

FUTURE FOR RARE

**A NEW VISION FOR RARE
CONDITIONS POLICY IN THE UK**

REPORT FOR MINISTERS

The findings of this report were informed by the Future for Rare expert summit held in collaboration with

ABOUT GENETIC ALLIANCE UK



Genetic Alliance UK is an alliance of over 250 charities and support groups working together to improve the lives of people in the UK with 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 2 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.



The Future for Rare campaign is being coordinated by Genetic Alliance UK to inform what the future of rare conditions policy in the UK should look like after 2026 when the current UK Rare Diseases Framework ends.

The campaign is being delivered in collaboration with a Future for Rare Steering Group. This multi-stakeholder group includes lived-experience representatives, charity leaders, research and industry experts who ensure the process remains inclusive, transparent, and focused on delivering high-quality evidence to the government.

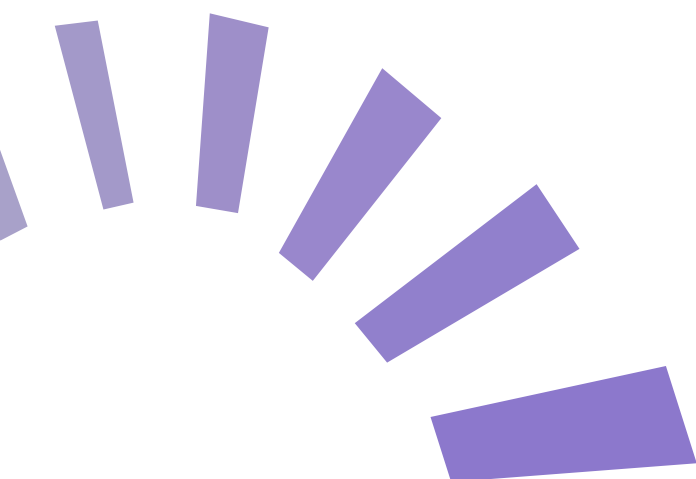
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EXECUTIVE SUMMARY

More than 3.5 million people in the UK live with a rare condition. Yet they rely on a healthcare system that was not built with them in mind, and which too often fails to recognise their needs.

Through our Future for Rare campaign, people living with rare conditions, families support organisations, clinicians, researchers and industry have set out a shared vision. They want an equitable healthcare system that values people with rare conditions and delivers for them, wherever they live and however rare their condition is.

The UK Rare Diseases Framework ends in January 2027. Over its lifetime, it has supported the delivery of national registries, the Rare Disease Research UK platform, new models of care coordination, undiagnosed clinic pilots and a NICE quality standard for rare conditions care, and much more across the four nations. These are hard-won foundations, and letting the Framework lapse would put them at risk just as they begin to deliver. We are not asking governments to start again. We are asking them to renew and refresh what has been built together, across all four nations, because no nation can do this alone.

The timing could not be more important. As each UK nation reshapes its health service, there is a chance to put rare conditions at the heart of that change. Rare conditions bring together complex, lifelong needs that cross specialities, settings and services. A system designed to meet those needs

from the outset will cost less than one adapted later, and will be stronger for everyone who uses it.

Genetic Alliance UK calls on governments across the UK to:

- 1. Renew a UK-wide policy framework and appoint a National Clinical Lead for rare conditions in each nation.**
- 2. Resource a national core data offer and embed diagnostic capability across the health system.**
- 3. Organise care through sustainable clinical networks and establish an annual national patient experience survey.**

The opportunity is here, and the time to act is now. Together, we can build a health system that values people with rare conditions and supports them to live well at all stages of their life.

ABOUT THIS REPORT

This report summarises the key messages of Future for Rare, Genetic Alliance UK's campaign to inform the next iteration of UK rare conditions policy.

Written by Genetic Alliance UK for Ministers and government teams in all four nations, it draws on extensive community engagement, including a UK-wide survey and call for evidence, and multi-stakeholder input from 11 expert thematic working groups and an expert summit held in collaboration with LifeArc, Rare Disease Research UK and the Rare Disease International Mirror and Action Group.

RECOMMENDATIONS

The Future for Rare campaign identified three priority actions for the UK to build a stronger, more coordinated and future-ready rare conditions ecosystem.

01

POLICY RENEWAL AND CLINICAL LEADERSHIP

The UK Rare Diseases Framework ends in January 2027. A UK-wide successor would safeguard the important initiatives developed under the current Framework, build on the progress made and avoid duplication across the four nations. A resourced National Clinical Lead in each nation should hold accountability for delivery of the new policy.

02

DATA VISIBILITY AND SYSTEM-WIDE DIAGNOSTICS

Many people with rare conditions cannot be found in NHS data. Governments should resource a core data offer built on national registries, genomic infrastructure and standardised coding, and move from isolated diagnostic initiatives to lasting diagnostic capabilities across the system.

03

SUSTAINABLE SERVICE NETWORKS AND SYSTEM EVALUATION

Coordinating care too often depends on individual clinical champions or falls to families. Existing specialist networks should be connected, resourced and expanded, with core roles funded by NHS provision. An annual rare conditions patient experience survey would measure progress nationally.

THE TIME TO ACT IS NOW

As all four nations reform their health systems, rare conditions must be integrated from the outset. With the UK Rare Diseases Framework ending in January 2027, Ministers must seize this opportunity to dismantle the systemic inequities faced by people affected by rare conditions.

THE RARE REALITY

Rare conditions are individually rare, but collectively common. Over 3.5 million people in the UK live with one of over 7,000 known rare conditions. 7 in 10 rare conditions affect children and it is estimated that more than half of childhood deaths are associated with a rare condition.

Behind each of these figures is a person navigating a system that was not built with them in mind. For many, the defining experience is the diagnostic odyssey itself: years of appointments, referrals and uncertainty before an answer, if one comes at all. When a rare condition is diagnosed, coordinated support is the exception rather than the rule.

Even among the best-understood rare conditions, fewer than a third have NICE guidance, over a quarter have no specialised NHS service at all, and genetic testing is available for less than half. For the rarest conditions, the picture is starker still.

This systemic fragmentation incurs significant societal and economic costs. Analysis highlighted in recent MHRA reporting estimates that delayed diagnosis and limited treatment options for rare conditions cost the NHS £340 million annually, with £4.7 billion incurred in health-related disability support and £14.9 billion lost in economic productivity.

Only 1 out of 20 rare conditions have an approved treatment or medicine to help. Small affected population numbers make it hard to gather enough evidence and push up costs, while current assessment processes are better suited to common or short-term illnesses than to the lifelong conditions many rare conditions are.

Rare conditions receive only around 7% of the NIHR and MRC's combined research budget, despite representing a fifth of the population's lifetime health burden. Underinvestment in evidence compounds underinvestment in care, and the economic cost compounds with it.

'Too many of us have deteriorated drastically while awaiting diagnoses, unable to access life-improving treatments. Too many of us have a plethora of consultants and specialists to deal with in a fragmented way. Too many HCPs do not understand our conditions and try to fit us into the box of their own knowledge rather than listen to our experience. Affected person, Wales

The only pathway available to husband is to go through A&E every time he relapses, before getting the specific medication once admitted. This is tedious, time wasting and stressful. A much better pathway would be to provide the medication at a point when relapse just occurs (as soon as a blood test indicates this) or soon after, and avoid A&E completely. Carer, England

**1 in
17**

**people are affected
by a rare condition
at some point in
their lives**

'Regardless of diagnosis or rarity of disease... not relying on the parent to relay info and remember what happened and chase when things should be going on or followed up would make a world of difference to us.' Carer, Scotland

'As a person with a chronic rare condition, I cannot access services or diagnosis. My wait time so far has been 4 years. Health professionals having more knowledge makes no difference because anyone who offers to help me is told there is no funding.' Affected person, Northern Ireland

THIS IS THE TIME TO ACT

Strategically, the UK rare conditions landscape presents each nation with a set of needs and opportunities. While it represents an underserved population with urgent clinical needs, it is simultaneously a unique opportunity for the UK life sciences sector.

Advanced therapies, genomic medicine, and precision health are transforming rare conditions into a frontier of modern medical innovation. The UK could very well be a potential leader in this space, but delivering these breakthrough therapies requires more than regulatory pathways. It demands a fully functioning rare conditions ecosystem spanning early diagnosis, interoperable data, and connected clinical networks. Without this infrastructure, life sciences breakthroughs cannot reach people with rare conditions, and global investment will migrate elsewhere.

The current UK Rare Diseases Framework ends in January 2027. As each home nation undertakes major health system reforms such as England's 10-Year Health Plan and parallel programmes in Scotland, Wales, and Northern Ireland, Ministers have a unique opportunity to embed rare conditions into the future of healthcare.



A VISION FOR THE FUTURE

Delivering a better future for the UK rare conditions community is achievable, affordable, and entirely within our grasp.

Through Genetic Alliance UK's extensive multi-stakeholder Future for Rare campaign, there was overwhelming consensus that any future rare conditions policy must be built on the principle of driving equity for people living with rare conditions.

This means that people affected by a rare or suspected rare condition, should have:

- equitable, ongoing access to timely diagnosis;
- specialist expertise for clinical management;
- high-quality services and care coordination;
- approved treatments at the earliest appropriate opportunity;
- research opportunities;
- holistic coordinated support;
- advocacy and information to navigate health and wider public services and the support to live well throughout their lives.

Access to the above should not depend on individual circumstances, where a person lives, socio-demographic characteristics, their diagnosis or presentation, the rarity of their condition, or whether condition-specific infrastructure happens to exist, nor should it require repeated proof of needs that a diagnosis alone does not convey to non-clinical assessors.

Parity of opportunity

The rare community was clear that equity does not mean delivering identical services everywhere. It means parity of opportunity, through clear pathways, effective signposting and timely referral.

A system in which self-advocacy is the price of good care is not an equitable one. Families should not have to act as clinical coordinators or relentless self-advocates to achieve the basic standard of care. Defaulting system navigation to individuals affected by rare conditions magnifies inequity, leaving those without the resources, capacity or confidence to fight for care at a severe disadvantage.

Achieving parity of opportunity requires embedding equitable pathways directly into health infrastructure. By establishing formal connections between primary care, regional hospitals, and national centres, clinical expertise can travel to the patient. Standardised care coordination and clear referral pathways will help to ensure every individual receives timely diagnosis, appropriate treatment, and lifelong support regardless of their postcode or condition.

Reducing inequity

Rare condition policy must actively reduce inequity, not merely avoid worsening it. Existing inequity for people living with rare conditions operates across multiple dimensions, including disparity between rare and common conditions, variance between well-resourced rare conditions and ultra-rare or non-genetic diagnoses, and systemic barriers linked to geography, ethnic background, socio-economic status, or other characteristics amplified by a rare condition.

Tackling these disparities requires structural action rather than incremental adjustments. Equity should be driven through systemic change that shifts mindsets, updates health data infrastructure, and alters the broader landscape through intentional policy and service development. At the same time, national strategy must actively seize immediate opportunities for specific cohorts within the community, ensuring that rapid advances in genomics, diagnostics, and life sciences translate directly into improved clinical care for every person with a rare condition across all four nations.



Delivering for the UK

This is a vision for a United Kingdom where research and innovation for rare conditions are actively encouraged, funded, and embedded as a national priority.

Advancing clinical trials, genomic innovation, and targeted therapies directly transforms outcomes for people affected by rare conditions while attracting high-value global life sciences investment, building resilient health data infrastructure, and driving wider economic productivity. By building an environment that accelerates discovery and streamlines delivery pathways, a forward-looking rare conditions policy delivers profound, long-term benefits for the economy, the health service, and the entire UK population.

This is the standard set by the rare conditions community and it forms the basis of the recommended strategic actions in this report. To bridge the gap between principle and practice, ministers across the four nations must take urgent strategic action now, adopting these solutions across all four nations to build a health system that delivers equitable outcomes for every person and family affected by a rare condition.



Broad reform programmes are, by design, built around the needs of the many. Long-term conditions strategies, priority pathways and neighbourhood models are organised around common conditions with predictable presentations and a large number of people affected. With more than 7,000 rare conditions, each affecting a small and dispersed population, no generic pathway can capture the range of needs involved, and no local team will see enough people with any one condition to build expertise alone. Without dedicated policy, people with rare conditions risk falling through the gaps between strategies, services and nations, and the inequities they already face will be built into the new structures rather than designed out of them.

At the same time, a renewed rare conditions policy for the UK can help each nation deliver its own priorities. Faster diagnosis is prevention. Standardised coding and connected data are the foundation of digital transformation. UK-wide registries and trial cohorts give every nation the scale that life sciences investment depends on. Each nation has already developed strengths, from Wales's National Clinical Lead and SWAN clinic to Northern Ireland's undiagnosed neuromuscular clinic pilot, Scotland's CARDRISS data and healthcare professional awareness initiatives to England's undiagnosed condition pilot centres, support for rare conditions research and the inclusion of rare conditions in the NHS England Core20PLUS5 programme. A four-nation Framework allows these models to be shared and built upon, rather than duplicated or lost.

***'Rapid evidence generation. Rapid science. Rapid data. All on a national and four-nation approach, because we need numbers for rare to succeed.'* Healthcare Professional, England**

Delivering for each nation

Each UK nation is reforming its health service and setting ambitions for prevention, digital transformation and life sciences growth. A renewed policy for rare conditions in the UK does not compete with this work, and rare conditions cannot simply be absorbed into it. Rare conditions require a dedicated, four-nation approach.

WHAT CAN A UK RARE CONDITIONS POLICY DELIVER FOR EACH NATION?

Health policy is devolved, and each nation has set its own priorities for reform. By pooling expertise and data across the UK, it gives every nation the scale needed to improve diagnosis, care and research. That makes a Framework in each nation's interest. Below, we set out how a rare conditions policy can help each nation deliver its own health goals.

Wales

Healthcare priorities

A renewed UK rare conditions policy could align with the Healthcare Inspectorate Wales Strategy and NHS Wales Performance Framework to support early intervention and reduce health inequalities. Linking the Digital Rare Care Centre and Rare Disease Data Dashboard to UK coding would strengthen service planning. Continuing to support the work of the SWAN clinic and extending specialist networks into primary and emergency care, supported by the Strategic Genomics Workforce Plan and Mental Health and Wellbeing Strategy 2025–2035, would enable earlier diagnosis and psychological support.

Life sciences and economic priorities

Wales contributes to international research through the James Lind Alliance Priority Setting Partnership and a national biobank. As Wales cannot build trial cohorts for all rare conditions alone, four-nation networks could combine patient cohorts across the UK. This would widen trial access, encourage commercial investment and strengthen Wales's appeal to life sciences investors.

England

Healthcare priorities

A renewed UK rare conditions policy would enable delivery of the 10-Year Health Plan for people living with rare conditions. Integrated neighbourhood teams that link with specialist networks and implement NICE quality standards would improve care coordination and support the shift from hospital to community, reinforced by continued work to reflect rare conditions in Core20PLUS5. Integrating rare conditions data would strengthen the single patient record, and a National Clinical Lead would provide accountability throughout NHS England reform. The policy would also ensure that rare conditions are built into the National Care Service from the outset, help shape the Mental Health Strategy for England, and that education and employment policy is developed with people living with rare conditions in mind.

Life sciences and economic priorities

A renewed policy would provide a framework for delivering the Life Sciences Sector Plan. Rare conditions are among the areas with the greatest potential for breakthroughs in bioscience. Research registries, clinical trial cohorts, the Genomic Medicine Service and the MHRA's facilitative approach to regulation all depend on scale, which is where we need to maximise the potential. Four-nation collaboration would allow the UK to combine patient cohorts, widening access to trials and strengthening the life sciences economy.

Scotland

Healthcare priorities

With the Programme for Government 2026–2031 proposing to replace 14 territorial Health Boards with two Strategic Health Boards, a renewed policy could build rare condition pathways and services into new structures from the start. It would complement the proposed Long Term Conditions Framework, which is currently limited in scope for rare conditions, by ensuring people do not fall through gaps, and support delivery on the commitment to strengthen support for children with long-term conditions. Consistent coding, TURAS e-learning and CARDRISS data would identify people sooner and connect rural and island communities to expertise through UK-wide networks.

Life sciences and economic priorities

Rare condition trials need more patients than any one nation has. UK-wide registries would give Scotland that scale, supporting the Programme for Government commitment to grow clinical trials capacity and strengthening the triple helix of NHS Scotland, academia and industry. Better evidence on outcomes would also inform Scottish Medicines Consortium decisions, helping to attract investment.

Northern Ireland

Healthcare priorities

With Assembly elections planned, a new policy would provide continuity and stability as the Northern Ireland Rare Disease Action Plan is delivered. Northern Ireland is shortening the route to diagnosis through its undiagnosed neuromuscular clinic pilot and virtual MDT for complex paediatric cases. A renewed Framework would build on these successes faster, drawing on proven UK models, so more people are diagnosed sooner. With under two million people, Northern Ireland cannot sustain expertise in every rare condition alone. Four-nation networks and the NICE quality standard could support new service specifications and cross-border access when required.

Life sciences and economic priorities

Collecting patient-reported outcomes and dynamic consent through My Care could deliver more if the data links to UK registries, and the Framework is the opportunity to drive that link. As part of UK-wide networks with access to trial cohorts, a Rare Disease Centre of Excellence could make Northern Ireland more attractive to funders and industry. Priorities noted through stakeholder consultation for the UK framework helped secure research funding to address unmet needs.

RECOMMENDATION 1: POLICY RENEWAL AND CLINICAL LEADERSHIP

The UK Rare Diseases Framework ends in January 2027. It has built momentum and foundations that would be costly to rebuild: national conditions registries, the UK Rare Disease Research Platform, care coordination models and, most recently, the NICE quality standard for rare diseases. If the Framework lapses without a successor, these assets lose the policy structure needed to sustain and expand them.

A successor must remain UK-wide. No nation alone has the rare population numbers, clinical expertise or research capacity to plan services, design trials or build an evidence base for every rare condition. A four-nation approach allows expertise to be pooled, data to be shared across borders and patients to be referred along common pathways. It avoids four health systems duplicating the same effort and gives the UK the scale that clinical trial sponsors look for, supporting the ambition to lead Europe's life sciences economy by 2030.

The timing matters. Each nation is redesigning its health system, through England's 10-Year Health Plan and parallel programmes in Scotland, Wales and Northern Ireland. A renewed policy can ensure these reforms account for people with rare conditions from the outset. That means making them identifiable in digital systems and connecting them to coordinated pathways for diagnosis, lifelong care and treatment. Building this in now costs far less than adapting systems later.

The Future for Rare campaign has identified a clear, evidence-based vision for the successor to the UK Rare Diseases Framework. It offers governments a ready foundation to build on.

Clinical Leadership

Accountability for delivery of a new rare conditions policy should sit with a senior, named National Clinical Lead for Rare Conditions in each nation. These leads should hold authority comparable to the National Clinical Directors who have driven improvement in cancer care in England. Wales has already shown that the model works for rare conditions: its National Clinical Lead for Rare Diseases coordinates care and connects local services across the nation.

To be effective, these roles need three things:

1. They should sit within the health service rather than be political appointments, so that they provide continuity across changes of government.
2. Their remit should cut across specialties, linking clinical care with research and with public services beyond health, because rare conditions do not belong to a single therapy area.
3. They should be properly resourced, with a budget, convening power and a network of named counterparts at regional, health board and integrated care board level.

Oversight should be built in from the start. Independent oversight boards and annual audits need dedicated funding rather than relying on discretionary staff time or goodwill. People living with rare conditions, their carers and support organisations should be funded partners in designing, delivering and evaluating policy, with lived experience treated as evidence.

Policy renewal and named leadership provide ministers with a practical, low-cost framework to protect existing public investment, reduce duplication, and hold the system accountable for care quality. In doing so, they create the ideal platform to expand this scope, unlocking vital opportunities to transform care for people with rare conditions while driving wider economic growth through health system efficiencies and life sciences innovation.



RECOMMENDATION 2: DATA VISIBILITY AND SYSTEM-WIDE DIAGNOSTICS

Many people with rare conditions cannot currently be found in NHS data. Routine clinical systems use broad coding frameworks that lack specific descriptors for most rare conditions, so individuals are recorded under generic categories or miscoded. Health services therefore cannot reliably say how many people have a given condition, where they live or what care they receive. This gap undermines service planning, trial recruitment and the measurement of outcomes. It also makes it difficult to show whether policy is working.

A resourced core data offer

A significant proportion of people living with rare conditions are invisible in health service data. Routine clinical systems rely on high-level coding frameworks, and many rare conditions have no specific descriptor.

Governments must establish a resourced core data offer, underpinned by national condition registries, genomic infrastructure and newborn screening data, and supported by clear policy frameworks. This starts with embedding standardised terminology, such as ORPHA codes and specialised ontologies, directly into electronic health records during procurement and updates. Doing so avoids the higher cost and operational friction of retrofitting later.

Clinical datasets must interoperate across settings and national borders, so it can support automated care alerts, genomics integration, research analysis and early intervention. People undergoing diagnostic investigation should be tracked as a distinct cohort, so their care stays coordinated and they can access support before a formal diagnosis. Underpinning all of this, transparent governance with clear standards of privacy, security and public benefit will sustain public trust and high participation in research registries.

Because rare conditions involve small, dispersed populations, they represent the most demanding test of health data systems. Structuring data infrastructure to capture rare conditions effectively improves data quality, coding accuracy, and interoperability across the entire health service.

'Alignment of data is crucial. Patients are getting treated by different consultants nationally and a consensus on how to treat, diagnose and obtain data would vastly improve unknown quantities such as natural history.' Researcher, Scotland

'It is increasingly clear that there is a need for better data collection, understanding of natural history and response to treatments, and comparison to quality standards.' Healthcare professional in Scotland



Diagnostic capability across the system

The UK has strong genomic infrastructure and specialist expertise, but diagnostic capacity remains concentrated in regional centres. Genetic testing is available for fewer than half of even the better-understood rare conditions, and the many rare conditions that are not genetic often lack a clear diagnostic pathway. A person with a rare condition may have a route to diagnosis that too often depends on where they live or which specialty they see first. Delayed diagnosis and limited treatment cost the NHS an estimated £340 million a year, before wider costs to disability support and productivity.

Policy should shift from funding isolated diagnostic initiatives to building lasting capability across the system. Awareness of rare conditions, genetic and non-genetic, should be part of training and CPD, particularly in primary care, where most diagnostic journeys begin. Genomic testing should be standardised across specialties, following clinical need rather than point of entry. Care and support should extend to people whose symptoms suggest a rare condition before diagnosis is confirmed. Diagnosis, data, research and service design should be connected, so each diagnosis improves the speed and accuracy of the next.

Taken together, these measures shorten the diagnostic odyssey and spare families repeated tests and avoidable admissions. Faster, more targeted support improves quality of life while easing pressure on the NHS. They also give life sciences partners the well-characterised data needed to accelerate research and investment, underpinning every other element of a renewed policy.



‘Advances in diagnostics such as improved gene panels can help non-specialists diagnose a rare disease without any specific knowledge of that condition. But this only works on a practical basis when access is universal & the time taken to deliver a result permits meaningful clinical intervention. Turnaround times in genomics are currently failing to achieve this, and a lack of genetic counsellors compounds the problem.’ Industry representative, England

‘Capacity in clinical genetics services should be expanded through workforce growth, clearer referral pathways, and reduced regional variation. Genetic testing and counselling, including specialist family planning advice, should be timely, equitable and integrated into routine care where appropriate...Ensure fair and consistent access to assisted reproduction and reproductive choices. Clear national guidance and equitable commissioning for assisted reproduction, including IVF and PGT-M, are needed to avoid postcode variation.’ Support organisation representative, England

‘Due to lack of awareness of rare conditions, it took us nearly 10 years to get a genetic diagnosis. I strongly believe that increased awareness amongst GPs, paediatricians, AHPs and families would improve people’s lives significantly.’ Carer, England

RECOMMENDATION 3: SUSTAINABLE SERVICE NETWORKS AND SYSTEM EVALUATION

People with rare conditions often have complex needs that span several specialties, settings and regions. At present, coordinating that care depends heavily on individual clinical champions, or falls to patients and families, who must find their own way through disconnected services. Over a quarter of even the better-understood rare conditions have no specialised NHS service, and fewer than a third have NICE guidance. The result is variation in care that reflects postcode, persistence and resources more than clinical need.

Sustainable service networks

The UK already has the foundations of a better model in its highly specialised services, managed clinical networks and Rare Disease Collaborative Networks. The task is to connect, resource and expand them, linking primary care, local providers, regional centres and national expertise. Specialist advice should reach local clinicians through virtual clinics, shared care arrangements and clear referral pathways. People would then travel long distances only when hands-on specialist care adds real value. Shared standards, pathways and data protocols would reduce unwarranted variation between areas and nations.

Networks must also be resilient. A service that rests on one clinician can leave a whole patient group without expert care when that person retires or moves on. Clinical academic training pathways, succession planning and specialist retention should be built into how networks are funded. The roles that hold networks together, including care coordinators, psychological support and network managers, should be funded as core NHS provision rather than through short-term grants.

'I actually think that the way rare disease is treated in Centres of Excellence with MDTs could be pushed as a wider model for healthcare. Where it works well, some rare condition patients have a far superior experience.' Support organisation representative, England

The evidence for this is encouraging. The Rare Care Centre in Perth, Australia, reports a return of

four Australian dollars for every dollar invested in care coordination. In the UK, the SWAN clinic in Wales has shown measurable reductions in secondary care use since introducing coordinators.

Networks should also link routine care with registries, research programmes and trial pipelines, so that taking part in research is offered to patients as a matter of course.

Measuring what matters

There is no consistent, UK-wide measure of what it is like to be diagnosed, treated and cared for with a rare condition. Without one, it is hard to judge whether policy is succeeding, to compare areas or to see where services fall short. The national cancer patient experience surveys run across the four nations and show how a regular survey can become a trusted tool for improvement and accountability.

Ministers should commit to an annual rare condition patient experience survey. A weighted sample across a defined set of conditions offers a credible starting point that can broaden over time. The Health Data Research Service should be funded to lead the work, drawing on data already collected wherever possible to limit the burden on families and clinical staff. Results should be read alongside staff experience and service activity data and benchmarked against the NICE quality standard. The method should adapt as new therapies and diagnostic technologies arrive.

Networks and measurement reinforce each other.

The survey shows where networks are working and where people are falling through gaps, and well-organised networks provide the means to act on its findings. Together, they give ministers a shared, evidence-based view of progress and a clear account of what has changed.

'A consistent national set of measurable outcomes or performance indicators for rare diseases would support tracking progress against the priorities set out in the framework.' Industry representative, England

DELIVERING BETTER OUTCOMES FOR PEOPLE AFFECTED BY RARE CONDITIONS

People living with rare conditions have made their priorities clear: they want to be believed, valued, and treated as partners in their own care. They need timely diagnoses, specialist expertise, coordinated support, and equal access to treatment, regardless of where they live or how rare their condition is. Above all, high-quality care must become the default standard, not a reward for endless self-advocacy.

What can we achieve if we act now?

The recommendations outlined in this report are not standalone policy measures; they build the essential foundation required to deliver these exact outcomes for the 3.5 million people in the UK affected by a rare condition. By establishing a UK-wide Framework, we create the basis to capture and share best practice, explore joint initiatives, and ensure broader health and social care policies in each nation are capable of delivering for people affected by rare conditions.

Supported by national clinical directors, a core data offer, and sustainable clinical networks, this infrastructure gives all four nations the capability to measure progress, collaborate effectively, and deliver equitable care across the UK.

Timely and meaningful diagnosis

What people need: People need a diagnosis that is timely, meaningful and equitable, because it opens the door to coordinated care, informed choices and lifelong support. They need that diagnosis communicated with care, with access to genetic counselling, clear information and early contact with support organisations. And those who remain undiagnosed, or whose condition is suspected but unconfirmed, need care and support without waiting for a label.

How governments can deliver it: Investment in diagnostic capability and sustainable networks, set out in our foundations, gives services the capacity to reduce time to diagnosis. Governments can set targets to shorten that time and to narrow variation between nations, regions and population groups. Pathways can be designed so that support follows need rather than a confirmed diagnosis.

Why it matters: Faster diagnosis shortens years of uncertainty for families and provides them access to timely healthcare interventions and support. It also saves the NHS the cost of repeated tests, unnecessary appointments and emergency admissions during the diagnostic odyssey.

The challenge is that many people are still waiting too long for a diagnosis, so the focus now needs to shift to delivery: making sure the infrastructure, workforce and commissioning arrangements are in place to turn ambition into consistent outcomes.' Industry representative, England

'I think we need to work to improve provision for the many children whose diagnosis remains unclear, and the experience of 'the diagnostic odyssey' within the reality of the current limits of genetic testing. The emotional impact of this must be addressed.' Healthcare professional, England

Rare care in the NHS

What people with rare conditions need: People with rare conditions, whether genetic or non-genetic, need equitable access to timely diagnosis, specialist expertise and holistic support to a clear and consistent standard, wherever they live in the UK, whoever they are and however rare their condition. This means clear pathways, effective signposting and timely referral, so they have the same opportunity and quality of care as people with more common conditions. They need good information, a real say in decisions about their care, a named healthcare professional who knows them, and an assessment that looks at their whole life, not just their symptoms. They also need to be recognised within the NHS, so that services know who they are and plan around their needs.

How government can deliver it: Governments can adopt the NICE quality standard for rare diseases as the benchmark for a renewed rare conditions policy in each of the four nations, and report progress against it publicly. They can set national standards for access to specialist expertise, with clear referral pathways for both diagnosed and undiagnosed patients, and make funded, sustainable networks, including expanded Rare Disease Collaborative Networks, core NHS infrastructure. These networks connect primary care, local services, regional centres, national expertise and research, moving knowledge rather than unnecessarily moving patients. They can be made resilient through funded leadership and coordinator roles with protected time and clear succession plans, while virtual multidisciplinary teams and specialist advice services link primary, secondary and tertiary care. Rare conditions education, decision-support tools and rapid access to advice can be embedded across the workforce, and standardised coding with an NHS-wide rare conditions flag in electronic health records can ensure people are identified and their needs are visible wherever they are seen.

Why it matters: Networked care spreads scarce expertise further and reduces unwarranted geographic and socio-demographic variation. Earlier referral and better-informed clinicians mean fewer avoidable emergency admissions and less fragmented care. A shared, independently developed benchmark and better NHS data give ministers a ready-made measure of success and the evidence to plan services, target investment and demonstrate improvements.

Data and digital

What people with rare conditions need: People with rare conditions need to be digitally findable and not lost in the system. Inconsistent identification, coding and registration mean they are too often invisible to care, research, clinical trials and service planning. Rare condition communities also need to be partners in decisions about what data matter, how they are governed and how they are reused.

How the governments can deliver it: Governments can establish a well-governed core data offer across all four nations. It would focus on sustainable national condition registration services and genetic screening, enabled by appropriate legislation and policy. Governments can agree core data standards, coding and ontologies, and embed rare condition requirements into electronic health record source systems from the outset, without requiring one single UK-wide system. Sustainable funding, governance and accountability must accompany these commitments.

Why it matters: High-quality, connected data are a prerequisite for future genomic and AI-enabled approaches. Connected data can support earlier identification, service planning, research readiness, recontact and clinical trial opportunities. Treating rare condition data requirements as an exemplar rather than an afterthought can strengthen the wider health system.

Research

What people with rare conditions need: People with rare conditions need research that leads to better care and new treatments, with equal opportunities to take part wherever they live. UK rare condition research is fragmented across nations, funders and conditions, and funding tends to favour conditions seen as treatable or with a clinical champion.

How governments can deliver it: Government should connect existing infrastructure into rare disease Research and Innovation Hubs, with at least one in each nation within three years. Investment in population-based registries, and linking genomic data to health records across all four nations within five years, would give these hubs the data they need. Rare conditions should be named explicitly within the Health Data Research Service and in national life sciences and AI strategies and investment programmes. This should be backed by a cross-nation funding approach, with long-term investment across the whole research pathway that can follow the research across UK borders. Patient involvement and workforce development should be built in from the start. The UK should be a full and active partner in international rare condition collaboration, particularly for research, to build and share knowledge, optimise care, while accelerating the pace of global research, regulatory and innovation agendas. This is crucial to avoid duplication of effort and resources.

Why it matters: No single nation has a large enough population to sustain trials, registries or cohorts for every rare condition. Working across the four nations gives research the scale it needs. Funding routes currently differ between nations, so shared hubs, shared data standards and coordinated investment would help funding go further and give researchers and patients in every nation the same opportunities. A connected research base also makes the UK more attractive for trials and for launching the treatments they produce.

‘Further national and international investment into research of rare conditions [is needed]. The previous wave of investment seems to have slowly died.’ Researcher, England

Advocacy, rights and representation

What people with rare conditions need: People living with rare conditions and their carers need to be recognised as valued partners in shaping policy, research and services. They need equitable access to advocacy, coordinated support and information to navigate health and wider public services. That access should not depend on diagnosis, geography or the availability of condition-specific infrastructure.

How the governments can deliver it: Governments can adopt consistent standards for meaningful involvement across national rare condition programmes. Those standards should treat lived experience as a primary form of evidence alongside clinical, research and service data. Governments can include people with lived experience in governance and oversight groups and publish progress against agreed measures. They can sustain advocacy infrastructure by recognising patient organisations as strategic partners. Flexible involvement, targeted outreach to underserved communities and accessible communications can widen participation.

Why it matters: Advocacy, rights and representation are the means through which people living with rare conditions can exercise choice, influence decisions and access equitable care and support. Implementation must not disproportionately benefit those who already have greater opportunity to participate and are able to self-advocate.

it is most important to ensure people are provided with the assistance needed to make their voices heard, not just to receive lip service.’ Affected person, Wales

Treatment access

What people with rare conditions need: People with rare conditions need timely, equal access to new treatments, wherever they live in the UK. Around 95% of rare conditions have no treatment, and where medicines do exist the UK is falling behind. In 2025, England ranked 11th in Europe for the availability of non-oncology orphan medicines, and Scotland 9th.

How governments can deliver it: Government should make the UK a first-choice launch market for rare condition treatments. This means dedicated regulatory and HTA routes for rare and ultra-rare conditions across all four nations, which recognise the full value of treatments to people affected, families, the NHS and the economy, and manage the uncertainty of rare condition evidence rather than penalising it. Strong orphan drug incentives, an improved Early Access to Medicines Scheme, and coordinated four-nation horizon scanning and commissioning would give developers a clear, predictable route from licence to patient.

Why it matters: Companies launch treatments and place trials where they see a viable route to market. Without one, the UK risks losing treatments altogether, and some approved treatments have already been withdrawn. A single, predictable UK route would make the UK an attractive place to launch, bringing earlier access for patients, more clinical trials and continued investment in UK life sciences.

‘Access to medicines should be a standalone priority area. This is an area the UK is performing terribly in comparison to the rest of the world.’ Affected person, Northern Ireland

‘Please can we have access to the medications that have been approved in most of Europe and North America. The clinical trials have been transformative for the children who have had a space on them.’ Support organisation representative, England

Collaboration

What people with rare conditions need: People living with rare conditions need advocacy, navigation, meaningful involvement and accountability to be consistently embedded across the rare conditions landscape. That requires aligned action across local services, communities, devolved systems and national policy bodies, rather than a single owner or a single UK-wide system.

How governments can deliver it: Governments can set common UK-wide expectations that respect devolved delivery. Ownership can be shared at three levels. Local ownership covers identification, coding and data capture at source. Co-ownership with rare condition communities shapes governance and use. National ownership embeds rare condition requirements into infrastructure across the four nations. Principles should be specific enough to drive action while allowing flexibility in implementation across devolved health systems and different community contexts.

Why it matters: Rare condition policy areas depend on each other, including governance, care coordination, data, advocacy and non-healthcare interventions. The working groups identified tensions between national consistency and local flexibility, and between ambition and deliverability. These will need to be closely monitored during implementation.

‘International engagement and collaboration is crucial. However, it is still challenging, post-Brexit, to know what we can and cannot engage with. Our RD-IMAG is important here. We need a strong focus on national registries, disease registries, health data, and what can be pooled (and how) to advance diagnosis, treatment, care, research etc.’ Researcher, England

Support for living well with a rare condition

What people with rare conditions need: Living well is the outcome that matters most to people and families. They need support that spans healthcare, education, social care, employment and welfare. They need someone accountable when things go wrong between services. They need professionals who understand rare conditions, so they no longer have to explain a complex diagnosis at every encounter or prove again and again what their needs are. And they need public services flexible enough to respond to needs that are complex, multi-system and fluctuating.

How governments can deliver it: Governments can build rare condition awareness into initial training and continuing professional development for teachers, social workers, employment advisers, benefit assessors and health professionals, following the established model of equality, diversity and inclusion training. They can ensure that diagnostic reports, clinical assessments and care plans produced by the health service are recognised across education, social care, welfare and local government.

From first contact, every family can receive a proactive pack setting out their rights and the support available, with a named non-healthcare coordinator in the multidisciplinary team to help them access it. Where families choose and trust them, charities and patient organisations can be recognised as partners in delivering that role. Success will be seen in a rising proportion of families accessing non-healthcare support, with uptake tracked by geography, ethnicity, language, disability and income so that no community is left behind.

Why it matters: For people affected by rare conditions, this means the stability to stay in education, to remain in or return to work, and to take part fully in family and community life. For government, it means fewer avoidable escalations, less duplicated administration across departments, and more people able to stay in employment and contribute to the economy.

‘Support for families and carers is a significant gap, particularly access to respite and emotional support. Transitions into adulthood, education and independent living are not well supported... the system is not designed around the reality of rare conditions.’ Affected person, England

Mental health and wellbeing

What people affected by rare conditions need: People need psychological support from before diagnosis onwards, not only when they reach crisis. Parents, carers and siblings need to be recognised in their own right, because a rare condition affects the whole family.

How governments can deliver it: Governments can embed psychologically informed, trauma-aware and whole-family care as standard across rare condition services. National mental health policies can explicitly recognise people with rare conditions and their families, with carers' assessments and sibling support designed around the realities of living with a rare condition.

Why it matters: Early, preventative support costs less than crisis care. Governments will be able to show people reporting that they feel better supported year on year, alongside a fall in crisis-driven admissions.

‘Being diagnosed with a rare condition is isolating and frightening. Along with dealing with the physical, the mental toll is horrific. Being able to reach out and to find information, links to any information, helps a little.’ Affected person, Northern Ireland

Coordinated care

What people with rare conditions need: Coordinating care too often falls to families by default, and good coordination has become a reward for those able to advocate loudest. The CONCORD1 definition sets out what people should expect instead: everyone involved in a person's care, including the person, their family and their professionals, working together to avoid duplication and achieve shared outcomes throughout life. That means care coordinated across specialities, settings and locations, across conditions that affect several parts of the body, and through every move between services. It should be family-centred and holistic, covering medical, psychological, educational and vocational needs, with equal access regardless of diagnosis, circumstances or where people live. People also need transitions treated as a continuous process, not a single handover. These should be anticipated from diagnosis and planned for throughout a person's life journey.

How governments can deliver it: Governments can make care coordination a commissioned and accountable part of rare condition care, rather than a hidden burden on families. They can adopt the CONCORD1 definition as the shared foundation across all four nations, and embed the learning from the RareCare and CONCORD2 evaluations of the current Framework in what comes next. In line with the NICE quality standard, every person with a rare or undiagnosed condition can have a named professional to support their care, written into service specifications. Governments do not need to start from scratch on transition, because proven models already exist. Structured programmes such as Ready Steady Go and T-KASH, and training led by transition coordinators can be built into the care coordinator role as standard. Regional Rare Conditions Leads can oversee services, standardise pathways and handle complaints. Interoperable systems can ensure records, care plans and emergency information follow the person. A national directory of services can help professionals signpost correctly. Patient organisations can be commissioned as partners rather than left to fill gaps. Succession planning in every service specification can ensure coordination never depends on one individual.

Why it matters: Care coordination has received the least investment and improvement under the current Framework, despite its central role in safety, experience and outcomes. Proactive coordination prevents fragmentation, reduces duplicate tests and appointments, and cuts the failures at transition that lead to deterioration and emergency care. The evidence of return is strong. The Rare Care Centre in Perth, Australia, a service dedicated to care coordination, reports a return of four Australian dollars for every dollar invested. In the UK, the SWAN clinic has shown measurable benefits in secondary care use since care coordinators were introduced. Coordination is also a patient safety requirement, because failing to act on specialist advice in emergency care can have life-threatening consequences.

'No one NHS service taking the 'lead' in my care so I am struggling to get different healthcare teams to work together and arrange further appointments and referrals.'
Affected person, Wales

'There is no care coordinator, no specialist care, the services we need are not commissioned at all. All the services don't talk to each other so we have to be administrators, legal specialists, advocates and financial managers as well as carers.' Carer, England

'Communication between NHS services within different Health Boards and also with care between Wales and England relies on me having to take copies of hospital letters to the specialist hospital in England. Surely there should be a better way in 2026 for hospitals/the NHS to share and access hospital records instead of relying on a patient to have to do additional admin burden.'
Affected person, Wales

There is also an opportunity to better reflect transition points across the patient journey, including the transition from paediatric to adult care, where continuity of care can be challenging.' Industry representative, England

THE TIME FOR ACTION IS NOW

Genetic Alliance UK is calling on Ministers to act urgently to:

- 1. Renew a UK-wide policy framework and appoint a National Clinical Lead for rare conditions in each nation.**
- 2. Resource a national core data offer and embed diagnostic capability across the health system.**
- 3. Organise care through sustainable clinical networks and establish an annual national patient experience survey.**

Every outcome in this report can be delivered and tracked by acting on these three recommendations. The annual patient experience survey and core data offer, read alongside staff experience and service activity data and benchmarked against the NICE quality standard, will measure progress across all four nations. Annual reports from each National Clinical Lead will give a clear public account of what has changed.

Success should be judged not by processes completed but by whether people feel valued, supported and able to live well. That must be true for everyone, not only those who advocate loudest. To achieve this, people with lived experience must help shape how success is measured and see their voices change decisions.

Gaps in access and outcomes must narrow across nations, regions and communities, including for those still awaiting a diagnosis. A system that achieves this will also be more effective and efficient, delivering faster diagnosis, fewer emergency admissions, more carers in work and greater research investment. And because rare conditions test every part of the system at once, with often complex, lifelong needs that cross specialties and services, getting it right for rare lays the foundations for getting health and social care right for all.

With the UK Rare Diseases Framework ending in January 2027, Ministers must decide what comes next. The current Framework has laid strong foundations, from national disease registries and the UK Rare Disease Research Platform to specialised care coordination models and a growing research ecosystem. The next policy must build on them.

All four nations are reforming their health systems, and each has ambitious plans for life sciences. Design these reforms and plans around common conditions alone, and existing inequities will be locked in. Build rare in from the start, and they will deliver a better, more equitable healthcare system for everyone.

For the 3.5 million people living with a rare condition in the UK, every day of delay means longer diagnostic odysseys, fragmented care, avoidable crises and families left to navigate complex systems alone. Closing the gap between what our health services could offer and what people actually experience cannot wait.

Genetic Alliance UK and the rare conditions community stand ready to work with Ministers across all four nations to deliver a future where diagnosis is swift, care is joined up and no one is left behind because of their rare condition.

An equitable future for rare conditions is within reach. The time to act is now.

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Working group co-chairs

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Prof. Tim Barrett, University of Birmingham	Rare care in the NHS
Kerry Leeson-Beevers, Alström Syndrome UK	Care coordination
Zoe Morrison, NHS Wales	Care coordination
Rosie Lindup, BioIndustry Association	Access to medicines
Sue Millman, Ataxia UK	Access to medicines
Prof. AJ McKnight, Queen's University Belfast	Research
Liz Barnard, Kidney Research UK	Research
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Rebecca Cosgriff, LifeArc	Data, digital and technology
Toni Mathieson, Niemann-Pick UK	Data, digital and technology
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Nick Meade, Genetic Alliance UK	Governance
Sheela Upadhyaya, Rarely Ordinary	Governance

Future for Rare steering group organisations

The following organisations were represented on the Future for Rare steering group: the Association of Medical Research Charities, the Association of the British Pharmaceutical Industry, Ataxia UK, Beacon for Rare Diseases, the BioIndustry Association, Breaking Down Barriers, the British Society for Genetic Medicine, Cambridge Rare Disease Network, the Faculty of Pharmaceutical Medicine, the Federation of Specialist Hospitals, Gene People, Genomics England, LifeArc, Medics for Rare Diseases, Metabolic Support UK, the Neurological Alliance, the Northern Ireland Rare Disease Partnership, the PHG Foundation, the Rare Autoimmune Rheumatic Disease Alliance, Rare Disease Research UK, the Royal College of Paediatrics and Child Health, the Royal College of Physicians, the Specialised Healthcare Alliance and Unique.

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