built up during the COVID-19 pandemic, and address some of the long-term challenges faced by the country, including a growing and ageing population, chronic conditions (including rare diseases) and inequalities in health outcomes.

The act supports a more joined-up system, ensuring that every part of England is covered by an integrated care board (ICB) and integrated care partnership (ICP), which bring together NHS, local government and wider system partners to empower them to put collaboration and partnership at the heart of planning.

ICBs are responsible for commissioning NHS services against local population needs and are responsible for working with providers to review capacity for healthcare services in their area. When commissioning healthcare services, ICBs have a duty to consider the health and wellbeing of the people of England (including inequalities in health and wellbeing), as well as the quality, sustainability and efficient use of services provided or arranged by both themselves and other relevant bodies (including inequalities in benefits from those services). ICBs are formally accountable to NHS England (NHSE), which formally oversees both ICBs and providers, and regularly assesses whether they need any support to deliver required standards and performance.

This new legislative framework focuses the ICBs on their local population's health, presenting an opportunity for specialised services, commissioned to care for patients with some rare conditions, to be fully integrated into the design and delivery of local pathways of care for patients. The Roadmap for integrating specialised services within Integrated Care Systems (https://www.england.nhs.uk/publication/nhs-england-commissioning-functions-for-delegation-to-integrated-care-systems/) publication sets out a phased and managed approach to integrating commissioning of specialised services, currently commissioned nationally, with wider ICB commissioning responsibilities.

The expectation for 2023 to 2024 financial year is that ICBs and NHSE will set up joint working arrangements overseen by a joint committee. This means that, from April 2023, they will have joint responsibility for the specialised services that have been assessed as suitable and ready for greater involvement of ICBs. It is the intention that ICBs will take on statutory delegated commissioning responsibility for these services from April 2024, subject to system readiness.

Not all responsibilities will be delegated to ICBs, however, and NHSE will continue to retain responsibility for commissioning highly specialised services, where patient numbers are typically less than 500 people per year, as well as for clinical genomic services inclusive of genomic counselling. Regardless of delegation status, all services will continue to have national standards attached to them and NHSE will continue to be the accountable commissioner for all specialised services, including those serving people with rare conditions.

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