GENETIC ALLIANCE UK

MORE THAN YOU CAN IMAGINE

Opportunities for improving the lives of people with rare conditions

About Genetic Alliance UK

Genetic Alliance UK is an alliance of over 200 charities and support groups working together to improve the lives of people in the UK with lifelong and complex genetic, rare and undiagnosed conditions.

We advocate for fast and accurate diagnosis, good quality care and access to the best treatments. We actively support progress in research and engage with decision makers and the public about the challenges faced by our community.

We run two long standing projects:

Rare Disease UK: A campaign focused on making sure the UK Rare Diseases Framework is as successful as possible, and to ensure that people and families living with rare conditions have access to a final diagnosis, coordinated care and specialist care and treatment.

SWAN UK: The only dedicated support network in the UK for families affected by a syndrome without a name – a genetic condition so rare it often remains undiagnosed.

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Foreword

Rare conditions are individually rare but collectively common, with over 3.5 million people in the UK living with a rare condition.

To mark Rare Disease Day 2025, alongside this report, Genetic Alliance UK are publishing *'More than you can imagine: an anthology of rare experiences'* (the Anthology), a collection of creativity showcasing the impact rare conditions can have on a person's sense of self, relationships and wellbeing.

More Than Zebra by Matilda Tumim - instagram.com/tumimandprendergast

While the Anthology shares individual experiences of rare conditions, it is impossible to ignore the shared challenges that underpin each story. Delays in getting an accurate diagnosis, a lack of understanding among healthcare professionals, poorly coordinated care and barriers to accessing services and treatment are common challenges that shape a person's rare experience.

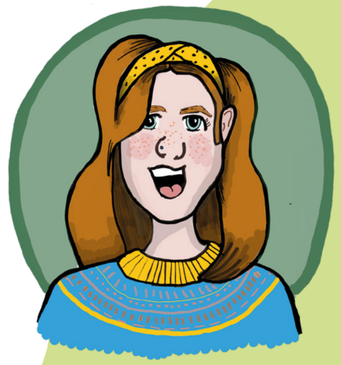
The UK Rare Diseases Framework (the Framework) was published in 2021 with the intention of tackling these challenges head on. The ambition of the Framework was to 'develop positive change in how we diagnose, treat and care for patients with a rare disease' and 'ensure that rare disease patients receive the best possible care'.

Over the past four years, substantial progress has been made. The Framework has certainly facilitated positive change, but much more needs to be done to achieve the ambition of delivering the best possible care for people with rare conditions.

The existing Framework has laid strong foundations and since its publication, new opportunities have emerged. Scientific developments have deepened our understanding of rare conditions and exciting innovations such as Artificial Intelligence (AI)-driven diagnostics, genomic testing, cell and gene therapies, and digital care support have progressed, presenting opportunities to improve health outcomes and reduce inequalities.

We cannot afford to lose momentum at this critical point. As we enter the final year of this Framework, we must make preparations for what comes next. We are calling on the governments of the UK to commit to renewing the UK Rare Diseases Framework.

3.5+
million people in the UK
are living with a rare condition



The shared experience of rare conditions

The shared experience of rare conditions has been captured by Rose Matheson (illustrator and paramedic, Scotland). Rose attended Genetic Alliance UK's Member Meeting in January 2025 where members of the rare community shared the most common challenges of living with a rare condition and discussed what would help improve their experience of care.

This illustration reflects the key points of that discussion and provides a visual representation of the shared experience of rare conditions.

The second part of the illustration showing how healthcare professionals view rare conditions can be seen on **page 15**.

Rose's images can be viewed on Genetic Alliance UK's website.

Introduction

In the UK, 1 in 17 people will be affected by a rare condition during their lifetime. That is 1 in 17 people who face the possibility of a long journey to diagnosis, poorly coordinated care or barriers to accessing services or appropriate treatment. Too often members of our community report having to 'battle' and 'fight' to get the care they need.

This report gives a broad overview of how the existing UK Rare Diseases Framework (January 2021 – January 2026) has sought to address some of the challenges facing people living with a rare condition. We have explored the progress that has been made, and with the help of the rare conditions community, have identified what needs to happen next.

The key messages in this report are drawn from Genetic Alliance UK's Patient Empowerment Group papers and our recent Rare Disease Day reports; *'Good Diagnosis' (2022)*, '*Coordinating Care' (2023)* and *'Stats behind the Stories' (2024)*. We also reflect on our collaboration with the Specialised Healthcare Alliance to deliver a '*Manifesto for Rare Diseases*', and would like to thank them for informing this report with findings of their recent member survey. We would also like to thank the Northern Ireland Rare Disease Partnership for their important input on the progress that has been made in Northern Ireland.

Genetic Alliance UK can only deliver our advocacy because our members and people living with rare conditions share evidence and their stories with us. We would like to thank them.

As this report demonstrates, there remains significant unmet need within the rare conditions community. A new Framework is essential if we are to improve the experiences of people with rare conditions. This report is intended to start the discussion about what a new Framework may look like.

We will continue to work alongside and on behalf of our community to campaign for the governments of the UK to renew the UK Rare Diseases Framework.

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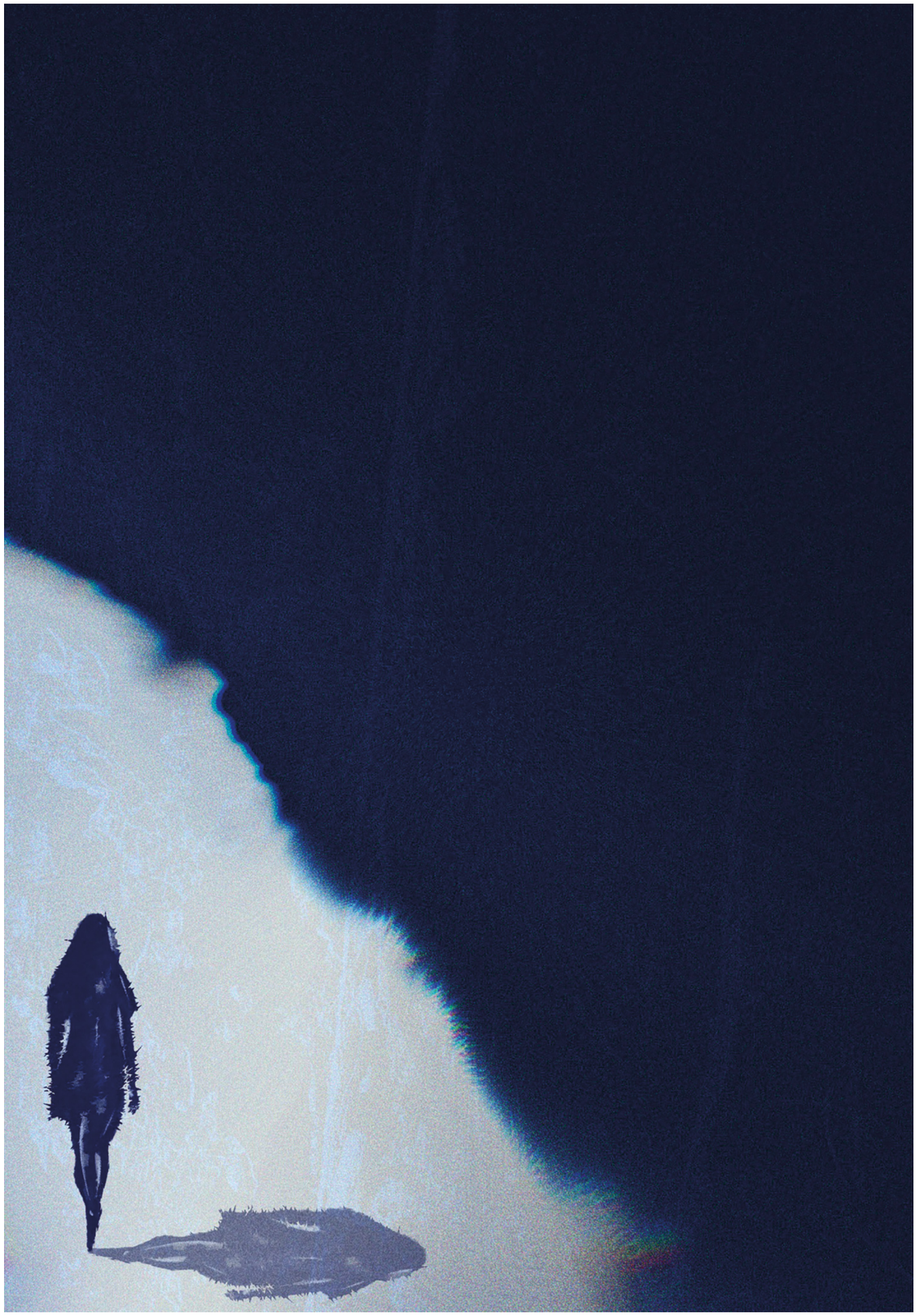


'Invisible Battles' poignantly depicts the impact of Pregnancy Associated Osteoporosis (PAO) including the loneliness that women feel on their journey to diagnosis and beyond, and the loss of connection between mother and child.

Taken from 'More than you can imagine: an anthology of rare experiences'. Art by Rhyanna Halasovski (Studio RLH), contributed by PAO UK.

The UK Rare Diseases Framework

The story so far

The UK Rare Diseases Framework, published in 2021, aims to improve the diagnosis, treatment, and care of the 3.5 million people living with rare conditions in the UK.

The Framework was co-signed by the health ministers of all four nations and was developed following the National Conversation on Rare Diseases Survey, which gathered over 6,000 responses from individuals, support organisations, and healthcare professionals.

The Framework prioritises four key areas:

1. Faster diagnosis

2. Increased awareness among healthcare professionals

3. Improved care coordination

4. Better access to specialist care, treatments, and medications

Five underpinning themes support the priorities: **patient voice**, **national and international collaboration**, **pioneering research**, **digital, data and technology**, and **policy alignment**.

At a UK level, The UK Rare Diseases Framework Board ensures coordination across the nations, while the UK Rare Diseases Stakeholder Forum facilitates ongoing collaboration.

As healthcare is a devolved matter, each nation is responsible for creating its own Rare Diseases Action Plan with input from the rare conditions community.

Has the Framework delivered?

The Framework has facilitated some important progress at both UK and national level, including:

1. Rare Disease Research UK, which promotes collaboration between researchers, patients, and charities. Eleven research nodes have been set up to focus on specific and cross-cutting rare disease topics.

2. An Independent Advisory Group was established to develop a Rare Disease Quality Standard, designed to promote high-quality, cost-effective care for individuals with rare conditions.

3. Scotland and Northern Ireland have developed rare conditions information hubs to provide essential resources for healthcare professionals and people with rare conditions.

4. Wales has introduced SWAN Clinics, where specialist nurses coordinate care for individuals with undiagnosed genetic conditions.

5. Rare disease registries have been implemented in England, Wales and Scotland to collect essential data for research and treatment.

A comprehensive list of individual items of progress can be found in the progress reports published by each nation.

Although a project has been commissioned to develop metrics for assessing the England Action Plan, their absence to date means it is challenging to evaluate the impact of the progress made. It is also difficult to contrast and compare how the Framework has been implemented across the four nations due to the different approaches to delivery in each nation.

Challenges and considerations for the future

The differing approaches between nations complicate comparisons and fiscal constraints, exacerbated by the Covid-19 pandemic, have limited the implementation of new initiatives. The absence of ring-fenced funding to support the Framework's delivery has hindered progress, with cost-neutral actions limiting the scope for innovation.

Despite these challenges, there is strong support for renewing the Framework. A survey by the Specialised Healthcare Alliance emphasises the need for greater investment and coordinated action to address the needs of those with rare conditions.

To be successful in meeting the needs of the rare conditions community, the delivery of a new Framework needs to be supported by:

- **Ring-fenced funding for each nation to support Framework priorities**
- **Well-resourced delivery teams for effective implementation**
- **Ongoing involvement of the rare conditions community**
- **Enhanced collaboration across the four nations, including joint initiatives**
- **Development of clear metrics to monitor progress and evaluate outcomes**
- **Regular reporting of action plans and progress to facilitate comparison and collaboration**

Genetic Alliance UK calls for renewed commitment from UK governments to support a refreshed Rare Diseases Framework for 2026.

Delivering the Framework across the UK

Northern Ireland

Northern Ireland published its first Rare Disease Action Plan in 2022, outlining 14 key actions. The Department of Health's NI Rare Diseases Implementation Group oversees its delivery through five working groups.

Progress includes work to establish a national rare diseases information hub. Efforts to improve healthcare professional education and training have advanced through collaborations with the Northern Ireland Rare Diseases Partnership (NIRDP) and a rare disease society has been formed at Ulster University's School of Medicine.

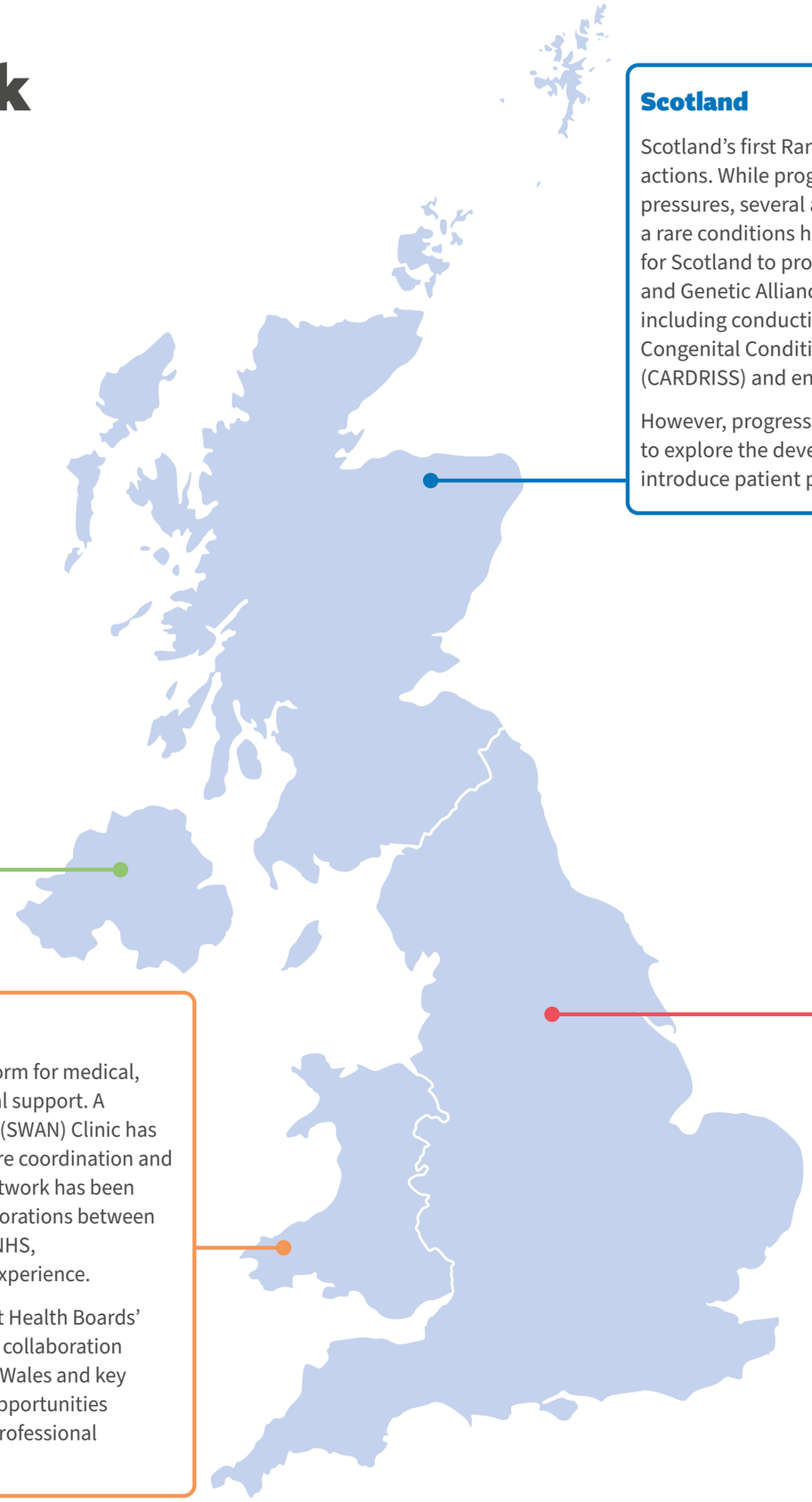
New care pathways for several paediatric and adult services have been developed, and a patient and carer survey on research experiences has been launched. A £12m LifeArc Centre for Acceleration of Rare Disease Trials, led by Queen's University Belfast, in consortium with the University of Birmingham and Newcastle University, will focus on improving the efficiency and availability of rare disease trials.

Progress to review newborn screening and establish a rare disease registration service has been slower. However, research resources have now been secured to support registry efforts.

Scotland

Scotland's first Rare Disease Action Plan, published in 2022, outlined 18 actions. While progress has been limited due to fiscal constraints and NHS pressures, several actions are moving forward. These include the creation of a rare conditions hub on NHS Inform and collaboration with NHS Education for Scotland to produce signposting videos. The Office for Rare Conditions and Genetic Alliance UK have supported the delivery of a number of actions, including conducting a healthcare professional survey. Efforts to expand the Congenital Conditions and Rare Diseases Registration and Information Service (CARDRISS) and enhance rare disease research in Scotland are also underway.

However, progress on diagnosis and care coordination has been slower. Actions to explore the development of a care coordination service for Scotland and to introduce patient passports have not been progressed.



Wales

The Rare Disease Implementation Network (RDIN) oversees the Welsh Rare Disease Action Plan, first published in 2021 and refreshed in 2023 with 29 actions. Welsh Government funding enabled a clinical lead and coordinator to support its implementation. The 2024 progress report outlined three focus areas: education, data linkage, and clinical care coordination.

Key achievements include developing a rare disease dashboard and patient-reported measures (PREMs and PROMs) to support evaluation and the launch of a Digital Rare Care Centre Pilot in Southeast Wales, which will provide an online platform for medical, social care, and professional support. A Syndrome Without a Name (SWAN) Clinic has demonstrated improved care coordination and a Rare Disease Research Network has been established to foster collaborations between academic institutions, the NHS, and individuals with lived experience.

While fiscal constraints limit Health Boards' full engagement with RDIN, collaboration with the Genomics Plan for Wales and key partners offers promising opportunities to enhance diagnosis and professional awareness.

England

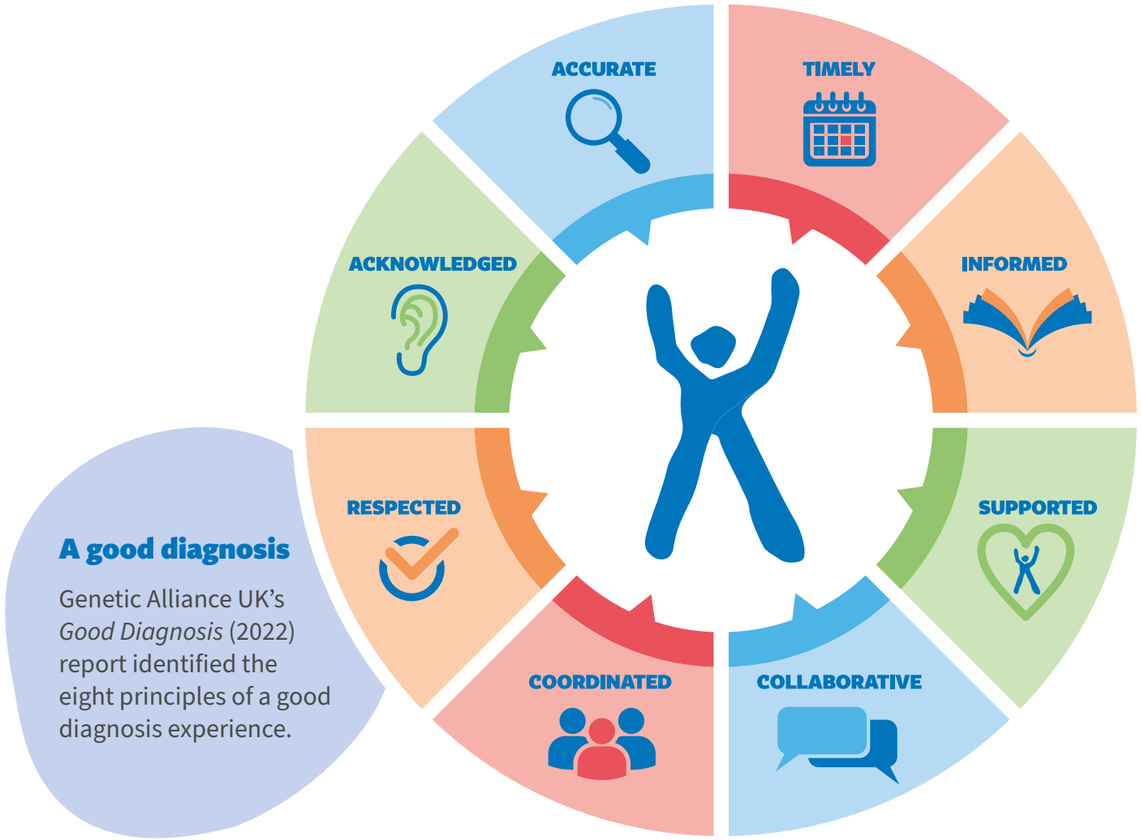
England's Rare Disease Action Plan, first published in 2022, has been updated annually, with the fourth and final iteration expected in February 2025. The England Rare Diseases Framework Delivery Group is responsible for the delivery of the action plan and introduced six focus areas within the priorities and underpinning themes of the Framework in 2022: non-genetic and undiagnosed conditions, mental health support, coordination of care, clinical research delivery, registries and monitoring and evaluation.

Progress has been made in several areas including the launch of the Generation Study, inclusion of the definition of care coordination in all new and revised service specifications from 2023, and improved functionality of the Be Part of Research Platform.

However, some actions have faced delays. Of the 29 actions reviewed in the 2024 progress report, eight were completed, seven are ongoing, and 15 have been extended. Commitments made before the UK Rare Diseases Framework came into effect are also missing. For example, work on the development of alert cards has not progressed.

Diagnosis

More than 1 in 3 people with rare conditions experience delays of over five years between the onset of symptoms and a final diagnosis, a process known as the 'diagnostic odyssey.' This delay can limit access to appropriate care and support. Faster, more accurate diagnoses improve patient outcomes and reduce unnecessary referrals, tests, and treatments, benefiting both individuals and the NHS.



A good diagnosis

Genetic Alliance UK's *Good Diagnosis* (2022) report identified the eight principles of a good diagnosis experience.

Research has shown that diagnostic delays cost NHS England over

£3.4 billion

across a 10-year period*

*Research undertaken by Imperial College Health Partners.

Progress under the existing Framework

1. **Whole genome sequencing (WGS) for faster diagnosis:** WGS enables healthcare professionals to examine a person's complete genetic code to identify genetic variants that may be causing a rare condition. In England, the Generation Study has been introduced to pilot the use of WGS to screen healthy newborns for over 200 genetic conditions that are not currently covered by the NHS newborn blood spot screening programme.

2. **Syndrome Without A Name (SWAN) Clinics:** SWAN clinics in Wales offer support to individuals with undiagnosed conditions. These clinics provide expert advice, genetic data re-analysis, and additional support. Pilot SWAN clinics are being developed in England.

3. **Newborn screening for rare conditions:** The UK Newborn Screening Committee (UK NSC) oversees the newborn blood spot screening programme, which currently screens for nine genetic and rare conditions, soon to be ten. The rare conditions community has criticised the UK's delay in expanding screenings compared to other countries. In response to calls to improve how decisions are made by the UK NSC, the Blood Spot Task Group has been established. In addition, an in-service evaluation (ISE) of newborn screening for severe combined immune deficiency is in progress and planning for an ISE of newborn screening for spinal muscular atrophy is underway. While these developments are welcome, it is important to note that ISEs are not UK-wide and may lead to inequity.

4. **National rare disease registration services:** In England (National Congenital Anomaly and Rare Disease Registration Service), Wales (Congenital Anomaly Register and Information Service) and Scotland (Congenital Conditions and Rare Diseases Registration and Information Service for Scotland) are also gathering data to inform healthcare planning and research.

Opportunities for a future Framework

1. **Consider non-genetic conditions:** While current efforts focus on genetic conditions, 20% of rare conditions are not genetic. England's 2023 Action Plan delivered a workshop to explore how to better support individuals with non-genetic rare conditions. Insights from this workshop should inform national action plans to improve diagnosis for these conditions.

2. **Supporting people on their journey to diagnosis and beyond:** Many individuals report dissatisfaction with the information and support they receive during the diagnostic journey. A future Framework should seek to improve the experience of diagnosis and could include a diagnosis care plan to provide better support throughout this process.

3. **Capitalising on advancements in Artificial Intelligence (AI):** AI technologies like machine learning hold great potential to speed up diagnoses. AI can analyse large amounts of medical data, including genetic information, to identify patterns that may go unnoticed, potentially helping healthcare professionals diagnose rare diseases more quickly and accurately.

4. **Expanding newborn screening:** A future Framework should prioritise expanding the UK's newborn blood spot screening programme to improve early detection of rare conditions, in line with international best practice. The findings from the Generation Study on WGS should also be evaluated to determine how it can be integrated into newborn screening for earlier diagnosis.

5. **UK-wide SWAN clinics:** A future Framework should be informed by evaluation of the pilot SWAN services in Wales and England and consideration should be given to how similar services are rolled out across the UK.

Raising awareness of rare conditions among healthcare professionals

There are around 7,000 rare conditions, with new ones regularly identified through scientific advancements. Some are so rare that only a few families in the UK are affected, and others are so new that they may only affect one person.

Rare conditions often present with vague or common symptoms, making it unrealistic to expect healthcare professionals (HCPs) to know them all. However, since 1 in 17 people will be affected by a rare condition in their lifetime, it is essential for HCPs to be prepared to deliver high-quality care and support.

HCPs who are knowledgeable about rare conditions may consider them as potential diagnoses, reducing misdiagnosis and treatment delays. Recognising the limits of their expertise may mean that HCPs may be more willing to seek additional information and refer patients to specialists or signpost to appropriate support services. This can improve access to timely and effective treatment.

Progress under the existing Framework

Several initiatives have been launched across the UK to improve rare disease education for HCPs:

1. **Clinical leads for rare conditions:** NHS Wales and Northern Ireland have appointed clinical leads for rare diseases. These roles provide specialised knowledge which may help improve diagnosis and early detection, and enhance care coordination.

2. **GeNotes educational resource:** The GeNotes platform, part of the UK's genomics initiative, provides HCPs with access to information on genetic and rare conditions, helping them integrate this knowledge into patient care.

3. **Rare conditions information hubs:** Northern Ireland and Scotland have developed or improved information hubs that allow HCPs to educate themselves on rare conditions, providing a valuable resource for medical professionals.

4. **Incorporating rare conditions into formal education:** Efforts have been made to include rare diseases in healthcare curricula. For example, the School of Medicine at Ulster University and the NI Rare Diseases Partnership have integrated rare diseases into undergraduate and postgraduate programs. Cardiff University has also introduced a rare condition teaching week for medical students.

5. **Independent Advisory Group (IAG):** Led by Medics For Rare Disease, the IAG is working to define competency expectations for HCPs and develop strategies to improve rare disease education.

Opportunities for a future Framework

1. **Wider integration into medical curricula:** Rare conditions education should be mandatory across all medical and health-related programmes. A more consistent approach is needed to ensure all HCPs are equipped to recognise and manage rare conditions early.

2. **Develop a digital platform for HCPs:** A centralised digital platform would allow HCPs to easily access information and seek advice from specialists, addressing knowledge gaps, particularly for general practitioners.

3. **Establish rare conditions champions:** A network of 'rare conditions champions' could raise awareness, provide guidance, and support HCPs in recognising and addressing rare diseases.

4. **Expand Continuing Professional Development (CPD) opportunities:** CPD opportunities focused on rare diseases, such as online modules and workshops, would ensure HCPs stay informed about new developments.

5. **Promote interdisciplinary collaboration:** Collaboration between general practitioners, specialists, geneticists, and patient advocacy groups is essential to raising awareness and improving care for rare disease patients.

6. **Build on existing initiatives for Rare Disease Day:** A coordinated, UK-wide approach to use Rare Disease Day to improve awareness and promote education on rare conditions should be considered.

How healthcare professionals view rare conditions

In January 2025, Genetic Alliance UK held a workshop with four HCPs to explore their views on rare conditions.

The workshop explored:

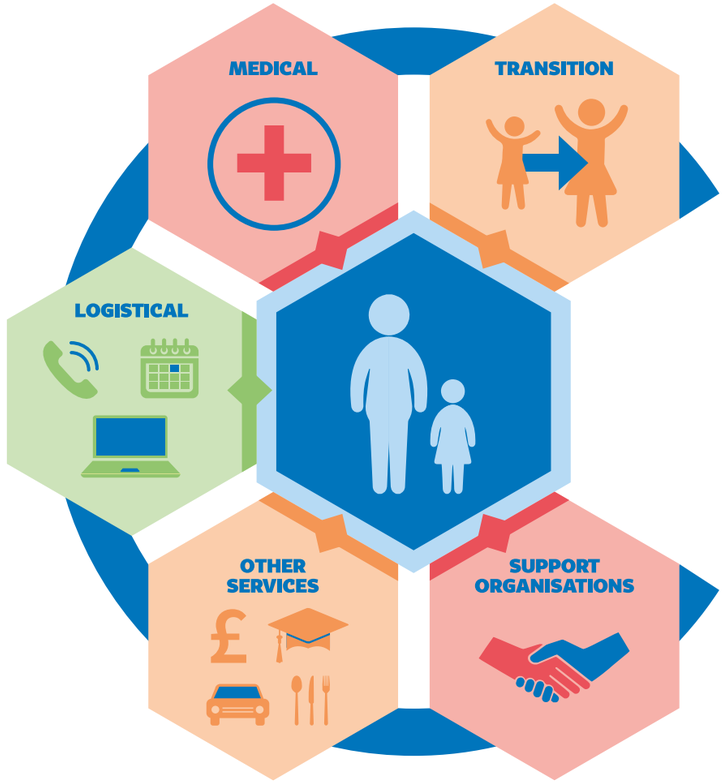
- **the perceived biggest challenges to providing care for people with rare conditions**
- **the tools needed to help HCPs support people with rare conditions better**
- **what all healthcare professionals should know about rare conditions**

This valuable discussion was captured in this illustration by Rose Matheson. Their illustration reflects the key points of that discussion and provides a visual representation of the HCPs views on rare conditions.

Care coordination

Coordination of care is crucial for individuals with rare conditions, as it involves organising and sharing relevant information across services to ensure safer, more effective care. Effective care coordination must address not only medical needs but also mental health, social care, and support for work or education. Access to coordinated care should be available to everyone, regardless of their diagnosis, circumstances, or location.

People with rare conditions often face complex health needs that require multiple healthcare services. However, many lack coordinated care and are left to manage it themselves, which adds to the burden of managing their condition, particularly for those needing ongoing, specialised care.



Genetic Alliance UK's 2023 report '*Coordinating Care: Learning from the experience of people living with rare conditions*' highlights that the value of care coordination for people with rare conditions and healthcare professionals can be seen across the following themes:

- Coordinating complex medical care
- Delivering well-organised logistical support
- Assisting an effective move from children's services to adult services
- Bridging the gap between healthcare and other services
- Integrating support from rare condition charities and support groups

Progress under the existing Framework

Efforts to improve care coordination for rare diseases have faced challenges due to funding limitations, resource constraints, and the strain on NHS systems following the pandemic. While many ambitious initiatives have not progressed, some successes have been achieved:

1. Northern Ireland has developed Rare Disease Care Pathways for inherited cardiac conditions, inherited metabolic disorders, immunology, haematology and cystic fibrosis. Cross-border collaboration to develop these tools could help create a unified approach across the UK, and maximise use of resources.

2. Wales has established rare conditions as 'Community of Practice,' focusing on multidisciplinary clinical pathways.

3. NHS England supports Rare Disease Collaborative Networks (RDCNs), bringing together providers to improve knowledge, research, and patient experiences for specific rare conditions.

4. In Wales, specialist nurses assist patients and families through the pilot SWAN clinic, providing coordinated care for those with undiagnosed conditions.

5. Scotland has incorporated rare conditions into broader healthcare policies, such as the Mental Health Strategy and Anticipatory Care Planning.

The CONCORD study has demonstrated the value of coordinated care and outlined a roadmap for improvement, but national action plans have struggled to implement meaningful changes. Initiatives like care coordinator roles, patient passports, and alert cards have not advanced, despite their potential to enhance care quality.

Opportunities for a future Framework

1. **Introduce new roles to support care coordination:** A future Framework should explore the introduction of dedicated care coordinators for rare conditions. These professionals would implement care plans, provide self-management information, and connect health and social services. Although they wouldn't need to be experts in specific conditions, they should be knowledgeable about the healthcare landscape and adept at finding resources and specialists. Standardising these roles across the UK, with sustainable funding, would improve care coordination.

2. **Explore the rollout of patient passports and alert cards:** Patient passports and alert cards can enhance communication between patients and healthcare providers, reducing errors, preventing unnecessary tests, and streamlining treatment. These tools can be particularly helpful for emergency healthcare settings. Cross-border collaboration to develop these tools could help create a unified approach across the UK, and maximise use of resources.

3. **Ensure mental health support for patients and families:** Mental health services for people with rare conditions are often limited. A future Framework should ensure mental health support both during the diagnostic process and after diagnosis. Organisations like Rareminds offer valuable resources that should be incorporated into care pathways to improve the overall wellbeing of patients and caregivers.

By advancing these initiatives with sustained funding, the UK can create a more effective, coordinated care system that supports people with rare conditions throughout their care journey. Collaboration between the four nations will be key to achieving these improvements.

Access to services, treatment and drugs

Rare conditions are often life-limiting or life-threatening. For the 1 in 17 people affected in the UK, accessing the right medications, expert care, and support services is essential for managing their condition and avoiding unnecessary treatments and referrals. Despite progress, significant challenges remain in obtaining effective treatments and accessing specialised services.

Specialised services

Specialised services provide expert care for specific rare conditions, typically through multidisciplinary teams. Access to these services is associated with improved patient outcomes.

A 2020 survey by Genetic Alliance UK found that:

85%

of patients who rated their care as 'very good' had access to a specialist centre, compared to **only 12%** of those who rated their care as 'very poor'

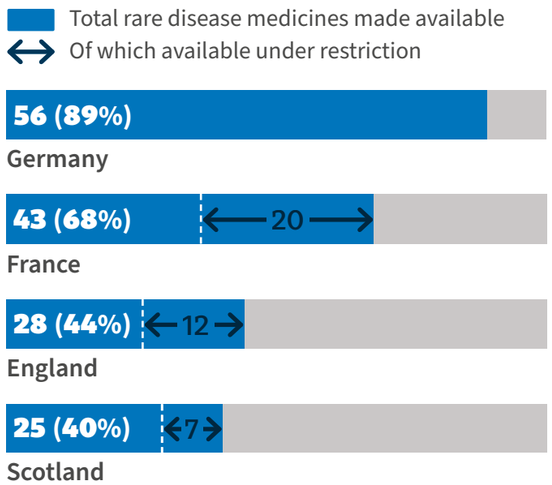
However, specialised services are not available for all rare conditions. Where they do exist, people with rare conditions benefit from 24/7 emergency support and expert care at multiple locations. Where they do not exist, people face fragmented care, poor coordination and delayed specialist appointments. Our member organisations are concerned that Integrated Care Boards (ICBs) could potentially exacerbate these challenges in England.

Access to these services is also hindered by difficulties in obtaining referrals, travel distances, and long waiting times. Some individuals may need to seek care in other UK nations, but strict referral criteria can delay or limit access.

Medicines

In recent years, there has been progress towards the development of medicines for rare conditions, with an increasing number of medicines becoming available. However, it remains true that for the vast majority of rare conditions there is no medicine. Currently, only 1 out of 20 rare conditions have an approved treatment or medicine. Worse, where medicines have been approved as safe and effective by regulators they can be slow to be judged cost-effective and many are either not available on the NHS in the UK or only available for a limited population.

The UK compares poorly to European countries on these measures. According to the 2024 European Federation of Pharmaceutical Industries Associations WAIT study, in January 2024, of 63 (100%) rare disease medicines approved by the EU 2019-2022:



Germany made access decisions on average 96 days after market authorisation, with France taking 597 days on average, England 394 days and Scotland 401 days (the study ignored the Medicines and Healthcare Products Regulatory Agency (MHRA) Early Access to Medicines Scheme).

By addressing these challenges, the UK can improve access to specialised services, treatments, and medications for those with rare conditions, ensuring more equitable and timely care for this often-overlooked group.

Progress under the existing Framework

1. The Medicines and Healthcare products Regulatory Agency (MHRA) has reviewed pathways to improve access to medicines, such as the Innovative Licensing and Access Pathway (ILAP).

2. Northern Ireland has made strides in developing rare disease pathways and collaborating across nations to address challenges related to medicine delivery.

3. There is also a focus on repurposing medicines and increasing the UK's readiness for Advanced Therapy Medicinal Products (ATMPs), including cell and gene therapies.

Opportunities for a future Framework

1. **Harness AI in diagnosis and treatment:** The potential of artificial intelligence (AI) in improving diagnosis and treatment development for rare conditions should be explored further.

2. **Personalised medicines and innovative commissioning:** A future Framework should support the use of personalised medicines, including n-of-1 therapies (n-of-1 therapies are individualised therapies developed for and used by one or very few people living with a rare condition), and innovative commissioning processes to bring these treatments to patients faster.

3. **Improved access to NHS specialised services:** Clearer guidance is needed to ensure that patients can easily access specialised services, including cross-border care arrangements.

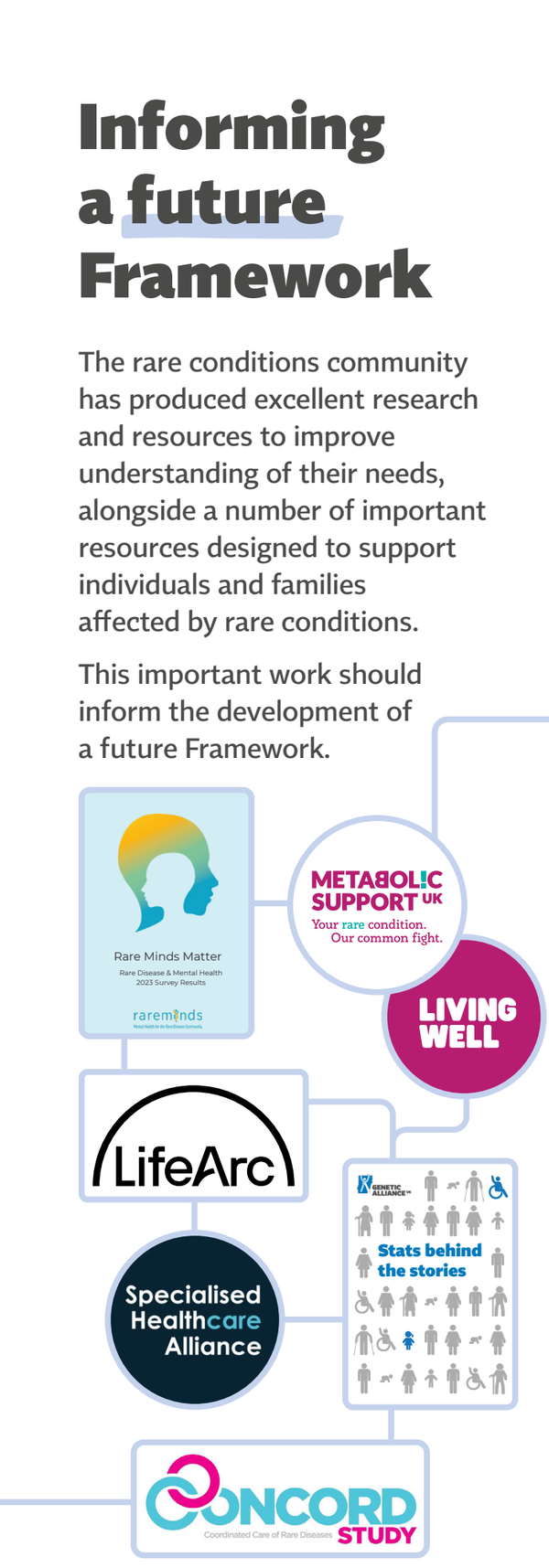
4. **Enhance equity of service provision:** Expanding data on rare conditions would help develop comprehensive diagnostic and care pathways for all individuals, improving equity in service provision.

5. **Ensure the effectiveness of the Innovative Medicines Fund:** The Innovative Medicines Fund should be effectively utilised to improve access to the latest treatments for rare conditions.

Informing a future Framework

The rare conditions community has produced excellent research and resources to improve understanding of their needs, alongside a number of important resources designed to support individuals and families affected by rare conditions.

This important work should inform the development of a future Framework.



CONCORD

The CONCORD study laid the foundations for evidence-based development of better coordination: it drew up a definition of care coordination and categorised the elements that contribute to care coordination to support conversations between advocacy groups, commissioners, policy makers and researchers; it assessed what is happening on the ground in the UK and the impact of poor care coordination on individuals and families; and it explored the preferences of those using health services and the professionals delivering them in terms of how care should be coordinated. The follow-up work, CONCORD2, is live until mid-2026 and will report in detail on the costs and benefits of different approaches to care coordination in different settings and scenarios. All of these findings should inform a new Framework.

Genetic Alliance UK Stats behind the stories

Little is known about the 3.5 million people in the UK affected by rare conditions. Genetic Alliance UK's 2024 '*Stats behind the stories*' report analyses 163 rare conditions from the Orphanet database with a prevalence between 5 in 10,000 and 1 in 10,000, which together could account for 80% of people with rare conditions.

The report identifies the need to segment and better understand UK data about who has rare conditions, and which rare conditions they have. The findings provide an introductory insight as to what would be possible with a well-resourced robust study.

LifeArc

Despite welcome recent attention from policymakers, the stark fact is that 95% of rare diseases lack an effective treatment, leaving the 1 in 17 people who live with them in limbo. This is all the more frustrating given recent advances in scientific understanding. As things stand, the rare disease R&D ecosystem faces an acute market failure, driven by complex interacting issues. These include fragmented data, diagnostic bottlenecks, small patient populations, and a convoluted regulatory landscape geared towards 'traditional' pathways for more common diseases.

To unblock progress, LifeArc convened an expert cross-sector Taskforce to map the UK rare disease R&D landscape and identify opportunities for the UK to improve the situation so that we can realise the market potential of rare disease and deliver much needed drugs to those living with rare disease. The resulting report identifies ten actionable recommendations to inform policymakers. These include developing a UK-wide data strategy, better evaluating the cost of rare diseases to society, 'concierge'-style support for innovators, and the creation of national coordinator roles.

Rareminds mental health survey

The 'Rare Minds Matter – Mental Health Survey 2023' provided a post-pandemic 'snap-shot' of the mental health of the UK's rare community. 695 respondents represented over 188 rare conditions.

The survey revealed the significant impact that a diagnosis of, and living with, a rare condition can have on mental health and wellbeing. This was apparent not just for affected individuals, carers, and family members, but also on the frontline staff and volunteers providing support too.

Key findings:

- Living with a rare condition creates additional and particular challenges for mental health and wellbeing.
- Rare conditions can affect the protective function usually played by couple and family relationships to support mental health and wellbeing.
- The quality of the relationships, and interactions with, healthcare professionals have a significant impact on the mental health and wellbeing.
- Mental health support is desirable, but accessing appropriate services is not always easy.
- The level of support provided by rare condition group leaders can impact their own mental health and wellbeing.

Rareminds Wellbeing Hub launched in 2024, with free and unlimited access to resources. Sections include Dealing with Diagnosis, Relationships, Uncertainty, Change and Loss, You and Your Feelings, Navigating Healthcare and Sources of Support.

Metabolic Support UK Living Well campaign

Quality of life is a key term in healthcare, used to assess how treatments impact an individual's wellbeing. But what does it truly mean to live well with a rare disease?

In 2023, Metabolic Support UK's Thoughts into Action project explored this question from the rare disease community's perspective. The research revealed that people with rare diseases rely on a 'net' of support structures, including identity, employment, benefits, food, healthcare, transitions, and mental health, that enable them to live well. Gaps in these areas can cause individuals to fall through the 'net,' leading to a worsened quality of life.

The Living Well Movement is advocating for policy change to address the full impact of rare conditions. By championing a holistic approach, the movement seeks to shape UK-wide policies that ensure people with rare diseases not only survive but thrive.

Specialised Healthcare Alliance member survey

The Specialised Healthcare Alliance (SHCA), in partnership with Genetic Alliance UK, **published a report reflecting on progress made to deliver the UK Rare Diseases Framework**. Advances have been made in every UK nation. The Framework and accompanying nation specific action plans have raised the profile of rare conditions across Government and driven closer coordination between and across departments and external stakeholders.

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 ring-fenced funding to accompany the delivery of the Framework, and resultant limited ambition and accountability within governments.

Building on progress made, and addressing the above challenges, together, the SHCA and Genetic Alliance UK are calling for the UK Rare Diseases Framework to be refreshed, renewed and recharged after 2026.

A Framework for the future

The UK Rare Diseases Framework has made significant progress, but there is still much to be done. Many people with rare conditions continue to face poor experiences in accessing care and often describe their journey as a ‘battle’ or ‘fight.’

A refreshed Framework offers the opportunity to create lasting change. By building on recent progress, embracing new ambitions, and leveraging innovations, we can transform the diagnosis, treatment, and care for those with rare conditions.

To achieve this, a robust governance structure is essential to enable UK-wide collaboration on rare conditions, along with a renewed Framework to drive the work forward. With less than a year left in the current Framework, we must act now to ensure momentum is not lost.

Genetic Alliance UK is urging the UK governments to urgently renew their commitment to rare conditions and ensure the UK Rare Diseases Framework is renewed for 2026 and beyond.

A future Framework should:

1. Build on existing foundations

The new Framework should build on the progress made by supporting existing initiatives like national registries, the NICE quality standard for rare diseases, and the UK Rare Diseases Research Platform. Successful pilot projects, such as the SWAN Clinics and Rare Disease Care Pathways in Northern Ireland, should be expanded across the UK. Research reports like the CONCORD study can help identify gaps in care and guide further improvements.

2. Maximise new innovations

Scientific advancements in rare conditions, such as AI-driven diagnostics, genomic testing, and gene therapies, offer hope for improved outcomes. The renewed Framework should accelerate access to these innovations. Engaging with the rare conditions community will be crucial to align these innovations with patient needs and maximise their benefits.

3. Involve the rare conditions community

Involving people affected by rare conditions and support organisations is essential for ensuring the Framework meets the needs of the community. The UK Rare Diseases Stakeholder Forum should be expanded to include more diverse voices.

A new Framework must involve all of the key stakeholders in improving outcomes for people living with rare conditions in the UK, including those who may have been peripheral to the implementation of the first UK Rare Diseases Framework. For example, rare cancer charities who share many of the common challenges of the rare conditions community and who are often underserved by existing cancer policies.

This also means looking beyond support organisations and widening engagement to include representatives from each nation’s national health services and representation from clinical communities. By doing so, there will be greater opportunities to explore initiatives that could be delivered by, and within, the NHS to improve the standard of care people receive. This has the potential to accelerate progress in delivering improved care coordination.

4. Improve understanding of rare conditions

Despite the significant number of people affected by rare conditions in the UK, data on prevalence and patient experiences remains fragmented. The renewed Framework should include efforts to systematically assess the prevalence of rare diseases, allowing the NHS to better organise services and create more effective care pathways.

5. Improve experiences of healthcare

The Framework should focus on improving the overall care experience for people with rare conditions. Positive care experiences build trust in the healthcare system. Actions such as investing in care coordinators, improving healthcare professional education, and expanding access to resources like Rareminds’ Wellbeing Hub and Genetic Alliance UK’s ‘Rare Resources’ should be prioritised.

6. Strengthen cross-border collaboration

A future Framework must continue and strengthen cross-border collaboration between the four nations. Pooling resources and sharing successful pilot projects, such as rare condition alert cards, will ensure equity in implementation and allow initiatives to benefit the entire UK population.

7. Be adequately funded and resourced

The existing Framework has been limited by the lack of funding. Without sufficient investment, progress has been slow, particularly in improving care experiences. The new Framework must include ring-fenced funding for services and initiatives. Metrics and benchmarks should be used to evaluate success and demonstrate cost savings and used to make rare conditions initiatives a driver of economic growth within the NHS.

Next steps for Genetic Alliance UK

Genetic Alliance UK will continue to work closely with the rare conditions community to campaign for a new Rare Diseases Framework, so that the UK maintains momentum to improve how we diagnose, treat and care for the 3.5 million people affected by rare conditions.

We will continue to liaise with the governments of the four nations to support the implementation of the existing UK Rare Diseases Framework and to inform discussions around the future of this important policy. We will be sharing this report, and our ‘*More than you can imagine: an anthology of rare experiences*’ with each nation’s civil service team.

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