Accelerating access and reducing variation

Measuring the impact of the **UK Rare Diseases Framework** on access to care, services and treatment in England

Introduction

The publication of the UK Rare Diseases Framework in 2021 was a significant moment for individuals living with a rare condition in the UK: a joined-up programme of work signed up to by all four UK nations aimed at advancing care and better access to treatments for rare diseases.[1] The framework has driven actions across the four nations, with each country publishing an individual action plan setting out how they will deliver on its priorities, strengthening collaboration across Government departments and with the rare diseases community. Subsequent progress reports have followed, tracking progress made and areas where additional work is required.[2,3,4,5]

Originally scheduled to conclude in January 2026, the UK Rare Diseases Framework was extended for a further year in autumn 2025 until January 2027, to enable engagement with the rare disease community on a potential successor.[6] This creates a timely opportunity to evaluate what the Framework has helped to deliver, and what a future framework should include.

This report assesses the progress made in England, specifically looking at priority 4 of the framework: *'improve access to specialist care, treatment and drugs'*. This priority has been a key focus, accounting for over half of the 40 actions committed to across the action plans for England. However, after a strong start, momentum has stalled. Despite ongoing challenges in care for rare diseases and access to treatment, each action plan for England published after the first in 2022 has seen fewer actions committed to than the previous one, and some existing actions have been scaled back or paused due to funding constraints.

As a consequence, the rare disease action plans have not lived up to their full potential in improving patient care, and the rare disease gap persists as those with more common conditions remain more likely to access the latest treatments. The Government must commit to the creation of a new Framework that is designed to address the shortcomings of previous plans, and is accompanied by dedicated funding and robust accountability mechanisms. This Framework should build on existing progress and address the challenges in rare disease care identified in the previous rare disease action plans for England.

To support the Government in England as it develops proposals on what a future Framework might look like, this document:

- Analyses the effectiveness of the existing Framework, within the context of priority 4, with case studies of three actions that highlight both the opportunities of the Framework and the constraints that have affected delivery, notably a lack of ring-fenced funding and accompanying political accountability
- Sets out two key priorities for a new Framework to deliver:

    1. **Joined-up regulatory and access reform that addresses the challenges affecting the reimbursement of rare disease treatments**
    2. **Faster, earlier, widespread and equitable uptake for NICE-approved new medicines to ensure that all those living with a rare condition benefit equally from innovation in rare diseases**

Sobi welcomes the Government's continued engagement with the community in delivering the UK Rare Diseases Framework, both through formal structures – such as the Rare Diseases Forum and the Patient Advisory Group – and informally, applying feedback and learnings from the community in the delivery of new actions.

The Government has also announced further detail on a series of medicines access pilots as part of the UK-US Economic Prosperity Deal, which specifically reference rare diseases as a priority area of focus. Sobi welcomes these proposals, which represent an important opportunity to test new approaches to improving patient access to innovative medicines for people living with rare conditions. Whilst further detail is required on their scope and implementation, the pilots are a positive step in acknowledging and moving towards addressing some of the long-standing challenges set out in this report.[7]

Sobi looks forward to working with the Government and the rare conditions community to contribute to the development of a new, ambitious Framework and future action plans.

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Sobi is a global biopharmaceutical company unlocking the potential of breakthrough therapies. We work to ensure those living with the rare and specialised conditions we treat, and those that care for them, have access to our treatments, and the support and care they need to live the life they want and deserve. However, we know we cannot change the world of rare conditions on our own; accomplishing this requires strong partnerships with patients, partners, and stakeholders across the entire rare disease community.

Context:
The rare disease gap

Advancing timely and coordinated access to care, services and treatment is essential for people living with rare conditions. Individuals living with these conditions often face delayed diagnosis, poorly coordinated care, services spread across different specialist centres, and limited or no treatment options.[8] Only 5% of rare diseases have an available treatment option.[9]

Rare diseases are characterised by small patient populations and affect fewer than one in every 2,000 people.[1] It is estimated that half of those with a rare disease are children,[10] who face additional practical barriers to clinical trial participation.[11] Alongside this, there are broader challenges with clinical trial recruitment for rare disease treatments,[12] due to the small patient populations, which means that building an evidence base to demonstrate safety and efficacy is more difficult and costly.[9]



In **England**, rare disease treatments are assessed through the Single Technology Appraisal (STA) pathway, whereas treatments for ultra-rare conditions – affecting fewer than one in every 50,000 people – are assessed through the Highly Specialised Technology (HST) Pathway, providing they meet a strict set of criteria, including a statement that no more than 300 people in England can be eligible for the technology within its licensed indication, and that it must be the first licensed indication for that therapy.

The HST pathway has a higher cost-effectiveness threshold of up to £300,000 per Quality-Adjusted Life Year (QALY), significantly greater than the thresholds used in the STA process (£25,000-£35,000). Due to the strict eligibility criteria, as of January 2025, only 35% of 50 assessments for non-oncology orphan medicines were appraised via the HST pathway – with all qualifying treatments recommended. Since the introduction of refined routing criteria in March 2025, only one of the 10 products considered by the prioritisation board have been recommended for the HST pathway.[13,14,15]

The higher costs associated with research and development, combined with small patient populations, create higher treatment costs per patient. However, treatments for the many rare conditions that do not meet the HST eligibility criteria set by the National Institute for Health and Care Excellence (NICE) are assessed for cost-effectiveness according to the same criteria as more common conditions, despite the additional barriers that need to be overcome in developing and manufacturing treatments.[13]

This creates a *'rare disease gap'*: there is a lack of incentives for developing new treatments for rare diseases, and for treatments that are developed, it is very difficult to demonstrate cost-effectiveness under standard assessment routes. A recent ABPI survey found that 11 of 18 companies were planning to launch less than 75% of their rare disease pipeline in the UK over the next five years as a result of these challenges.[16]

England ranks 11th for the availability of non-oncology orphan medicines in European countries – behind similarly sized European markets, including Germany, Spain and Italy – all of which rank within the top five.[17]

The UK Rare Diseases Framework aims to improve the quality of care, services and treatment in each of the four nations. Each nation then decides how best to deliver on the priorities of the Framework, and there are differences across the UK in the assessment routes used to determine whether rare disease treatments should be made available to NHS patients.

In the UK, an estimated 3.5 million people live with a rare disease,[18] and people with rare diseases are three times more likely to be unemployed.[19] Better access to rare disease treatments has considerable untapped societal benefits.

A recent analysis estimated that should the UK become the third best country in Europe for access to orphan medicines, this could lead to 76,000 more patients receiving treatment – creating £1.1 billion in productivity benefits from reduced absenteeism and workforce participation.[19] Over 10 years, it is estimated that these productivity gains could be as high as £9 billion.[19] However, the current appraisal system does not allow for reflection of gains that reach beyond direct clinical outcomes and address benefits such as reduction in hospital stays, ability to stay in work and other related outcomes that have wider societal impact.

In **Scotland**, a more flexible approach to appraising rare disease treatments is taken by the Scottish Medicines Consortium (SMC). The SMC has adopted a Patient and Clinician Engagement (PACE) process to evaluating medicines to treat rare conditions, aimed at providing patients and clinicians with a stronger voice in determining the value of rare disease treatments.[20]



The SMC does not have a strict or defined willingness-to-pay threshold, meaning that the patient voice and clinical input hold greater significance during the appraisal process.[21]

The Innovative Medicines Fund (IMF) was created to help address some of these challenges by providing time-limited access to innovative non-cancer medicines, while further evidence is gathered. Medicines can be funded on the IMF for up to 5 years, before reappraisal by NICE.[22] However, due to a variety of challenges with its implementation, the IMF has not been used to its full potential and has failed to deliver on its stated objectives for accelerating access to innovative non-cancer treatments.

Contributing factors include a lack of certainty of long-term funding, as well as restrictive entry criteria that are difficult to meet for rare disease medicines.[23] In particular, rare disease treatments must be deemed to be 'plausibly cost effective' to be considered for the IMF, presenting a commercial barrier for manufacturers – especially when evaluated against STA routing criteria.[23] Even if a treatment qualifies for the IMF, with small patient numbers and slow progression,[24] five years is often too short a timeframe to gather data to demonstrate cost-effectiveness. Following the five-year period of managed access, if a medicine is then not recommended by NICE for routine commissioning, companies are required to continue supplying the medicine to patients free of charge.[25] As many rare diseases are lifelong,[26] manufacturers carry significant risk, which further disincentivises use of the IMF pathway.

In an analysis of IMF submissions between October 2024 and June 2025, only 1 in 14 cases was recommended – with the most common reasons for not proceeding with the IMF including a lack of company engagement – for reasons outlined above, including the additional risk taken on by manufacturers due to requirements to supply medicines free-of-charge – or recommendation via routine commissioning.[27]

As a result, there has been a massive underspend on budget. **In both 2023/24 and 2024/25, NHS England spent £2 million on drugs funded through the IMF, which equates to approximately 0.6% of the £340 million budget, allocated to the IMF.[22]**

The challenges with NICE methodology and the ineffectiveness of the IMF for rare disease treatments have led to increasing 'terminations' of appraisals – including non-submissions of licensed treatments to NICE, or termination of submissions to NICE by manufacturers. Termination rates have more than doubled since 2008, increasing from 8.95% (2008–2018) to 18.95% (2019-2025).[28] While there are many factors that influence non-submissions, a survey of rare disease companies found that NICE's approach was having an impact:[29]

- Since 2018, there have been 16 out of 64 medicines indicated for rare diseases and approved by the MHRA which companies decided not to submit to NICE for evaluation[29]
- Out of the 18 companies that responded, 11 reported that they expected to launch less than 75% of their rare disease pipeline in the UK over the next five years[29]

In 2026, the STA cost-effectiveness threshold increased to £25,000 to £35,000 per QALY (from £20,000 to £30,000) following the recent UK-US trade agreement.[30] Whilst this will benefit treatments for more common diseases, it will not impact the approval of more rare disease treatments. There is therefore a continuing need for policy action on medicines evaluation to address the rare disease gap.

Additionally, even when rare disease treatments in England are approved by NICE, uptake can be slow and variable across different geographies. Complex commissioning arrangements, challenges with clinical preparedness, a lack of targeted incentives for accelerated adoption, as well as a lack of system-wide accountability mechanisms, can all impact the speed at which patients can access these treatments.[31] The need to improve the measurement of uptake has been recognised by DHSC with the 2024 Voluntary Scheme for Branded Medicines Pricing, Access and Growth (VPAG) agreement including a commitment to continue the development of tools measuring the uptake of NICE recommended medicines across Integrated Care Boards (ICBs).[32] There are opportunities to continue to strengthen data collection and build on these commitments to measure uptake of approved medications for rare diseases against anticipated levels, and ensure action is taken where and when appropriate.

Spotlight on priority 4:
Evaluating the effectiveness of the England action plans in addressing the rare disease gap

The Government has acknowledged that addressing these challenges and the health inequalities they cause will require a system-wide approach, and notes that partners across the system are working to make improvements which will lead to accelerated development of, and access to, rare disease treatments.[2] **21 of the 40 actions introduced across the five England rare diseases action plans have focused on this priority as a result.** Of those, 12 have been recorded as delivered, nine are in progress, and one is an additional action introduced this year.[33]

Many of these actions have the potential to be important vehicles in addressing these challenges. However, the lack of ring-fenced funding allocated to delivery of the Framework has hindered progress. **Please see Appendix 1 for a table outlining the stage of completion of each of the 21 actions introduced.** In the table, we assign an indicative level of the likely impact each action could have, or has had, on individuals living with a rare condition. We consider five of the actions to have the potential to have genuine impact on patients. Eight actions are marked as likely to have low impact, either because they have been scaled back since first introduced or refer to actions that were already in place. Evaluation of the other eight actions is contingent on additional information on their impact being made available.

The following three actions highlight both the ambition of the England's action plans in delivering progress against priority 4, as well as the challenges that have hindered delivery.

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



Stage of completion: ***Concluded***
Level of impact on patients: ***Low***

This action, introduced in 2022, referred to how the changes introduced in the NICE methods review would support timely patient access to innovative medicines for people with rare diseases.[34] That included:

- The introduction of a disease severity modifier under the STA programme in place of the previous end-of-life modifier, informed by evidence that the public place greater value on treatments for severe diseases than treatments at the end of life
- The requirement that a summary of information for patients is introduced, as well as more flexibility to adapt consultation timelines for each appraisal

As with some of the other actions introduced across the action plans, the changes have not delivered the anticipated benefit for rare disease treatments. This is despite the assertion in the plan that as rare conditions are also severe, committees will be able to give additional weight to the treatment benefits for these conditions.

However, challenges around uncertainty, and undervaluing chronic, lifelong conditions in terms of QALY gain, remain.[35] As a result, few medicines have qualified – a recent analysis suggested that 72% of all appraised treatments do not qualify for the severity modifier,[36] which has led to a more limited impact on determining whether a treatment receives a positive recommendation. Companies have suggested that 34% of treatments that did not qualify for the severity modifier, would have previously qualified for the end-of-life criteria, making them eligible for a higher cost-effectiveness threshold.[36]

Whilst this action has now been marked as complete, it is important that NICE revaluates whether the severity modifier is working in practice, to support access to rare disease treatments.

ACTION 25: Review effectiveness of Early Access to Medicines Scheme (EAMS), Innovative Licensing and Access Pathway (ILAP) and the Innovative Medicines Fund (IMF) in supporting access to treatments



Stage of completion: ***Concluded***
Level of impact on patients: ***Low***

Action 25 was introduced in the 2023 England Action Plan, which stated that *'Over the coming year, NHS England, MHRA and NICE will review the impact of these initiatives, including their effectiveness in improving access to treatments for people living with rare diseases. This will aid in transparency, provide an understanding of how the schemes are working for rare diseases medicines, and identify any potential gaps or improvements'*.[37]

Subsequent action plans included an overview of the numbers of rare disease treatments going through these schemes, and it is welcome that the first three products selected in the first application round for the refreshed ILAP are for the treatment of rare diseases.

Since 2023, EAMS scientific opinions have been granted for only two oncology medicines, suggesting the programme remains ineffective in supporting access to rare disease treatments.[38] Similarly, only five medicines are currently being funded through the IMF, with three indicated for the treatment of rare diseases.[39]

The action does not include any detail on what measures will be introduced to address challenges identified with existing access pathways, despite a survey being run of companies on their experiences of using them. Whilst the 2024 Action Plan noted that a joint written response to the findings from the MHRA, NICE and NHS England would be published, this has not been made publicly available.

It is important that any future Rare Diseases Framework includes actions that go beyond describing how these schemes are currently utilised and clearly address identified barriers to utilisation, including a clear and regular process of review to assess delivery and a commitment to refine the schemes to make them more sustainable and useful in the long term.

ACTION 14: Map and measure the geographic spread of patients accessing highly specialised services



Stage of completion: ***Concluded***
Level of impact on patients: ***Medium***

This action was introduced in the 2022 Action Plan in response to considerable variability in whether – and how easily – people with rare diseases are able to get access to care, services and treatment.[34] It included an important exercise in monitoring overall uptake of treatments for rare diseases and assessing geographical variations in access. However, given the limited scope, which focused on highly specialised services, the analysis does not provide a complete picture of the adoption of treatments for all rare diseases.

The 2024 Action Plan shared the topline findings of this exercise, noting that there was equitable geographical access in 9 out of the 10 drugs that could be measured. However, this only represents a small proportion of the rare disease treatments that are available on the NHS, with 10 rare disease treatments being approved by NICE between 2022-24 alone. The study also found that geographic uptake was variable, with three of the 16 measured geographies having uptake below expected levels. Where previous analysis identified unexpected variation, providers have been asked to develop action plans to address inequities and variation.[40]

It is welcome that the original intention for this exercise to be repeated annually is now being followed up on, and a repeat of the analysis across all highly specialised services to assess progress and inform further action is planned for the 2027 to 2028 financial year. This positive step should be accompanied by:

- Publication of the full analysis completed so far, to support monitoring and accountability where uptake is lower than expected levels, as well as transparency in how uptake is measured
- Expansion of the exercise outside of highly specialised services, to help track uptake of a wider range of rare disease treatments
- A commitment to an annual audit, moving beyond 2027-28

Two priorities for a new UK Rare Diseases Framework

These case studies highlight the opportunities presented by the UK Rare Diseases Framework: a vehicle for joined-up working across government departments towards shared actions that aim to improve access to care, services and treatment for people living with rare conditions. However, they also point to the wider challenges that have led to a stalling of momentum to date.

Whilst the challenges faced across the system are well noted in the Framework and subsequent plans, with some positive ideas and plans highlighted, a lack of sustainable, long-term political commitment has led to restricted positive gain for patients.

Dedicated funding and a local infrastructure of accountability would demonstrate the political commitment to drive change. There is also an opportunity to capitalise on existing commitments in the policy environment.

With careful development and implementation, supported by a successor Framework, these could help address existing access challenges. These include:

- The new, flexible regulatory framework for rare diseases by MHRA, aimed at accelerating access to rare disease treatments[9]
- The 2025 UK-US trade agreement's commitment to double spending on new medicines[41]
- The implementation of the new Single National Formulary – if driven by the objectives of priority 4 of the UK Rare Diseases Framework, this could be a vehicle to address variation and drive uptake of new rare disease treatments.[42]

Building on these opportunities, we have set out two core solutions that we believe a new Framework must deliver.

1. Joined-up regulatory and access reform that delivers on the challenges affecting the reimbursement of rare disease treatments

The current UK Rare Diseases Framework has contributed to strengthened engagement between DHSC, MHRA and NICE. For example, the Framework has coincided with the introduction of a streamlined MHRA-NICE aligned pathway – delivering regulatory approval and licensing decisions in parallel.[43] ***However, for this to be successful, closer collaboration and integration between DHSC, MHRA and NICE is required.***

There is now disparity across the organisations about how they are addressing the unique challenges associated with ultra-rare conditions. The MHRA's draft regulatory framework is designed for ultra-rare conditions, stating it is *'for therapies for traditionally described rare conditions with a prevalence of typically 1 in 50,000 or less in the UK.'*[18] This prevalence is typically the definition of an ultra-rare condition and equates to a maximum of 1,200 people in England. By developing this draft framework, the MHRA recognises the specific challenges related to evidence generation within these conditions. The first of NICE's routing criteria for the HST programme shares this definition, with an eligible treatment for a condition needing to have a point prevalence of 1 in 50,000 or less in England.[14] However, NICE also includes a subsequent criteria that no more than 300 people in England can be eligible for the technology in its licensed indication, for it to be assessed via the HST – limiting eligibility even within ultra-rare conditions.[14] As a result, treatments for rare conditions affecting 301 or more people face a cliff edge and are assessed through the same route as treatments for common conditions that may affect millions of people, despite facing similar evidence and pricing challenges.

A review of the criteria for the HST pathway to enable all ultra-rare conditions to be assessed via the HST route should be undertaken. This would support a more equitable approach for people with ultra-rare conditions and would also bring the reimbursement system into closer alignment with the rare diseases regulatory framework.

As noted earlier, Action 25 of the England Rare Diseases Action Plan committed to reviewing the effectiveness of Early Access to Medicines Schemes (EAMS) in supporting access to treatments.[2] As part of the EAMS submission process, companies must first apply for a Promising Innovative Medicine (PIM) designation before obtaining a scientific opinion, which poses a further resource strain, which can particularly impact smaller manufacturers. However, currently less than half of PIM submissions obtain a scientific opinion due to the bureaucratic hurdles and significant data demands,[44] and even if access through EAMS is achieved, there is no advantage of impact on the overall time to reimbursement.[45] However, despite clear challenges in implementation Action 25 has not yet led to any changes in the operation of EAMS. ***Greater high-level commitment is needed to recognise the need for better access to rare disease treatments through EAMS and to streamline the process accordingly.***

Prior to a managed access agreement being finalised, it is also important there is more effective scheduling of committee meetings. This includes greater alignment on data requirements – for example through the acceptance of surrogate endpoints by MHRA and NICE; as well as coordination between MHRA and the administrations in the devolved nations, so that duplication in engagement between manufacturers and the MHRA and then NICE is reduced.

We recognise the challenges that NICE faces in assessing rare disease treatments and understand that developing a sustainable and effective solution to bridging the rare diseases gap will require cross-sector collaboration. ***The Government's recent commitment to double medicines spending by 2035 to 0.6% of GDP,[41] presents an important opportunity to consider reforms to how NICE's methods and processes are applied to rare disease treatments*** such as introducing greater flexibility in the QALY approach to reflect relevant gains that reach beyond direct clinical outcomes and address benefits such as reduction in hospital stays, ability to stay in work and other related outcomes that have wider societal impact.

The EFPIA Patient's W.A.I.T Indicator survey, which reports annually, not only highlights the rate of availability for new treatments, including a section on non-oncology orphan treatments, it also now outlines time to availability of new treatments across 36 countries.[17] ***We would also recommend setting an explicit goal for the UK to rank among the top three top three in Europe in terms of availability of non-oncology orphan medicines, within the survey.***

Recommendations

- Strengthen cross-agency collaboration between DHSC, MHRA and NICE to ensure the successful implementation of the 2026 aligned pathway and rare disease regulatory reforms.
- Review the HST criteria to enable equitable processes for all ultra-rare conditions.
- Reduce bureaucratic hurdles within EAMS that discourage manufacturers from applying.
- Seek greater alignment between MHRA and NICE on definitions and evidence expectations to improve coordination and streamline the pathway from early access to reimbursement.
- Reform how NICE's methods and processes are applied to rare disease treatments.
- Define an explicit goal to improve England's ranking for the availability of non-oncology orphan medicines in the EFPIA Patient's W.A.I.T Indicator survey, to rank among the top three in Europe.

2. Improving uptake to ensure that approved medicines are reaching patients

Once rare disease treatments are approved, as outlined in the case study of action 14 of England's action plans, uptake can be inequitable across the country – meaning people with rare diseases are not benefiting consistently from the promise of new innovations. The NHS has developed an Innovation Scorecard that monitors the uptake of NICE-approved medicines. The reporting helps identify variation in uptake at a regional, ICB, and trust level, which can then be acted upon. The scorecard monitors 234 medicines, organised into 18 medicine groupings.[46] However, there is not consistent incorporation of rare disease treatments into the scorecard, making it difficult to use as a mechanism to assess uptake. ***There is a need for a redesigned Innovation Scorecard to better account for rare disease treatments and enable the measurement of uptake of innovative treatments.***

The 10 Year Health Plan has also promised the creation of a Single National Formulary (SNF), which aims to put an end to the postcode lottery created by local formularies, and ensure patients across the country are getting access to the medications that they need.[47] Although we support the aim to drive rapid and equitable adoption of innovations, there is a concern that sequencing of products in the formulary beyond recommendations made by NICE may lead to restricted patient access to innovative treatments. The Life Sciences Sector Plan highlighted the SNF's potential to reduce bureaucracy, freeing up healthcare staff and administrative time, suggesting the SNF will be driven by innovation.[48] ***As the SNF is developed, there is an opportunity to embed reporting of uptake to support regional accountability and reduce variation, while ensuring patients are given access to the treatments that they need.***

Recommendations

- DHSC/NHSE should establish accountability mechanisms to strengthen action by trusts and ICBs with lower than anticipated uptake of treatments for rare diseases and reduce geographical inequities in access to treatment.
- DHSC/NHSE should redesign the Innovation Scorecard to include reporting of uptake for rare disease treatments, to facilitate identification of regions, ICBs and trusts with low uptake and strengthen transparency.
- DHSC/NHSE should ensure the Single National Formulary is designed to expand access to innovative medicines in their first 5 years following NICE approval. This should include treatments that can drive efficiencies in the delivery of patient care, and should embed uptake reporting requirements into the design to strengthen regional accountability and reduce unwarranted variation, while maintaining patient and clinical choice.

Conclusion

The Framework is due to conclude at the end of this year, creating an important window to review its effectiveness and understand areas where it can be strengthened, to inform what comes next. Whilst important progress has been delivered, the potential of the Framework remains largely untapped.

To support the Government as it engages with the community to inform what comes next, Sobi has published this report to set out what those solutions should involve. These include:

1. **Joined-up regulatory and access reform that addresses the challenges affecting the reimbursement of rare disease treatments**

2. **Faster, earlier, widespread and equitable uptake for NICE-approved new medicines to ensure that all those living with a rare condition benefit equally from innovation in rare diseases**

The UK-US Economic Prosperity Deal, combined with the implementation of a Single National Formulary, present a real opportunity to translate priority 4 into actions with impact, building on the important work completed to date. Sobi welcomes the Government's commitment as part of that Deal to pilot new approaches to managed access, which pulls out rare diseases as an area of focus, and to explore how the productivity benefits of new medicines can be reflected in medicines assessment.[7] Subject to further detail on their implementation, these pilots have the potential to address a number of the challenges identified in this report, particularly around managed access for rare disease treatments and better recognition of the wider societal value that innovative treatments can deliver.

The next Framework should build on these opportunities and should be accompanied by clear funding and accountability mechanisms to ensure that opportunities are not missed to address the challenges set out in this report and improve outcomes for people living with a rare condition.

Appendix 1:
Evaluating the 21 priority 4 actions introduced in the 2022-2026 England rare diseases action plans[33]

Key: Level of impact on patients

- ***High*** This is a new action that has had a demonstrable patient benefit
- ***TBD*** Not enough information available to determine the impact of the action
- ***Low*** The action is not new and/or has not had a discernible impact on patients

ACTION	Details	Stage of completion
ACTION 11: Support rapid access to drugs for patients with rare diseases	Past progress reports indicate a 'preparedness template' with industry has been delivered to develop a shared understanding of the service implications for new drugs, though detail on its use and impact is lacking.	Concluded
ACTION 12: Develop a strategic approach for gene therapies and other ATMPs	This is an internal-facing action looking at strengthening horizon scanning of new ATMPs expected to be introduced, to help understand expected service implications of delivery. Whilst important, it would be helpful for more information to be published on what changes have been introduced.	Concluded

ACTION	Details	Stage of completion
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	As set out above, no actions have been taken in response to this review, despite the fact changes to NICE's methods are not working in the assessment of rare disease treatments.	Concluded
ACTION 14: Monitor overall uptake of drugs for patients with rare diseases and map geographical access to those drugs	This is an important action that sets out the extent of variation in access to NICE-approved treatments. It is welcome that NHS England has committed to repeating the analysis, and it is important it is replicated beyond highly specialised services.	Ongoing
ACTION 15: Map the rare disease research landscape to identify gaps and priorities for future funding	The 2023 National Institute for Health Research report set out important investment in rare diseases research, supporting the establishment of the UK Rare Disease Research Platform.	Concluded
ACTION 16: Reduce health inequalities in NHS highly specialised services	Whilst important action has been taken on health inequalities through the England action plans, the impact of this action has been less easy to define – including considering health inequalities at HSS annual clinical meetings, in service development and commissioning decisions.	Concluded
ACTION 22: Improved 'findability' of people living with rare diseases using National Congenital Condition and Rare Disease Registration Service	Several measures have been introduced as part of this action, including a new web-based self-reporting system for public testing, a formal rare disease data set, with input from clinical experts, and the publication of the first Rare Disease Prevalence report.	Concluded
ACTION 23: Improve understanding of the impact of NHS England's specialised service commissioning activities on rare disease patients	Action taken here included a review of NHS England's patient impact assessments (PIAs), which involve factual summaries of what the impact of living with a particular rare condition is like on patients and their carers. It is unclear what the objectives of this review were and what the outcome has been. As a result of the NHS England merger into the Department of Health and Social Care, it is also unclear what the future of many of the activities in scope of this action will be.	Concluded
ACTION 24: Establish a highly specialised services (HSS) programme board and strengthen role of NHS England in commissioning wider services	This is an internal-facing action for NHS England that is difficult to assess until more evidence is available on the impact of commissioning changes of specialised services.	Concluded

ACTION	Details	Stage of completion
ACTION 25: Review effectiveness of Early Access to Medicines Scheme (EAMS), Innovative Licensing and Access Pathway (ILAP) and the Innovative Medicines Fund (IMF) in supporting access to treatments	The results of this review have not been published, and it is not clear what actions will be taken to address the findings. Whilst Sobi welcomes the fact rare disease treatments have been prioritised for the refreshed ILAP, it is not yet clear whether it will address the challenges affecting the original ILAP. Challenges with EAMS and the IMF are set out in this report.	Concluded
ACTION 26: Registration of national data for exemplar rare genetic conditions which cause an inherited predisposition to cancer	NHS England's National Disease Registration Service (NDRS) is establishing the National Inherited Cancer Predisposition Register (NICPR), to provide a consistent national data set for all hereditary cancer predisposition syndromes currently tested in the NHS. The aim is for the register to contribute directly to screening pathways where national programmes exist.	Ongoing
ACTION 27: Improving the Be Part of Research Platform	This action involves redeveloping and improving the Be Part of Research platform, to enable the service to better meet the needs of research into rare diseases. However, despite being introduced in 2023, progress has been slow; the 2026 progress report notes 'significant improvements are unlikely to be implemented until 2027'.	Ongoing
ACTION 28: Develop a plan to include rare diseases in NHS England's Core20PLUS5 framework	The objective to include rare diseases in the Core20PLUS5 framework (designed to reduce healthcare inequalities at both national and system levels) is an important acknowledgement of the specific inequalities people living with a rare condition face. Evidence gathering is now complete and will now be presented to regions.	Concluded* See action 40 for more information
ACTION 29: Commission portfolio level evaluation of England's rare diseases action plans	This is a general evaluation and monitoring exercise that is not specific to priority 4.	Ongoing
ACTION 33: Develop a funding mechanism that sufficiently incentivises centres to undertake whole body scans on individuals with rare conditions resulting in a genetic predisposition to developing cancer prematurely	NHS England has launched a procurement exercise to identify future providers of adult whole-body magnetic resonance imaging (MRI) scanning for designated cancer-predisposing conditions which carry a high risk of developing cancer at an early age. Subject to NHS England governance, successful providers are expected to be operational from 1 May 2026. It is currently too early to determine the effectiveness of this exercise.	Ongoing
ACTION 34: Review the effectiveness of the Innovative Devices Access Pathway (IDAP) pilot in supporting access to medical devices, including diagnostics, for people living with rare disease	No update was provided on this action in the 2026 Action Plan, though it has been marked as complete. The action is also not specific to rare diseases; none of the eight technologies selected to date are relevant to rare diseases.	Concluded

ACTION	Details	Stage of completion
ACTION 35: Publish and disseminate a health inequalities toolkit, for highly specialised services	This is an important new action, that has seen the toolkit disseminated among providers to enable them to identify and address health inequalities. However, it is too early to know if the toolkit will translate into action, with no detail provided on how accountability will be introduced to ensure it is used.	Concluded
ACTION 36: Map and measure the geographic spread of patients accessing highly specialised services	This action includes new detail to action 14, covering additional data and outcomes. Whilst action 14 is marked as having a high level of ambition, and Sobi welcomes the commitment to repeat and expand the analysis, this had already been noted as NHS England's intention in previous action plans.	Ongoing
ACTION 38: Reform of clinical trials regulations	This action, whilst important, was underway outside of the infrastructure of the UK Rare Diseases Framework.	Concluded
ACTION 39: Operational framework for individualised therapies	NHS England is establishing a national operating framework setting out how NHS-commissioned individualised therapies for rare diseases will be delivered to patients. It is anticipated that the plan will be finalised in March 2026.	Ongoing
ACTION 40 (new action): Addressing health inequalities for rare diseases through Core20PLUS5	Work as part of this action had already been set out in previous action plans, linked to action 26.	Ongoing

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