



DEBATE PACK

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Implementation of the UK Rare Diseases Framework

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Summary

This briefing has been produced ahead of the debate on the Implementation of the UK Rare Diseases Framework, in Westminster Hall on 24 March 2021 from 4:50pm until 5:50pm. Liz Twist MP will open the debate.

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1. Background

In January 2021, the Department of Health and Social Care (DHSC) published the [UK Rare Diseases Framework](#) which set out a 'community-led vision to build upon existing UK strengths and improve areas of weakness to deliver better health outcomes and improve the lives of those living with rare diseases'.¹ The Framework follows on from a [2013 strategy](#) which was due to end in 2020.

The Framework defines a rare disease as a condition which affects less than 1 in 2,000 people and covers over 7,000 rare diseases.² Whilst 80% of rare diseases have an identified genetic origin, other causes include disordered immunity, infections, allergies, deterioration of body tissues and organs or disruption to development while in the womb.³

Rare diseases are individually rare but can affect up to 1 in 17 people at some point in their lifetime, amounting to over 3.5 million people in the UK.⁴

Rare diseases can be 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.⁵

Rare Disease Day takes place on the last day of February each year, most recently on 28 February 2021. Its main objective is to raise awareness of rare diseases and their impact on patients' lives, amongst the general public and decision makers. Further information is available on the [Rare Disease Day website](#).

2013 UK Strategy for Rare Diseases

In 2013 the then Department of Health published the [UK Strategy for Rare Diseases](#) which aimed to:

[...] ensure that people living with a rare disease have the best quality of evidence based care and treatment that our health and social care systems, working with charities and other organisations, our researchers and industry can provide.⁶

The strategy set out 51 commitments, centred on:

- Empowering those affected by rare diseases
- Identifying and preventing rare diseases
- Diagnosis and early intervention
- Coordination of care
- The role of research⁷

¹ DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

² DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

³ DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

⁴ DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

⁵ DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

⁶ DHSC, [UK Strategy for Rare Diseases](#), 22 Nov 2013

⁷ DHSC, [UK Strategy for Rare Diseases](#), 22 Nov 2013

The strategy was due to come to an end at the end of 2020.⁸ Further information about background to the strategy and its progress is provided in Section 1 of a 2017 Commons Library Debate Pack, [Implementing the UK Strategy for Rare Diseases](#).⁹

Following the strategy, the four UK nations were required to produce individual implementation plans that set out how they would put the strategy into action, in line with their national policies and priorities.

The DHSC published their [Implementation Plan for England](#) in January 2018, setting out an overarching view and the actions the DHSC would take to coordinate the delivery of the commitments by its partner organisations.¹⁰ NHS England's [Implementation Plan for the UK Strategy for Rare Diseases](#),¹¹ published in January 2018, is intended to be read alongside it, and together the plans cover the 51 relevant commitments laid out in the strategy.

[Rare Disease UK](#) is a multi-stakeholder campaign run by [Genetic Alliance UK](#), working with the rare disease community and the UK's health departments to implement the UK Strategy for Rare Diseases. Responding to the publication of the 2018 DHSC and NHS England implementation plans, Rare Disease UK [described](#) the plans as 'promising news' to those affected by rare conditions.¹² Commenting on the decision to deliver two separate plans, Rare Disease UK expressed concern that this would pose a challenge for their delivery and monitoring.¹³

Updates of the implementation plan for England were published in [2019](#)¹⁴ and [2020](#).¹⁵

Dr Jayne Spink, Chair of Rare Diseases UK and Chief Executive of Genetic Alliance UK, [welcomed the 2019 update](#) but expressed concern that it did not provide a comprehensive overview of progress:

We welcome this progress update from Department of Health and Social Care. However, since many of the recommendations of the UK Strategy for Rare Diseases fall to NHS England to implement, the report does not give a comprehensive picture. Nonetheless, the Department of Health and Social Care progress report demonstrates the scale of change over the past six years – fundamental changes that will increasingly affect the NHS and prospects for people affected by rare diseases. The case for urgently reviewing and updating the UK Strategy for Rare Disease is compelling – before its end date in 2020'.¹⁶

⁸ DHSC, [UK Strategy for Rare Diseases](#), 22 Nov 2013

⁹ House of Commons Library, [Implementing the UK Strategy for Rare Diseases](#), 24 Mar 2017

¹⁰ DHSC, [UK strategy for rare diseases: implementation plan for England](#), 29 Jan 2018

¹¹ NHS England, [Implementation Plan for the UK Strategy for Rare Diseases](#), 29 Jan 2018

¹² Rare Disease UK, [Implementation plans for England published](#), 29 Jan 2018

¹³ Rare Disease UK, [Implementation plans for England published](#), 29 Jan 2018

¹⁴ DHSC, [UK strategy for rare diseases: 2019 update to the implementation plan for England](#), 27 Feb 2019

¹⁵ DHSC, [UK strategy for rare diseases: 2020 update to the implementation plan for England](#), 26 Feb 2020

¹⁶ Rare Disease UK, [Update to the Implementation Plan for England](#), 1 Mar 2019

NHS England published the latest [progress report](#) in November 2020.¹⁷

Genome UK

In September 2020, the Government published [Genome UK: the future of healthcare](#), setting out how the genomics community would come together to take advantage of advances in genetic and genomic science, research and technology for the benefit of patients.¹⁸

Referencing the 80% of rare diseases which have a genetic component, the Government set out proposals for rare and inherited diseases. This included the NHS in England being the first national healthcare system to offer whole genome sequencing (WGS) as part of routine care via the NHS Genomic Medicine Service, with an aim to extend WGS to all rare disease patients without a genetic diagnosis. The Government set out aims to continue to increase the diagnostics rate from genomic testing, continue to reduce the time people wait for a rare disease diagnosis, make it easier for researchers to feedback findings relevant to a patient's care and make it easier for patients with rare diseases to be enrolled in specialist clinical trials.

UK Research and Development Roadmap

In July 2020, the Department for Business, Energy and Industrial Strategy (BEIS) published the [UK Research and Development Roadmap](#), marking the 'start of a conversation' to identify:

- The strengths and challenges facing the sector
- The issues that need to be addressed
- How the Government wants to work with universities, business, the third sector and across government to cement the UK's reputation as a science superpower¹⁹

¹⁷ NHS England, [Implementation Plan for the UK Strategy for Rare Diseases – progress report](#), first published 11 Jul 2019, updated 10 Nov 2020

¹⁸ HM Government, [Genome UK: the future of healthcare](#), 26 Sep 2020

¹⁹ BEIS, [UK Research and Development Roadmap](#), published 1 Jul 2020

2. 2021 UK Rare Diseases Framework

In January 2021, the Department of Health and Social Care (DHSC) published the [UK Rare Diseases Framework](#), which aims to ensure that the lives of people living with rare diseases continue to improve.²⁰ The Government made a number of commitments, including using advanced diagnostic technology to help patients receive a faster diagnosis, increasing awareness of rare diseases amongst healthcare professionals and improving access to innovative treatments and specialist care.

The Framework set out the background work undertaken by the Government:

To identify the priorities for the next 5 years, we undertook a programme of engagement to understand the main challenges for those living and working with rare diseases across the UK and how these could be addressed. In October 2019 government launched the National Conversation on Rare Diseases Survey to seek the views of patients, their families, clinicians, researchers and rare diseases patient organisations. The survey ran for 6 weeks and received a remarkable 6,293 responses from the community. The results of the survey can be found at Annex A.²¹

The Framework then sets out how its priorities and underpinning themes were determined:

Following the survey, an Editorial Board of policy officials, representatives from clinical practice and patient organisations was formed to formally identify and refine the priorities and underpinning themes for the new framework. These ideas were further tested through stakeholder engagement with patient organisations, clinicians, researchers and industry representatives and were put to the UK Rare Disease Policy Board and Rare Diseases Advisory Group for discussion.²²

The Framework is being developed in two key phases.

Phase 1 is delivered through the January 2021 Framework document which sets out four priorities and five underpinning themes for improving the lives of those living with rare diseases across the UK. Phase 1 sets out a high-level vision for each of these priority areas, shared by all UK nations, providing a strategic direction for the UK's work on rare diseases across the next five years, at which point it will be reviewed.

In **Phase 2**, each nation will develop an action plan, highlighting steps that they will take to meet the aims of the framework in accordance with their own arrangements. With respect to health being a devolved matter, each nation will have the flexibility to deliver the aims of the framework in a way that is most effective for their population. Each UK nation will follow a set of core principles when delivering action plans

²⁰ DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

²¹ DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

²² DHSC, [UK Rare Diseases Framework](#), 9 Jan 2021

and implementing the framework. In a [December 2020 PQ response](#), DHSC Minister Jo Churchill said that the Devolved Administrations have agreed to publish the nation specific action plans within two years of the Framework publication.²³

Rare Disease UK [welcomed](#) the Framework and said that it was looking forward to working with the UK nations to help deliver its aims, and that 'it is now time' to put the Framework into action.²⁴

2.1 Priorities and themes

Health is a devolved matter and as such each UK nation will deliver the aims of the Framework in a way that is most effective for their respective populations. The Government has set out core principles when delivering action plans and implementing the framework. The principles have been set out in four key priorities, which have been highlighted as major challenges by the rare disease community. The strategy identified progress in these areas as being vital in meeting the commitment to improve the lives of those living with rare diseases.

Additionally, five underpinning themes were identified in which work will continue to be progressed to support the priorities of the Framework and improve the lives of those living with rare diseases.

Priority 1: helping patients get a final diagnosis faster

The strategy identified difficulties in obtaining the right diagnosis as a consistent and significant challenge faced by people in both the genetic and non-genetic rare disease community. The strategy highlighted a long and multi-step process to diagnosis, including multiple referrals, inconclusive tests and sometimes incorrect diagnoses, before a final diagnosis. The strategy also highlighted that some people living with a rare disease may never receive a diagnosis that addresses the underlying disease.

The strategy highlighted the potential of screening proposals, including the potential to use genomics in testing for many of the conditions currently included in the blood spot test.

The Government said that it expects that new, validated genomics approaches and diagnostic tools would contribute to further improvements in diagnosis and screening, including improved recognition of which patients should undergo advanced genetic testing.

Priority 2: increasing awareness of rare diseases among healthcare professionals

The strategy pointed out that many GPs will not routinely have the knowledge or experience to correctly identify patients suffering with a rare condition, with a similar gap in knowledge for paramedics and A&E staff with respect to emergency hospital care for rare diseases patients. The strategy set out the importance of healthcare professionals being

²³ [PQ 123605](#), 9 Dec 2020

²⁴ Rare Disease UK, [Our thoughts on the UK Rare Diseases Framework](#), 9 Jan 2021

aware of rare diseases more widely, alert to considering them and being provided with the necessary education and resources.

Priority 3: better coordination of care

The management of patients with rare diseases may require the expertise of multiple specialists who may not be located in the same locality. The strategy also highlighted the burden this places on patients and parents of children affected by rare diseases, and the challenges in ensuring continuity of care across paediatric and adult services. The strategy pointed to the advantages that technology and digital tools provide, which could allow patients to access services remotely and enable specialists from across the system to share information and discuss tailored care plans.

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

The strategy acknowledged the significance of the few rare diseases that have established treatment, and highlighted opportunities to develop innovative models of care across the healthcare system so that patients have their care delivered as locally as possible. The strategy noted that assessment of, and access to, rare disease medicines for small patient populations could provide challenges to health technology assessment bodies. It pointed to the work of the Early Medicines Scheme, the Accelerated Access Collaborative and the expansion of the Innovative Medicines Fund in improving access to treatments and medicines.

Theme 1: patient voice

The Government committed to putting the patient voice at the heart of its decision making and collaborate closely with patients and patient organisations. Commitments will be developed in consultation with patient representatives, with particular consideration given to ensuring representation from marginalised groups.

Theme 2: national and international collaboration

The Government emphasised on the need for international collaboration to support research and patient care, highlighting the Devolved Administrations and European nations as key partners.

Theme 3: pioneering research

The Strategy highlighted the July 2020 UK Research and Development Roadmap which set out an ambition to increase public funding for all research and development, including for rare diseases, to £22 billion per year by 2024/25 and to reach 2.4% of GDP by 2027. The Government said that it would continue to support and invest in innovative research for rare diseases and ensure that the outcomes are translated into frontline clinical care.

Theme 4: digital, data and technology

The strategy highlighted the benefits of telemedicine, video conferencing and digital resources in supporting patients, improving

efficiency and patient experience and supporting research. The government also highlighted the importance of data interoperability, easy sharing and access of patient data through rare disease registries.

Theme 5: wider policy alignment

The Government said that it was important that the additional needs of rare diseases patients such as housing adjustments and financial aid were reflected in wider policy development. The strategy also discussed a need for staff trained in genetics and genomics, workforce and long-term succession planning to consider the needs of patients in health and social care. The Government also said that the Framework would align closely with Genome UK (strategy for genomic healthcare) and other relevant strategies and policies.

2.2 Funding

In a [February 2021 PQ response](#) DHSC Minister Jo Churchill said that the Framework did not allocate any new funding:

The Government published a new United Kingdom Rare Diseases Framework in January 2021, which replaces the UK Rare Diseases Strategy. There is no new funding allocated in new Framework. However, the Government is committed to ensuring that the Framework is a success and will publish subsequent action plans that outline how the priorities and underlying themes in the Framework will be addressed. The details of the action plans will be released in due course.²⁵

The January 2021 Framework referred to the [UK Research and Development Roadmap](#), and said that the Government 'have set out an ambition to increase public funding for research and development, including for rare diseases, to "£22 billion per year by 2024/25"'.²⁶ Further detail of this investment was set out in the November 2020 Spending Review.²⁷

2.3 Next steps

Following the Framework's publication, the four UK nations are now set to develop action plans which will set out how the Framework's priorities will be addressed in each respective nation, taking into account the Framework's priorities and themes. Where possible, each nation will publish the action plans in 2021.

²⁵ [PQ 124728](#), 4 Feb 2021

²⁶ BEIS, [UK Research and Development Roadmap](#), published 1 Jul 2020

²⁷ HM Treasury, [Spending Review 2020](#), published 25 Nov 2020

3. Press articles

Please note: the Library is not responsible for either the views or accuracy of external content.

BMJ blog, 21 February 2021

[Having a rare disease is difficult- gaining access to good coordinated care shouldn't be!](#)

Guardian, 5 February 2021

[Up to 100 UK children a week hospitalised with rare post-Covid disease](#)

Independent, 3 February 2021

[Hospitals prepare for increase in children suffering rare disease triggered by Covid](#)

Telegraph, 10 January 2021

[Algorithm used to set vaccine priority order missed key vulnerable groups](#)

Science Business, News, 17 December 2020

[Rare disease patients at risk from no-deal Brexit](#)

HSJ, 18 September 2020

[Gene Therapy – will the NHS lead or follow?](#)

HSJ, 2 July 2020

[Exclusive: People with rare conditions 'deprioritised' by trusts during pandemic](#) *(subscription required)*

FT Special Report

[FT Health: The Future of Rare Diseases](#) *(subscription via House of Commons Library)*

The Parliament Magazine

[European Rare Diseases Supplement 2021](#)

A special supplement looking at European and global efforts tackling rare diseases.

4. Parliamentary material

4.1 APPG on Rare, Genetic and Undiagnosed Conditions

[The APPG on Rare, Genetic and Undiagnosed Conditions](#)

We work on a variety of issues that families and individuals with genetic conditions face. We aim to provide information, support families and influence the services needed by these patients. We also work to add patient voice into debates that matter to our community. Our work falls into different categories: reproductive techniques, diagnosis, medical research, healthcare and delivery, access to treatments and living and education.

The impact of Covid-19

The APPG held a virtual meeting to discuss the impact of Covid-19 on those affected by rare, genetic and undiagnosed conditions.

- [Minutes from the meeting on 4 November 2020](#)

4.2 PQs

[Rare Diseases: Health Services](#)

Asked by: Blackman, Bob | **Party:** Conservative Party

To ask the Secretary of State for Health and Social Care, what assessment he has made of the effect on patients with rare diseases of NICE's proposed changes to eligibility criteria for the Highly Specialised Technology pathway.

Answering member: Jo Churchill | **Party:** Conservative Party
| **Department:** Department of Health and Social Care

The National Institute for Health and Care Excellence (NICE) is reviewing the criteria used to decide whether a new technology should be evaluated through the highly specialised technologies (HST) programme as part of the review of its health technology evaluation methods and processes.

NICE is currently consulting on the guiding principles for the HST programme as part of the consultation on the case for change to its processes for health technology evaluation and expects to consult on the proposed HST criteria this summer. The purpose of reviewing the criteria is to make them clearer, more specific and the outcome easier to understand and more predictable for stakeholders.

09 Mar 2021 | Written questions | Answered | House of Commons | 160627

[Rare Diseases: Finance](#)

Asked by: Blackman, Bob | **Party:** Conservative Party

To ask the Secretary of State for Health and Social Care, what funding he plans to allocate to support the Rare Disease Framework.

Answering member: Jo Churchill | **Party:** Conservative Party
| **Department:** Department of Health and Social Care

The Government published a new United Kingdom Rare Diseases Framework in January 2021, which replaces the UK Rare Diseases Strategy. There is no new funding allocated in new Framework. However, the Government is committed to ensuring that the Framework is a success and will publish subsequent action plans that outline how the priorities and underlying themes in the Framework will be addressed. The details of the action plans will be released in due course.

04 Feb 2021 | Written questions | Answered | House of Commons | 124728

[Rare Diseases](#)

Asked by: Shannon, Jim | **Party:** Democratic Unionist Party

To ask the Secretary of State for Health and Social Care, what progress has been made on the UK Strategy for Rare Diseases during the covid-19 outbreak.

Answering member: Jo Churchill | **Party:** Conservative Party
| **Department:** Department of Health and Social Care

The Government is committed to improving the lives of those affected by rare disease and continues to implement the commitments made in the United Kingdom Strategy for Rare Diseases which expired at the end of 2020. Recent developments include progress with Rare Diseases Collaborative Networks and work to strengthen partnerships for the National Congenital Anomaly and Rare Disease Registration Service. Details on NHS England progress can be viewed in their November 2020 Implementation Plan Progress Report.

The new UK Rare Diseases Framework was published on 9 January, which provides the high-level direction for rare diseases and replaces the previous strategy. The Department, alongside NHS England, continues to consider the impact of COVID-19 on patients with rare diseases.

14 Jan 2021 | Written questions | Answered | House of Commons | 132918

[Rare Diseases: Diagnosis](#)

Asked by: Blackman, Bob | **Party:** Conservative Party

To ask the Secretary of State for Health and Social Care, what steps he is taking to reduce diagnosis times for people living with rare diseases; and whether the Rare Disease Framework will include targets on earlier diagnosis.

Answering member: Jo Churchill | **Party:** Conservative Party
| **Department:** Department of Health and Social Care

The Government recognises the importance of all patients being able to obtain a quick and accurate diagnosis. We plan to publish a new United Kingdom Rare Diseases Framework by the end of 2020 which identifies improving diagnostic times as a key priority. Measurable targets where

possible are not included in the UK Rare Diseases Framework but will be included in the subsequent nation specific action plans.

Advances in genomics and the commitment to incorporate whole genome sequencing into routine healthcare by the NHS Genomic Medicine Service are helping early detection of rare diseases. The recently published Genome UK Strategy outlines the Government's vision to help people live longer, healthier lives using new genomic technologies to diagnose and treat rare diseases.

10 Dec 2020 | Written questions | Answered | House of Commons | 124729

[Rare Diseases: Mental Health Services](#)

Asked by: Blackman, Bob | **Party:** Conservative Party

To ask the Secretary of State for Health and Social Care, whether the Rare Disease Framework will include measures to help people with rare diseases to access psychological support.

Answering member: Jo Churchill | **Party:** Conservative Party
| **Department:** Department of Health and Social Care

The Government plans to publish a new United Kingdom Rare Diseases Framework by the end of 2020. One of the underpinning strategic themes will be to ensure that the needs of rare disease patients are recognised in wider health and care services including mental health support. The appropriate steps to take to address these themes will be addressed in the nation specific action plans that will follow on from the Framework publication.

10 Dec 2020 | Written questions | Answered | House of Commons | 124727

[Rare Diseases](#)

Asked by: Morris, Anne Marie | **Party:** Conservative Party

To ask the Secretary of State for Health and Social Care, whether there is a timetable for devolved nations to agree an implementation plan to support the new rare disease framework.

Answering member: Jo Churchill | **Party:** Conservative Party
| **Department:** Department of Health and Social Care

All four United Kingdom nations have signed up to the development of a new UK Rare Diseases Framework, set for publication at the end of 2020. The Rare Diseases Framework will be followed by nation-specific action plans that will detail the steps each government will take to meet the shared priorities of the Framework. The devolved administrations have agreed to publish nation specific action plans within two years of the Framework publication.

09 Dec 2020 | Written questions | Answered | House of Commons | 123605

[Rare Diseases: Medical Treatments](#)

Asked by: Cameron, Dr Lisa | **Party:** Scottish National Party

To ask the Secretary of State for Health and Social Care, what steps his Department is taking to ensure that all rare disease patients in the UK have access to licenced treatments as soon as possible after approval by the European Medicines Agency.

Answering member: Ms Nadine Dorries | **Party:** Conservative Party
| Department: Department of Health and Social Care

The Medicines and Healthcare products Regulatory Agency has taken steps to recognise, for two years, future European Medicines Agency decisions for medicine licences including for rare diseases approved through the centralised authorisation procedure from January 2021 and provide United Kingdom licences with no additional assessment.

The Government has also established the Early Access to Medicines Scheme, which will continue to give patients with seriously debilitating conditions prompt access to medicines that do not yet have a marketing authorisation, when there is a clear unmet medical need.

24 Nov 2020 | Written questions | Answered | House of Commons | 114228

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