Refreshing the UK Rare Diseases Framework

Reflections on progress to date and lessons for the future

Introduction

The publication of the UK Rare Diseases Framework in 2021 was a significant milestone: a joined-up statement of intent to improve the lives of people affected by a rare condition in the UK committed to by all four UK nations. It was published following a national conversation on rare diseases.

The Framework identified four priorities to deliver improvements in care for people with rare conditions and committed to progress being made by each UK nation through nation-specific action plans. The four priorities are to:

1. Help patients get a final diagnosis faster
2. Increase awareness of rare diseases among healthcare professionals
3. Support better coordination of care
4. Improve access to specialist care, treatments and drugs

Advances have been made in every UK nation. Individual plans were published in 2022 setting out specific actions to be taken to deliver on the Framework's four priorities. England and Northern Ireland have subsequently published two papers outlining progress made, Wales published a refreshed plan and progress report in January 2024 and Scotland published its first progress report in August 2024. Throughout, the nations have worked constructively and collaboratively with the rare conditions community.

Despite the optimism which followed the publication of the action plans, barriers across all four nations have limited progress. Whilst the circumstances for this vary by UK nation, they can all be attributed in part to the absence of ringfenced funding to accompany the delivery of the Framework, and resultant limited ambition and accountability within governments – though the timing of the Framework's publication in January 2021 during the COVID-19 pandemic and a national lockdown did, naturally, limit initial progress.

The Rare Diseases Framework runs until 2026, and the Department of Health and Social Care (DHSC) has commissioned an evaluation of its effectiveness in England. The Specialised Healthcare Alliance (SHCA) has collected the views of its members through a survey to inform the development of this report and support our own assessment of the effectiveness of the Framework to date, across all four UK nations, to inform discussions around what a successor to the Framework should look like. The report has been drafted in partnership with Genetic Alliance UK.

Together, we are calling for the UK Rare Diseases Framework to be refreshed, renewed and recharged after 2026.

Overarching views on the UK Rare Diseases Framework

We surveyed the SHCA's members and supporters to collect quantitative and qualitative feedback on the Framework, and the results form the basis of this report. 12 responses were received, 9 from patient groups and 3 from corporate supporters, though not all respondents answered each question. Alongside the quantitative results, quotes from the qualitative survey questions are interspersed throughout this report, highlighted in blue. Please see Appendix 1 for a more detailed overview of the survey and its respondents.

Support for the four priorities

Our survey found broad support for the four priorities of the UK Rare Diseases Framework, reflective of the strong engagement held with the community to inform its development. **11 of the 12 respondents felt the four priorities are the right ones for patients affected by a rare condition**.

The Framework also included a set of underpinning themes: patient voice; national and international collaboration; pioneering research; digital, data and technology; and wider policy alignment. These themes have played a key role in supporting wider momentum across government for delivering advances in care for people affected by a rare condition and have led to some examples of key progress, notably the development of rare disease registries across the UK nations.

Engagement with the rare conditions community

The SHCA and Genetic Alliance UK have welcomed the DHSC's meaningful engagement with the rare conditions community. The UK Rare Diseases Forum, which includes both the SHCA and Genetic Alliance UK, has created an important feedback loop between patient groups, other external advocates and each of the four nations. The Forum is UK wide and holds four quarterly meetings a year; representatives of each of the four UK nations present on a rotating basis at one meeting a year on progress made against their respective action plans.

The establishment of the Independent Advisory Group framework, meanwhile, has provided the community with the opportunity to establish working groups on specific issues facing rare conditions patients, and provides a further feedback loop between stakeholders and policy teams across the four nations. The Quality Standards Advisory Group, for example, is supporting the development of a quality standard for rare disease, to improve equity of care for rare diseases across all four UK nations.

The chair of the UK Rare Diseases Forum feeds into the UK Rare Diseases Framework Board alongside three representatives of the rare conditions community. All nations' action plan delivery groups include representation and have welcomed external speakers from the community. Topic specific working groups such as non-genetic rare conditions and mental health have also involved members of the community, whilst England's Action Plan development process includes a Patient Advisory Group phase and consultation with minority groups through Breaking Down Barriers.

Our members reflected on how the Framework has created important momentum across government for the delivery of actions specifically focused on rare conditions.

> *"The Rare Diseases Framework has been an effective vehicle for convening and galvanising stakeholders from across the ecosystem to come together to address a number of issues and inequalities that impact outcomes for people affected by a rare condition. It's important that this work can continue beyond 2026."*

The Framework and its governance arrangements have helped to facilitate multi-stakeholder input on the design and delivery of specific actions, drive instances of closer coordination between and across disparate actors, and provide impetus, transparency and accountability on some projects that would not have existed otherwise."

However, we also heard that that there is more to do in providing rare condition patient groups with the opportunity to input into action plans: whilst more than half (6 out of 11) felt that processes for patient groups to shape and share feedback on individual nation action plans has been satisfactory, 5 found them to be very or somewhat unsatisfactory. One of those members felt more emphasis was needed on how future plans can be co-produced and subsequent actions informed by external stakeholders, such as rare condition charities.

It is important that opportunities for involvement in, and engagement with, each of the four nations' rare conditions delivery teams are made more visible, so that the rare conditions community are aware of what opportunities there are to feed into the development of future action plans.

Barriers to delivery

Despite the significant efforts of rare conditions teams across the four nations, ultimately progress in delivering on the four Framework priorities have been affected by:

Funding: Most significantly, the publication of the Framework was not accompanied by funding allocations to support its delivery in any of the four nations. The need for actions to either be cost neutral or delivered at low cost has limited their impact. Recent progress reports published by all of the devolved nations reflected on how financial constraints make it difficult to maintain progress.

Ambition limited by lack of capacity and accountability: As a result of the absence of funding, actions previously committed to have often been scaled back or delayed – with rare conditions teams in each nation not receiving sufficient resource and support from their respective governments to enable progress.

The COVID-19 pandemic did impact the scope of actions committed to as part of initial action plans, with resources focused on the pandemic response, though challenges around capacity and accountability have remained.

Owing to these challenges, 8 of the 12 respondents to our survey do not feel that the UK Rare Diseases Framework has so far lived up to its potential as a vehicle for improving access to care, services and treatment for people with a rare condition in the UK.

"Progress to meet the aims of the Rare Diseases Framework in each of the devolved nations has been limited due to a lack of direct funding to improve services for patients."

The remainder of this report reflects on progress that has been made in each UK nation across the four priority areas, before considering how the above challenges have affected progress. Specific examples of best practice and challenges are brought out from each of the four nations, based on both survey feedback and the SHCA Secretariat's own analysis of each nation's action plans. The report then concludes with reflections on what a successor to the UK Rare Diseases Framework should look like

after 2026 and how DHSC and devolved administrations can ensure it is informed by patient and community involvement.

Analysis of the four nations' action plans

About the action plans

Each of the four nations have taken differing approaches to producing their individual action plans and consulting with the community, and some nations' governance arrangements are more developed than others:

In England, actions plans are sponsored by DHSC and overseen by the Rare Diseases Framework Board. The Board holds two formal meetings a year.

In Scotland, action plans are sponsored by the Scottish Government and led by the Rare Disease Implementation Board.

In Wales, action plans are sponsored by NHS Wales and led by the Rare Disease Implementation Network, which meets quarterly to oversee the implementation of the plans by health boards.

In Northern Ireland, actions plans are sponsored by the NI Department of Health and led by the NI Rare Diseases Implementation Group. The group meets quarterly with five individual working groups to drive forward actions.

The overarching UK Rare Diseases Forum meets quarterly to provide updates on progress made across each nation with the wider UK rare conditions community.

"The feedback process for the England Rare Diseases Action Plan is clear and the timelines are well defined with opportunities to engage in both the Patient Engagement Group meetings and in the yearly written feedback forms provided by Genetic Alliance UK. This process was not in place for the devolved nations, meaning that it was difficult to input into the individual nations' action plans apart from sharing the updates with our communities."

Individual action plans are also at different stages of development:

34 actions have been committed to across three action plans. Development of the 2025 Action Plan is ongoing; DHSC have outlined its focus will predominantly be on the delivery of existing actions rather than commitment to new ones.

18 actions have been committed to, with a progress report published in 2024.

42 actions are being progressed against, including 12 new actions introduced in 2024 as part of a refreshed Action Plan.

14 actions have been committed to, with two progress reports since published. The latest progress report highlighted the significance of the restoration of power sharing in Northern Ireland in February 2024 in creating new opportunities to reassert the importance of the rare conditions agenda.

Please see Appendix 2 to this report for a list of all action plans and subsequent progress reports.

Priority 1: Help patients get a final diagnosis faster

England

As part of the 2022, 2023 and 2024 England Action Plans, seven actions are aimed at helping patients get a diagnosis faster. Of those:

- Two have progressed well and led to direct improvements for patients
- Four are ongoing, with some delayed against initial timelines
- One is behind schedule as a result of resource constraints

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Whole genome sequencing for rare diseases: Over 200 rare conditions are included in the initial list of the Generation Study, which will pilot whole genome sequencing of newborns in the NHS to enable faster diagnosis of rare conditions. Milestones committed to in the 2022 Action Plan have been achieved. **Transfer whole genome sequencing data to the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)**: Two initial conditions have been included before expansion to other indications.	**Improve the evidence base to support the UK National Screening Committee (NSC) make robust decisions about newborn screening for rare conditions:** Progress has proved slower than anticipated and the latest Action Plan stated that resourcing constraints have delayed progress against some action-specific monitoring and evaluation targets. One member shared with us frustrations around delays in decision-making for newborn screening of rare conditions, with only tyrosinemia recently added.
Pilot new approaches for patients with undiagnosed rare conditions: A service model has been agreed and funding secured to pilot two Syndrome Without a Name (SWAN) clinics in England, one for children and one for adults. Whilst this is welcomed, progress in rolling this work out has been slower than in Wales – as set out later in this report.	**Commission research on how best to measure the diagnostic odyssey:** There have been frustrating delays in commencing this work, with a research partner only recently contracted. It is important that engagement is held with patient groups once initial findings of this work are disseminated,

	building on the findings of the SHCA's own research on delays to diagnosis.

Scotland

The 2022 Scotland Action Plan set out three actions to improve the early diagnosis of rare conditions: the implementation of Genome UK, newborn screening, and expanding the functionality of the Congenital Conditions and Rare Diseases Registration and Information Service for Scotland (CARDRISS). All three are ongoing.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Genome UK: This action helped to support the publication of Scotland's first ever genomic medicine strategy, *Genomics in Scotland: Building our Future*, in April 2024. New Scottish Genomics Test Advisory Groups have been launched for both Cancer and Rare and Inherited Disease. SHCA members hope this will help to enable progress in the breadth and equity of access to genomic testing for rare conditions with a genetic cause in Scotland.	**Newborn screening**: The only update provided in the recent Action Plan was that Scotland continues to participate in the UK NSC. As in England, SHCA members are frustrated by delays to newborn screening across the UK, and the impact this then has on families. The next progress report must include specific actions to address delays to newborn screening in Scotland, including how it will pilot whole genome sequencing of newborns in the NHS and capture learnings from screening carried out in England.

Wales

Nine actions were introduced in Wales to deliver earlier diagnosis across six themes, from improving access to genetic testing to supporting the research ecosystem. Of these nine actions, four can be marked as complete with five ongoing.

In the 2024 progress report, four additional actions were committed to, focused on supporting research activity, increasing genomic testing within Wales and improving diagnosis and care for non-genomic conditions.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Increase access to whole genome sequencing testing for rare conditions: There has been welcome progress against this action, with increases in testing across three conditions analysed. SHCA members would now like to see an increase in the conditions tested for in Wales.	Whilst the actions around genomic testing in the initial plan are welcomed, SHCA members would like to see additional actions introduced as part of the next progress report. Whilst testing is an important part of improving rates of early diagnosis, there should also be a greater focus on areas such as newborn screening and improving mental

Health Care Research Wales are also supporting investment in rare conditions research through Wales Gene Park. The latest progress report sets out increases in participants entering research studies across different rare conditions.	health support through the diagnostic journey.

Northern Ireland

Three actions against this priority were committed to in the 2022 Action Plan, which are all ongoing: establish a national online Rare Disease Information Hub; agree a new Genomics Partnership model to deliver against the UK Genome Strategy; and review newborn screening.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Develop a Rare Diseases Information Hub: Progress on this action has been good, with funding now secured to develop the resource. It is important the relevant working group leading it works with the patient community to consider what resources will help to deliver on the Framework priority.	**Review newborn screening**: As with the other UK nations, members are keen to see accelerated progress against this objective. The most recent progress report did not include an update against this action, while the 2023 report noted only that Northern Ireland is participating in the wider national newborn screening research study.

Priority 2: Increase awareness of rare diseases among healthcare professionals

England

DHSC has committed to completing six actions over the course of the UK Rare Diseases Framework, including two new actions around the genomics workforce. There has been welcome progress in this area, with strong progress against all six of the actions.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
The commitments made in the England action plans have helped to enable significant progress in strengthening the genomics workforce, including through: ● **GeNotes:** a new educational resource that enables healthcare professionals to access new information on genetic and rare conditions, with a range of rare conditions included in its knowledge hub. It also allows NHS staff to access information about the Generation Study.	Members have welcomed the greater emphasis placed recently on increasing awareness of rare conditions without a genetic cause, which was included as a focus area in the most recent England Action Plan. Going forwards, it is important stand-alone actions are committed to non-genetic conditions, in addition to ongoing work to address them in existing policy for genetic conditions. Our survey findings highlighted how more work is needed to improve the recognition and

• **The genomics communication skills resource**: committed to in 2024, healthcare professionals have already fed back on the proposed online modules being developed.	management of rare conditions in non-specialist settings. Whilst work is being carried out developing the Rare Disease Education Hub, a dedicated online portal for rare conditions, it is important there is a focus on ensuring this reaches health professionals in non-specialist settings.

Scotland

The 2022 Scotland Action Plan committed to the delivery of five actions to improve awareness of rare conditions among healthcare professionals. Of these, strong progress has been made on one, whilst the other four remain ongoing.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Improving information about rare conditions on NHS Scotland platforms: The Rare Disease Implementation Board has overseen the development of the first dedicated Rare Conditions Information Hub on NHS Inform. The Hub includes important resources for individuals and healthcare professionals, including Genetic Alliance UK's Rare Resources Guides and information from the Office for Rare Conditions (Glasgow). Genetic Alliance UK and the Office for Rare Conditions were also supported to conduct a survey with healthcare professionals to identify their information needs, the results of which will inform the development of future resources.	**Optimise Rare Disease Day:** SHCA members would like to see more ambition here. The progress report highlights how the day is marked by the Scottish Government through engagement events, though it is important going forward this includes written updates, shared externally, on progress made against the four priorities of the Framework. SHCA members would also like to see greater opportunities for third sector organisations to raise awareness of rare conditions in NHS Scotland and would value evidence of work undertaken to date being communicated in the next progress report.

Wales

The 2022 Plan introduced 11 actions against this priority across three themes: a lead clinician for rare conditions, education and shared learning and improving awareness of rare conditions with data. Two new actions have been introduced for 2024 on clinical education and the development of rare conditions registries.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Welcomed actions are included in the Action Plan around increasing awareness of rare conditions, including in relation to genomics and genomic counselling. **Expand the Congenital Anomaly Register and Information Service (CARIS) team's work on rare conditions:** Progress has been good here,	Many of the actions committed to in the 2022 Action Plan are accompanied by very succinct text in the progress report, making it hard to assess what steps have been taken in response to the action and any deliverables met.

with work underway looking at how rare conditions registers can be improved to drive improvement in pathways of care.	Additionally, whilst all work to celebrate Rare Diseases Day and raise its profile is welcomed, it is important it is accompanied by tangible steps being taken to deliver on the Framework's priorities – alongside the communications activity referenced in the plan.

Northern Ireland

Three actions were committed to in the Northern Ireland Action Plan against this priority which remain ongoing: develop a Northern Ireland rare diseases registry, improve education and training, and raise awareness of rare conditions across a range of communities.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Education and training: Progress has been strongest here - a Rare Disease Society has been established at the School of Medicine, Ulster University, in collaboration with the NI Rare Diseases Partnership, which has helped to collaborate on healthcare education and training, from undergraduate education through to continuing professional development.	**Northern Ireland rare diseases registry**: SHCA members are disappointed by the absence of any government funding to support its development. Latest updates include a note on challenges created by limited resources and single-year funding periods for planning and investing in long-term strategic projects. Against this backdrop, the progress made by the NI Department of Health and NI Rare Diseases Implementation Group to secure external research funding to support it is welcomed, with incidence data now having been established for several rare conditions and congenital anomalies. However, it is noted that the lack of government funding will impact on the data available and may limit understanding of the prevalence and geographical spread of people affected by a rare condition in Northern Ireland and the distribution of services.

Priority 3: Support better coordination of care

England

Fewer actions have been committed to by the government and delivery partners on this priority than other areas, which may reflect the complexity of the challenge and the need for more evidence before progress can be made; DHSC has acknowledged this as a key area of focus going forwards. Only four actions have been committed to across the three action plans, including one additional one in 2024. Of those four actions:

- One has been completed

- Two are ongoing
- One has been subject to delays

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Implement networked models of care for patients with rare conditions: Initial networked models are being rolled out for inherited metabolic disorders and amyloidosis, with strong early progress made across both. This will then inform possible future models that could be rolled out in other areas. **Inclusion of a definition of coordination of care in all new and revised service specifications for specialised services:** Further progress has been made on the inclusion of a definition of coordination of care in all new and revised service specifications for specialised services, with updates currently being carried out by NHS England. It is important this is accompanied by accountability for individual services to demonstrate to NHS England how they are implementing effective coordination of care.	**Commission research to better operationalise coordination of care within the NHS**: The SHCA and Genetic Alliance UK welcome this commitment, as people with rare conditions often see a wide range of healthcare professionals, both specialist and non-specialist, from across different settings. However, as with other research commitments, the process for contracting out this research has proved lengthy – with initial findings only expected next year. It will be important that the outcomes of the research are acted on swiftly, supporting decisions on how effective care coordination can be implemented across the NHS.

Scotland

Six actions were included in Scotland's Action Plan on coordination of care. Whilst it forms the lowest of proportion of actions in England's action plans it is the highest in Scotland's. Of those actions, two are behind schedule, two are ongoing, and two are complete.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Mental Health and Wellbeing Strategy: The publication of the strategy in 2023 was an important milestone, setting out a national vision for people in Scotland to achieve the best mental health and wellbeing possible. The latest progress report notes the Rare Conditions Policy Team helped to ensure that the strategy met the needs of those living with rare conditions. We would like to see that team consider publishing a separate short paper reflecting on how the specific mental health challenges facing people affected by a rare condition can be addressed, linked to the proposed outcomes of	**Consider a future Care Coordination Service in Scotland.** The recently published progress report highlighted how this work has not progressed due to financial constraints. It is important work continues over the next year to consider how a national model for Scotland could be rolled out, building on local services already in place in different parts of the country. **Implement the "It's OK to Ask" campaign**: Whilst the work carried out as part of the campaign, including the value based health and care action plan and module for health

the strategy, given the unique issues facing these patients.	professionals on shared decision making, is welcomed – they do not refer to support for coordinating care for people with rare conditions. Our members would like to see this work continue, though with a specific focus on how they are delivering on the third priority of the Rare Diseases Framework. **Rare conditions patient passports:** The progress notes a practical route to take this forward has not been identified. One member noted that this is disappointing as there are examples of good practice from other nations that Scotland could utilise as a template.

Wales

Six actions have been committed to against this priority across four areas. Of those actions, two are complete and four are ongoing. Two new actions were committed to in 2024: develop patient-reported outcome measures for the SWAN clinics and improve consideration of equity, diversity and inclusion measures.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
SWAN clinics: Wales has been an exemplar in their introduction, hosting the first such clinic. It successfully assessed 105 referrals from across Wales as of January 2024. The clinic has also designated two Clinical Nurse Specialists (CNSs) as care co-ordinators for patients. One member told us how the clinic helped patients with undiagnosed rare conditions who would not have otherwise been diagnosed. **Rare disease patient pathway:** The launch of the pathway has been welcomed, helping to link patients to emergency services and enable patients and carers to collate their own information, to be shared with healthcare professionals when required.	**Ensure implementation of transition guidance with a named worker and digital care plan linked to a patient passport:** Progress against this action was initially slow, though the SHCA welcomes news that the Southeast region is developing rare condition transitional support to enable a smooth and supportive journey to adult services. It is important this is rolled out across Wales. **Ensure the mental health needs of rare conditions patients and carers are considered as part of the overall mental health strategy for Wales**: SHCA members have been disappointed to see only limited progress here. The latest progress report notes "prioritisation of resources limits opportunities to support this action."

Northern Ireland

Four actions were committed to in the 2022 Action Plan against this priority, with progress against all four ongoing.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Develop care pathways and new models of care: Work has been carried out to develop service specifications and clinical pathways for a variety of adult services, to set out pathways for diagnosis, support and care, and access to treatment.	**Expert Centre for Rare Diseases**: We welcome the completion of the scoping review for the centre, which the original Action Plan said would include a rare conditions specialisms co-ordinator. SHCA members are keen to gain additional clarity on the centre's scope and how it will improve coordination of care for patients accessing care across different specialities. It is important the public consultation demonstrates how the expert centre is joined up to other actions committed to in the Action Plan relating to this priority.

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

England

14 actions have been committed to in England to support access to specialist care, treatments and drugs, reflective of the broad cross-cutting nature of the priority. Six of these actions have already been identified as completed – from a review of geographical variation in access to treatment in highly specialised services, to the mapping of the rare disease research landscape by the National Institute for Health and Care Research (NIHR).

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Review of the uptake of treatments for patients with very rare conditions across highly specialised services: This review was carried out in 2023 and was followed by constructive engagement with external stakeholders on its findings. It enabled issues with uptake in specific regions to be identified and addressed. This work should be carried out annually, including newly available treatments, to assess new issues that may emerge and reflect on progress made. **Mapping of the rare conditions research landscape by the NIHR**: The NIHR report was an important and previously under-addressed piece of work that presented an overall picture of rare conditions research taking place across the UK, with a second phase of this project now underway to identify research gaps and priorities. It is important this second phase incentivises greater research in areas such as	**Capitalise on changes to the NICE methods review**: Success against this action is to be measured on the 'number of medicines for rare diseases receiving a positive NICE recommendation'. However, as set out in the SHCA's recent report, Valuing rarity: Assessing the impact of the NICE methods review, SHCA members feel many of the changes introduced in the review have made it harder to secure a positive recommendation for a rare disease treatment. It remains unclear what the outputs will be; it is important it includes an analysis of the impact of NICE's updated methods and processes on treatments for rare conditions. **Review the effectiveness of the access landscape for rare conditions treatments:** Some progress has been made here; it is hoped the revised Innovative Licensing and Access Pathway will help to ensure rapid

health inequalities and coordination of care, given the findings of the initial report.	access to promising new rare conditions treatments. It is important that the review also reflects on challenges facing the Innovative Medicines Fund since its introduction in 2022, to consider how it can be reformed to become a more attractive access route for manufacturers of rare conditions treatments that are accompanied by higher levels of uncertainty. **The strategic approach to ATMPs**: Though internally focused, members were frustrated this was not accompanied by stakeholder engagement on how it is supporting horizon scanning for these treatments and what the outputs have been.

Scotland

Four actions were included in the 2022 Action Plan on improving access to specialist care, treatments and drugs. Three of those are ongoing and one is complete, though the completed action refers to the introduction of the ultra-orphan pathway in 2018, prior to the development of the UK Rare Diseases Framework.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Recognising the role of specialist services: Ongoing work by NHS National Services Scotland to develop specific rare conditions services is an important development and has built on Scotland's genomics strategy. A horizon scanning process linked to the Scottish Medicines Consortium has been introduced and a Scottish Genomics Test Advisory Group for Rare and Inherited Conditions launched to review and plan for the addition of new tests to the Scottish Genomic Test Directory. SHCA members look forward to seeing the outputs of ongoing work to deliver a single new planning framework, which has the potential to improve early access to innovative treatments through genetic testing.	**Introduction of the ultra-orphan pathway:** The pathway is a separate route for ultra rare conditions treatments within the Scottish Medicines Consortium's processes, introduced in recognition of the unique barriers to access facing these treatments. Whilst welcomed, Scotland is the only one of the four UK nations to not include a specific new action focused on improving access to rare conditions treatments, and members were frustrated to see past actions referred to rather than new ones committed to. Members would like to see issues such as improving equity of access to very rare conditions treatments included in future progress reports, building on the work carried out by the highly specialised services team in England.

Wales

Four actions were committed to in Wales against two themes: access to treatment and access to specialist care. Four additional actions were proposed in the 2024 plan.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Ensure horizon scanning for new medicines for patients in Wales to allow timely awareness and availability of new medicines: The Blueteq High Cost Drugs (HCD) system was rolled out across NHS Wales from April 2023 to help improve speed of access. Additional actions committed to in the progress report are also welcomed around improving access to research on new treatments and supporting the use of repurposed and off-label medicines and devices.	Whilst the themes committed to in the Wales Action Plan are welcomed, it is unclear what new actions have been taken as a result: the progress update to **ensure continued access to orphan and ultra-orphan medicines in Wales** only refers to the statutory responsibility of the health technology assessment body in Wales to fund new treatments within 60 days of a decision. Meanwhile the action on **timely awareness of new medicines** only lists treatments that have been approved for use in Wales since 2022. We would like to see new actions committed to on this priority that go beyond the statutory responsibilities of government bodies.

Northern Ireland

Four actions were committed to in the 2022 Action Plan against this priority that are ongoing, covering improving access to treatment, specialist advice and research, and introducing a Rare Diseases Champion for Northern Ireland.

The following examples of progress and opportunities to go further are identified:

Progress made	Opportunities to go further
Improve access to treatments for rare conditions: A survey of HCPs has been disseminated to gauge their awareness of how to prescribe and make requests for non-commissioned, specialist medicines and their knowledge of existing access schemes. SHCA members welcome this work, given the very small patient populations for many rare conditions in Northern Ireland and the resultant issues this can create in accessing innovative treatments. The progress report says the data will be used to identify current challenges and solutions to overcome them, and it is important this is set out in the 2025 progress report.	**Introduce a Rare Diseases Champion for Northern Ireland**: The only update provided is on the development of an options paper exploring the feasibility and potential advantages of the role. SHCA members are keen to see analysis of how a prospective Rare Diseases Champion could help to 1) increase the visibility of rare conditions within the Department of Health and Social Care, 2) support funding allocation to the delivery of the Rare Diseases Framework, and 3) help to coordinate with other UK nations in building on actions committed to and considering how they can be replicated in Northern Ireland.

The case for refreshing the UK Rare Diseases Framework

The analysis set out in this report, alongside the accompanying feedback from SHCA members, illustrates the momentum created by the UK Rare Diseases Framework in improving care for people with rare conditions across the UK. All four UK nations have worked constructively in engaging with the rare conditions community to consider how progress can be made against the four priorities of the Framework, within the resources available.

The underpinning themes of the Framework have also delivered progress, such as through the development of rare conditions registries across the four nations. The establishment of Rare Disease Research UK, meanwhile, will continue to deliver advances in rare disease research long past the cycle of the Framework.

> *"The Framework has given a unifying policy focus to rare conditions, especially on common clinical priorities, across all UK nations that has markedly raised the profile of rare conditions."*

However, our findings also reflect the barriers to progress each nation's rare conditions policy team has faced – and whilst we recognise the challenging fiscal environment, governments in each nation have failed to provide the necessary ring-fenced funding to support the delivery of the Framework's ambitions. The subsequent lack of capacity has constrained the work of policy teams in each nation and in some cases led to scaled back ambition and the pausing of actions which could deliver significant benefit to people affected by rare conditions.

In spite of these challenges, the Framework has shown what is possible when all four nations work together with the community to consider how care for rare conditions can continue to be improved – from expanding access to whole genome sequencing, to the establishment of SWAN clinics and the expansion of training and educational materials on rare conditions for healthcare professions.

However, there is also frustration within the community that examples of best practice from one nation is not always replicated at the same scale, if at all, elsewhere: the introduction of patient passports, establishment of rare conditions information hubs, and roll out of SWAN clinics are some examples of this.

Without the Rare Diseases Framework, there is a risk that the progress it has enabled will be lost, with government priorities shifted elsewhere. A successor framework would enable initial lessons to be learnt from and opportunities for further progress to be delivered on, building on the foundations the existing Framework has created

> *"Without a renewed Framework, I fear that our community will get overlooked as the preventative agenda focused on population health increases. But we face big issues and those need more than an un-resourced action list."*

The SHCA and Genetic Alliance UK call for the UK Rare Diseases Framework to be refreshed, renewed and recharged after 2026, following a period of consultation with stakeholders on how the challenges set out in this report can be overcome.

December 2024

Appendix 1: Methodology

Survey questions were drafted in collaboration between the SHCA and Genetic Alliance UK. All 12 respondents operate in each of the four UK nations, though some only employ staff in England:

- 2 have employees in each of the four nations
- 4 only employ representatives in England
- 1 employs representatives in England, Wales and Northern Ireland
- 1 employs representatives in England and Scotland
- 2 charity respondents are entirely volunteer run

Appendix 2: Nation specific action plans and progress report

- UK Rare Diseases Framework, 2021. Accessed here

England

- 2022 England Rare Diseases Action Plan. Accessed here
- 2023 England Rare Diseases Action Plan. Accessed here
- 2024 England Rare Diseases Action Plan. Accessed here

Scotland

- Rare Disease Action Plan 2022. Accessed here
- Rare Disease Action Plan for Scotland – Progress Report 2024. Accessed here

Wales

- Refreshed Wales Rare Disease Action Plan. Accessed here
- Wales Rare Disease Action Plan Progress Report. Accessed here

Northern Ireland

- Northern Ireland Rare Diseases Action Plan 2022/23. Accessed here
- Northern Ireland's Rare Diseases Action Plan Progress Report Year 1. Accessed here
- Northern Ireland's Rare Diseases Action Plan Progress Report Update Year 1. Accessed here