GENETIC ALLIANCE UK

EQUITY for RARE

Delivering fair healthcare systems for people affected by rare conditions

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About Genetic Alliance UK

Genetic Alliance UK is an alliance of over 220 charities and support groups working together to improve the lives of people in the UK with genetic, rare and undiagnosed conditions.

Genetic Alliance UK supports people with genetic, rare and undiagnosed conditions by advocating for improved care, progressing medical research, increasing awareness and improving information and support.

We run two long standing projects:

RARE DISEASE UK

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
syndromes without a name

SWAN UK: The only dedicated support community 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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Everyone with a rare condition deserves fair and equitable care, no matter how rare their condition is. Equitable care means addressing individual needs, not treating everyone the same.

Equity may mean different things to different people, but we are united in working to end the diagnostic odyssey and securing timely access to specialised care and innovative treatments.

Learn more about Genetic Alliance UK's Equity for Rare campaign on our website.

Foreword

Over 3.5 million people in the UK live with a rare condition. For many of them, the NHS is failing to deliver equitable care. Not through design or neglect, but because the fundamental challenges of rarity - low prioritisation, low clinical awareness, and low evidence - remain unaddressed and are baked into our healthcare system.

This report confirms what our community has long felt: that people with rare conditions are navigating a system fundamentally not designed for their needs. While common conditions benefit from established clinical infrastructure, visibility, and prioritisation, those with rare conditions face a diagnostic odyssey, fragmented care, and limited access to treatments.

Our consultation with the rare conditions community found patchy care and profound variability across the UK. We found that this damaging variation is further compounded by ethnicity, gender, and socio-economic status, amplifying disadvantage for many. Some have also faced racism, misogyny, and other discrimination throughout their rare condition journey. We acknowledge the extensive work within our community exploring these intersections and have sought to highlight some of these vital contributions throughout this report.

However, this report adopts a specific focus. We examine how the defining, immutable characteristic of all rare conditions, their small patient population, is itself a fundamental driver of systemic inequity. This scarcity creates three challenges: low priority, limited evidence, and low clinical familiarity. Overcoming these is essential to delivering fair healthcare for people with rare conditions.

If it is hard to recognise the scale of this problem, that is partly because inadequacies are hidden by pockets of world-class care. The NHS is capable of delivering excellent rare condition care, we have centres of excellence, pioneering specialists, and groundbreaking research. But we have created a winner-takes-all ecosystem where a handful of rare conditions attract research funding, clinical trials, and specialist centres, while thousands of others remain in obscurity. We are concentrating excellence rather than spreading it.

This does not have to be our reality. With the UK Rare Diseases Framework ending next year and healthcare reforms underway across all four nations, we are at a decision point. We can seize this moment to integrate rare conditions into mainstream healthcare, or we risk entrenching these inequities for another generation.

Our report sets out five practical recommendations that address root causes rather than symptoms.

The questions now are: what do we actually need from a successor to the UK Rare Diseases Framework? What core principles must we demand from the four governments? This report provides answers grounded in evidence and the lived experience of people who have spent years being overlooked by healthcare systems that should serve them. As the successor framework takes form, their voices must be central to every decision. Genetic Alliance UK will continue to work to fulfill this goal for all 3.5 million people living with a rare condition in the UK.

Acknowledgements

The recommendations in this report emerged from collaboration with people affected by rare conditions, the organisations that support them, clinicians, researchers, and policymakers across the rare conditions community.

We would like to thank everyone who contributed their time, insight, and lived experience to this work. Those who shared their stories through case studies, participated in surveys, and provided invaluable feedback: your contributions have been essential in grounding this report in real life experiences. The support organisations and advocacy groups who facilitated connections and ensured community priorities remained central: your expertise shaped our approach.

Your collective voices have shaped this work and represent a community committed to driving meaningful change for people affected by rare conditions.

Executive summary

Rare conditions affect over 3.5 million people in the UK. People affected by rare conditions face profound systemic inequity, often experiencing a complicated diagnostic odyssey followed by fragmented care and limited access to treatments.

Our Equity for Rare consultation highlights the inequities our community experiences, and found that equity for the rare condition community broadly means ensuring that people with rare conditions can navigate the healthcare system with the same dignity and efficacy as those with common conditions. While common conditions are not without their own challenges, they often benefit from established clinical infrastructure, visibility and prioritisation. By contrast, those with rare conditions must navigate a system fundamentally not designed for their specific needs.

This report acknowledges that rarity is seldom a standalone challenge and it intersects with broader drivers of inequity such as ethnicity, gender, and socio-economic status. We recognise the extensive work within our community exploring these intersections and have sought to highlight some of these vital contributions throughout this report.

Our report adopts a narrower focus, highlighting that a defining, and immutable, characteristic of all rare conditions is their small patient population. We have centred this analysis on how this scarcity is a fundamental driver of inequity for all rare conditions, resulting in three systemic challenges: low priority, limited evidence, and low clinical familiarity. Overcoming these three challenges is essential to delivering a fair healthcare system for those affected by rare conditions.

Genetic Alliance UK is calling on the governments of the UK to:

1. **Deliver a UK-wide map of rare conditions:** The four UK nations must urgently fund comprehensive rare condition registries and work in partnership to map all 7,000 rare conditions. Robust national data infrastructure is not optional, it is the foundation of equitable service planning, resource allocation, and healthcare delivery.

2. **Close the evidence gap through fair research investment:** Government and research funders must correct the imbalance in research investment by directing funding towards rare conditions. Decision-makers must also reform evidence standards to recognise that uncertainty is an inherent feature of rare conditions and should not be used as a barrier to access or innovation.

3. **Mandate system-wide accountability for rare conditions:** Healthcare systems must stress-test policies, commissioning decisions, and service delivery frameworks against the realities of low-prevalence conditions. This must include systematic auditing to identify gaps, eliminate inequities, and enforce consistent standards of clinical accountability across all services.

4. **Embed rare conditions into mainstream healthcare delivery:** Current reforms across the UK healthcare system present an opportunity to fully integrate rare condition care into routine service provision. Failure to act now will entrench existing health inequalities for the 1 in 17 people in the UK that will be affected by a rare condition at some point in their life.

5. **Commit to a bold successor to the UK Rare Diseases Framework:** UK governments must commit to a long-term successor to the UK Rare Diseases Framework that delivers measurable improvements for people with rare conditions. This successor must set clear targets, and the nations must respond with adequate funding, directly addressing the structural drivers of inequity identified in this report.

About rare conditions

Rare conditions are individually rare but collectively common, with over 3.5 million people in the UK living with a rare condition. There are around 7,000 known conditions, with new conditions regularly identified through scientific progress.

7 in 10 rare conditions affect children. It is estimated that more than 50% of childhood deaths are associated with a rare condition.

In the UK, 1 in 17 people will be affected by a rare condition at some point in their life. The experience of those living with these conditions is often defined by significant inequity, beginning with the 'diagnostic odyssey'. The most recent data on time-to-diagnosis for people with a rare condition shows that 1 in 4 in the UK waited at least 3 years between the first symptoms showing and receiving a confirmed diagnosis. The same data indicates that for the vast majority (95%) of the 3 year wait, people were within the healthcare system i.e. they were actively seeking help from the health service. For many, a diagnosis remains entirely out of reach. Every year, more than 6,000 children are born with a 'syndrome without a name' (SWAN), meaning their condition is so rare that it does not yet have a name.

Inequity persists in the management and treatment of these conditions. Only 1 out of 20 rare conditions have an approved treatment or medicine to help. Where there is an approved treatment, these are mostly for rare cancers.

People living with rare conditions often have complex health needs that require support across multiple health services, which means effective coordinated care is highly valued. However, most people do not receive this support and are responsible for coordinating their own care on top of managing their own condition. Only 1 in 10 adults in the UK living with a rare condition have a care coordinator to help organise different aspects of their care. Only 1 in 10 adults have a care plan in place. Children with rare conditions fare slightly better, with 4 in 10 children having a care plan in place.

Read more about rare conditions in our facts and figures web page

Over 3.5 million people in the UK living with a rare condition

1 in 17 people will be affected by a rare condition at some point in their life

There are over 7,000 rare conditions, with new conditions regularly identified through scientific progress

1 in 4 in the UK waited at least 3 years between the first symptoms showing and receiving a confirmed diagnosis

7 in 10 rare conditions affect children

6,000 children are born with a 'syndrome without a name' (SWAN), meaning their condition is so rare that it does not yet have a name

What do we mean by healthcare equity for rare conditions?

Health equity is a fundamental principle in public health that moves beyond the concept of uniform provision of health services to focus on fairness in health outcomes. The World Health Organisation defines health equity as 'the absence of remediable differences among population groups'. It is important to distinguish this from health equality, which implies that every individual or group is given the exact same resources or opportunities. In contrast, equity acknowledges that various populations face distinct social, economic, and health barriers; therefore, resources must be distributed according to specific needs to ensure everyone can achieve their full health potential.

While rare is often used as a single label, there are more than 7,000 identified rare conditions. In the UK, a condition is considered rare if it affects fewer than 1 in 2,000 people. Some rare conditions affect thousands of people in the UK. Because of these numbers, they may be familiar to the public and the medical profession. At the other end of the spectrum many thousands of rare conditions affect fewer than 100 people in the country. Across rare conditions, scientific attention is not distributed evenly. Some conditions that have been known for decades may have established registries, natural history studies, and active clinical trials. For other conditions, there may be no active research at all. The level of support varies from rare condition to rare condition, with some having well established charities involved in providing support, research and campaigning, with others operating on a voluntary basis with limited resources and capacity. This doesn't mean some have it 'easy', the healthcare system creates different barriers for different groups.

Achieving equity for the rare conditions community is therefore not about creating a new, separate definition of fairness. It is about ensuring the universal principle of care according to need is finally extended to the 1 in 17 people in the UK that will be affected by a rare condition at some point in their life. Their needs are currently overlooked by a healthcare system that systematically prioritises high-prevalence conditions at the expense of rare conditions. This requires a shift in how the NHS operates to ensure that the baseline of care for people with rare conditions is comparable to the safety, coordination, and speed experienced by those with common conditions.

What does equity mean for rare conditions?

Between November 2025 and February 2026, Genetic Alliance UK conducted a community survey and a series of workshops exploring the theme of equity. We asked the rare community to share their experience of rare conditions and tell us what equity for rare conditions means to them.

Our findings were captured by Scottish illustrator and paramedic, Rose Matheson. This illustration reflects our findings and provides a visual representation of what equity means to people affected by rare conditions.

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


GENETIC ALLIANCE UK
WHAT DOES EQUITY MEAN TO YOU?
VISUAL MINUTES BY ROSE MATHESON @ROSESUSCITATION
FORMAL DEFINITION: "the quality of being fair and just, especially in a way that takes account of and seeks to address existing inequalities." (Oxford English Dictionary)
"Our whole community can access the same standard of care regardless of: LOCATION, SOCIOECONOMIC BACKGROUND OR ETHNICITY."
"SUPPORTING INTERNATIONAL COLLABORATION TO SHARE BEST PRACTICE and resources to increase QUALITY OF LIFE AND LIFE EXPECTANCY."
"BEING SEEN AS A PERSON NOT A PROBLEM"
"GREATER AWARENESS among healthcare professionals so that rare diseases are RECOGNISED rather than overlooked."
Help accessing Personal Indepedence Payments.
"ACCOMODATIONS So that I can be INVOLVED in the local community e.g accessible business."
"FULL ACCESS to TESTING, SUPPORT, TREATMENT + RESEARCH for GENETIC CONDITIONS."
"INCLUSION"
"Working WITH the patient/carer as part of the team, keeping them INFORMED and EDUCATED."
"Access to EXPERTISE and RESEARCH OPPORTUNITIES EVEN FOR THE SMALLEST patient groups."
"HEARD and BELIEVED."
...FINANCIAL SECURITY
EMPLOYMENT
"Parents of children with rare diseases often have to drop out of employment leading to financial challenges..."
...Although they have access to disability allowances these are cut rapidly if the child dies...
DEBT
GRIEF
...this results in parents with a loss of money + employment as well as grieving the loss of a child and no easy route back into employment."
"HONESTY regarding EXPENSIVE personalised gene therapies that may struggle to ever be made available to the majority who could benefit from them via the NHS."
TREATMENTS
"Dedicated NURSING Support."
"...To have our case reviewed at an MDT meeting (and be PRESENT where appropriate."
"...Having a SUPPORT GROUP for ADVICE and INFORMATION."
"FASTER MORE ACCURATE DIAGNOSIS."
Access to health + social care without BARRIERS
"Access to an effective TREATMENT."
"Mental Health at all Stages of the diagnostic process Support and continuing thereafter."
"Having all her needs catered for at school"
"...Not having EXCESSIVE DURATIONS OF TIME between appointments."
"COORDINATED COMPASSIONATE care that DOESN'T DEPEND on GEOGRAPHY or PERSONAL ADVOCACY"
"EVERY family having quick receipts of Post Mortem
EVERY family has equal access to cardiac screening + genetic testing
A statutory obligation to document and report on cases that remain unexplained
EVERY family must be supported
EVERY professional must be trained in Sudden Unexplained Death in Childhood"
"...Being able to have the CONFIDENCE and ABILITY to do NORMAL things in life:
Get a job.
Use public transport."
"MENTAL HEALTH OR NEURODIVERGENCE NOT TO GET IN THE WAY OF BEING LISTENED TO"
"We would ALL like access to healthcare professionals that KNOW and UNDERSTAND our conditions. However, rare diseases are RARE by definition and it is very DIFFICULT for medics to be well informed on RARE CONDITIONS. EQUITY looks like sufficient TIME with our consultants + GPs to discuss an issue... ...them to be RECEPTIVE to the issue and them having time to Research a Solution OR REACH to peers or further research and come back to you. TIME to follow up if solutions resolved an issue or improved quality of life."
"A dedicated point of contact we are so rare that there is NO 'HOME' for us within the NHS and we have to rely on generalist services e.g GP services, A+E which is not equitable to other conditions that have dedicated specialist services."
"BEING INCLUDED within a workplace within the healthcare system."
...Having access to the WIDER MDT."
"As a woman who presents as generally fit and healthy, I want to be LISTENED TO when symptoms arise..."
"ADMITTING that they DON'T KNOW"
"ACCESSIBILITY & REASONABLE ADJUSTMENTS."
"ACCEPTANCE of the EFFECT of a debilitating condition in the WORKPLACE WITHOUT it lowering expectations of your ABILITY."
"EMPLOYABILITY FLEXIBILITY"
Treated the same as any other patient
Being able to TRAVEL UNAIDED Without being frightened of tripping over, being able to read a menu without a magnifying glass."
"...Being taken SERIOUSLY when Someone presents with SYMPTOMS."
CO-ORDINATING SPECIALIST CARE"
"...that means NO ONE is DISADVANTAGED Simply because their condition is UNFAMILIAR, COMPLEX or only affects a handful of people worldwide."
"...To be treated with CARE + UNDERSTANDING and not cast aside if there's NO TREATMENT AVAILABLE."
"...A Service that is NEEDS led NOT DIAGNOSIS led."
"MEANINGFUL"
LINKED UP CONSULTATIONS
POOLING of INFORMATION
"... A service that does NOT think TREATMENT is ONE SIZE FITS ALL."
"COUNCILS Should have a statutory minimum standard of services.
SCHOOLS Should be funded to supply the level of care required including properly managed transport provision.
HEALTHCARE TRUSTS Should be measured the same for rare disease patients as any other patient, not having different standards."
"...my son's condition being RECOGNISED by the government."
"WE DON'T expect every professional to have in-depth knowledge of the condition BUT We do expect sensible decisions to be made about who is most APPROPRIATE to help us at any given time."
Making medicines EASILY ACCESSIBLE to patients when needed
"FREE ACCESS to medicines relating to the condition."
"TIMELY ACCESS to appropriate Services"
Health Assessment
Treatment
Social Supports
Financial Support
"FINANCIAL SUPPORT Where necessary, especially for items that the general population DOESN'T WANT OR BELIEVE THEY NEED." e.g PEPS inhalers
"RESEARCH OPPORTUNITIES for rare CF mutations not eligible for current treatments."
"Being treated EQUALLY and INCLUSION for work and all life activities."
"ACCEPTANCE that our SPECIFIC DIETARY NEEDS are a MEDICAL NECESSITY NOT a personal choice."
EXCLUSION
SYMPTOMS
SOCIETAL
"Add LEARNING DISABILITIES + RARE DISEASES Literature to MANDATORY TRAINING for all hospital Staff."
"Not having to FIGHT for TREATMENT."
"Feeling safe that medical Professionals are up to date with knowledge re: my condition."
"EQUITY aligns closely with the ambitions of the UK RARE DISEASE FRAMEWORK"
"...Not having to REPEATEDLY EXPLAIN to clinicians who I have appointments with ABOUT MY CONDITION"
"EQUAL ACCESS to SPECIALIST CARE for people IN ALL PARTS OF THE COUNTRY"
"To be treated like everyone else DESPITE my disease but to understand that I have some limitation."
"ACCESS to Care CO-ORDINATORS to NAVIGATE a COMPLEX HEALTH SYSTEM."
"Being BELIEVED my rare illness is NOT anxiety, alcohol or an eating disorder."
"LONGER APPOINTMENTS available to reflect the complexity of living with a rare condition"
"Doctors co-ordinating with each other as treatments don't work against other health problems"
"Having the Same SHARED INFORMATION"
...HAVING SPECIALISED CARE FOR MY SPECIFIC CONDITION. (There's only one place in the UK which is in England but there's no support in Wales)"
"POSTCODE MAPPING to check for areas where Patients are not referred into rare disease services."
"A much more effective way of MARKING my MEDICAL RECORDS THAT I HAVE A RARE CONDITION So they can ask how it affects my treatment and what special arrangements are necessary."
"To not have to WAIT such a LONG time for appointments"
"SPECIALISTS doing the TRAVELLING for clinics to what LOCAL PHYSICIANS are UPSKILLED and CONFIDENT in the immediate care of us."
"Equal, timely access to treatments for ALL patients REGARDLESS of postcode, specialty hospitals or insurance coverage. (+ not = NG)"
"VISIBILITY"
"INCLUSION"
"There should NEVER be a POSTCODE LOTTERY for hospital treatment in-home care or for equipment needed."
"An UNDERSTANDING by the Medical fraternity & wider general community of the CHALLENGES that confront people with a hidden disability."
Being ACCEPTED as being rare with no NAMED DISEASE
"People need to be EMPATHETIC and realise we may not look ill but that doesn't mean we don't feel unwell. Just because we LOOK NORMAL doesn't mean we aren't STRUGGLING We are NOT attention seeking We need to be LISTENED to and taken seriously I feel unwell 90% of the time, but looking at me you'd never know. We learn to put on a good act because Unfortunately its what we have to do to SURVIVE"
"...Being taken SERIOUSLY when Someone presents with SYMPTOMS."
"ADJUSTMENTS being made to ensure people get the most out of their APPOINTMENTS."
"ENSURING EVERY OPPORTUNITY IS ACCESSIBLE"
Use of the NHS ENGLAND HEALTH INEQUALITIES TOOLKIT to support."
"To not be FORGOTTEN about."
"HOLISTIC TREATMENT alongside MEDICAL to tackle SOCIAL CARE MENTAL HEALTH NUTRITION EDUCATION"
"ACTUAL TANGIBLE SUPPORT"
Charities etc. have funding for research but very few have on the ground workers supporting those with the actual disease."
"Primary healthcare system that ENABLES Specialist referrals when suitable, not hinders them."
BROADER TRAINING for non-specialists
"EQUITABLE ACCESS for patients with rare diseases to access high quality SPECIALIST SERVICES with ROI experienced in the rare condition"
"UNDERSTANDING of ALL ASPECTS of MITOCHONDRIAL DISEASE"
"...Having doctors who are NOT experts in rare diseases to TRUST that we sometimes KNOW MORE or can put them in contact with experts doctors."
"DIGNITY"

What are the drivers of inequity for people with rare conditions?

It is well established that a complex range of systemic drivers contribute to the disproportionately poor healthcare experiences faced by those living with rare conditions. This report is deliberately narrow in focus as we seek to understand and explain how the inherent scarcity of rare conditions creates a fundamental driver for inequity in the context of healthcare delivery.

Small population sizes are an immutable characteristic of rare conditions, but the challenge is compounded by the fact that there are a large number of individual rare conditions. While a single rare condition is defined as affecting fewer than 1 in 2,000 people, there are upwards of 7,000 distinct conditions, with new ones regularly identified through scientific progress.

While the rare community is collectively large, it is fragmented into thousands of small cohorts. Some conditions may affect a few thousand people in the UK, others just a few. This disparity means there are often inequities within the rare community itself. However, no matter where a condition falls on the rare scale, people with a rare condition will likely experience an inequity by virtue of having a rare condition defined by having a small patient population.

This scarcity creates three key challenges:

Because the prevalence of any single rare condition is by definition low, it struggles to meet the thresholds required to be categorised as a significant public health concern. In systems where resources and attention are allocated based on the breadth of impact, these conditions fail to demonstrate a burden that competes with more common conditions. Consequently, rare conditions are often sidelined in policy discussions and strategic planning, as the individual impact, however severe, is diluted by the low number of affected individuals.

The rarity of these conditions ensures they remain outside the realm of public and professional familiarity. Without a critical mass of cases, there is no natural mechanism for a condition to enter the general consciousness of healthcare professionals or become a standard part of medical training and discourse. This lack of exposure leads to a cycle of invisibility; because the condition is not well-known, it is less likely to be identified or discussed, ensuring it remains invisible to the clinical community and the healthcare systems they operate in.

The small number of individuals affected by a rare condition creates an inherent barrier to the generation of robust data. Traditional methods of gathering evidence, such as large-scale clinical trials or comprehensive epidemiological studies, rely on significant participant numbers to achieve robust levels of evidence. In the absence of a large population, it is impossible to produce the same volume or depth of evidence that is standard for common conditions.

Without deliberate adaptation or mitigation by healthcare systems, these three fundamental challenges directly drive the inequities experienced by individuals with rare conditions. These challenges, whether taken individually or in combination, lead to several critical failures in the standard of care experienced by people with rare conditions.

> **[Equity is] our whole community has the same standard of care and access to specialists and care across the country regardless of location, social economic background or ethnicity. Our community has equal access to treatments if we are lucky enough for treatments to become available within the UK.**
> - Person affected by fibrodysplasia ossificans progressiva

Explainer Box

We use specific icons throughout this report to identify which of the three fundamental challenges, low priority, low visibility, or low evidence, is driving a particular inequity.

Where an icon appears alongside an issue, such as delayed diagnosis or restricted access to treatment, it identifies which challenge, or challenges, are driving that inequity. This approach clarifies which of these challenges, whether acting individually or in combination, create the systemic inequities experienced by those living with rare conditions.

Where an icon appears alongside a case study of an initiative or recommendation, it highlights which of the challenges are being addressed.

Intersectional barriers to rare conditions healthcare

Inequity in the context of rare conditions diagnosis is a multi-layered issue. Fundamentally, the three core systemic challenges: low priority within overstretched healthcare systems, low visibility in clinical training, and low evidence, that drive inequity are a result of small patient populations. It is these factors that drive a baseline of systemic inequity that affects everyone in the rare conditions community.

However, a person's experience is not defined by the rarity of their condition alone. Instead, these systemic barriers layer upon broader, pre-existing drivers of inequity, such as ethnicity, gender, and socioeconomic status, that are inherent across all of healthcare and can affect anyone regardless of whether their condition is rare or common.

Acknowledging and investigating the intersectionality of these factors is vital, as a failure to do so risks leaving behind the members of the rare condition community most in need of support. While this report focuses specifically on the three key challenges arising from population scarcity, we recognise that this analysis represents only one facet of a much broader and more complex conversation about equity for rare conditions. Addressing these issues requires a collaborative effort, and we acknowledge the invaluable work of other experts, advocacy groups, and researchers who continue to highlight how wider societal disparities compound the difficulties of living with a rare condition.

Achalasia Action

Achalasia Action's report, 'Misunderstood, mistreated and fighting to be heard', has shone a spotlight on how inequities operate at multiple levels for people in the UK to get an achalasia diagnosis. Achalasia is a rare condition, and rarity itself creates a baseline inequity in people's diagnostic journeys. The research identified further layers of inequity within the achalasia community, shaped by gender and income. The research was a service-user-led study combining a national survey of 350 people with achalasia, qualitative interviews and focus groups.

Rare condition inequity: achalasia and delayed diagnosis

The report documents how people routinely experience years of misdiagnosis, repeated referrals, and fragmented care before receiving an accurate diagnosis. Over half of survey respondents endured multiple misdiagnoses, most commonly reflux, anxiety, and eating disorders, before reaching the correct diagnosis. Over a quarter (28%) waited longer than three years for a correct diagnosis. These delays resulted in avoidable decline in physical health, psychological distress, social isolation, and issues with employment.

Gender inequity within the achalasia community

Gender intensifies barriers to diagnosis. Women were disproportionately more likely than men to face diagnostic delays and repeated misdiagnoses. 31% of women waited over three years for a diagnosis compared to 18% of men, and women were more likely to have symptoms attributed to anxiety (41% vs 31%). In practice, this means that women experience an added layer of dismissal when getting an achalasia diagnosis, with physical symptoms frequently reframed as psychological, and the disease remaining undetected for longer.

Income inequity and access to diagnosis

Successful diagnosis frequently depended on sustained self-advocacy, meaning those without the time, confidence, health literacy, access to technology, or energy to persist were structurally disadvantaged. Income also shapes who can escape diagnostic delay. When NHS pathways failed, some people were able to pay for private consultations and diagnostic tests. Sixteen percent of respondents received their diagnosis privately, often describing this as a desperate last resort rather than a choice. Those without financial resources remained stuck in delayed NHS pathways, creating a two-tier system where speed of diagnosis depends on ability to pay.

Core20PLUS5

Core20PLUS5 is a national NHS England approach to inform action to reduce healthcare inequalities at both national and system level. The approach defines a target population – the Core20PLUS – and five focus clinical areas requiring accelerated improvement.

Core20: This refers to the most deprived 20% of the national population as identified by the national Index of Multiple Deprivation (IMD). The IMD accounts for a wide range of social determinants of health.

PLUS: PLUS population groups are identified at a local level. These are populations that experience poorer than average health access, experience and/or outcomes. They include:

- Ethnic minority communities.
- People with a learning disability and autistic people.
- Inclusion of health groups, such as people experiencing homelessness, drug and alcohol dependence, and Gypsy, Roma and Traveller communities.
- Coastal communities and other socially excluded groups.

5: There are five clinical areas of focus which require accelerated improvement: Maternity, severe mental illness, chronic respiratory disease, early cancer diagnosis and hypertension case-finding and lipid management.

From ‘Inclusion in What’ to ‘Equity in What’: (Re)Thinking the Question of In/Equity in Precision Medicine and Health: Sasha Henriques

Although equity, diversity and inclusion are now widely discussed in genomics, rare disease services and research still operate within systems shaped by social inequality. Offering the same service to everyone does not guarantee fair outcomes. People’s experiences of care are influenced by overlapping factors such as race, ethnicity, disability, language and socioeconomic circumstances.

In clinical practice, the challenges of living with a rare or inherited condition often sit alongside experiences of racism, language barriers, stigma or inflexible healthcare systems. Delayed referrals, poor communication, lack of interpreter support and the labelling of families as ‘difficult’ can deepen mistrust and limit options. For some people, a rare condition does not exist in isolation but adds to existing disadvantages, increasing the emotional, financial and practical work required to secure appropriate care.

An ethnographic study conducted at the Wellcome Sanger Institute showed that diversity is often discussed in terms of improving representation within genomic datasets. While representation matters, diversity is more than numbers. The ways in which race, ethnicity and ancestry are defined and recorded were not neutral technical choices. They reflected historical and social assumptions that influenced which data were collected, how groups were described, and who benefited from the research. Simply adding more diverse data does not automatically address deeper structural inequalities.

Achieving justice in genomics, therefore, requires more than inclusive language or statistical correction. It calls for careful reflection on how services are designed, how research categories are constructed and how power operates within institutions. Addressing the additional burdens faced by people living with rare and genetic conditions means embedding equity into everyday clinical practice and into the foundations of genomic research.

Sasha Henriques is the Director of Equity & Assurance at Genomics England. She is also a Registered Genetic Counsellor and incoming Chair of the Association of Genetic Nurses and Counsellors.

Full article: From ‘Inclusion in What’ to ‘Equity in What’: (Re)Thinking the Question of In/Equity in Precision Medicine and Health.

Genetic Alliance UK would like to thank Sasha Henriques for providing her insights and research to inform our ‘what does equity look like for rare conditions?’ workshops.

NHS Race and Health Observatory: The Seven Anti-Racism Principles

The Seven Anti-Racism Principles, developed by the NHS Race and Health Observatory, serve as an evidence-based framework designed to guide healthcare organisations in moving beyond performative pledges towards tangible, systemic change.

The work is structured around the following seven core principles.

- **Understand and acknowledge**: Recognising that structural, institutional, and interpersonal racism directly impact health outcomes. Organisations must be clear about where accountability lies for improvement.
- **Demonstrate leadership by naming racism**: Leaders are encouraged to engage continuously with how racism affects patients and the public, actively working to dismantle these barriers rather than using neutral language.
- **Meaningfully involve communities**: Ensuring that racially minoritised individuals and groups are involved at every stage of service development. This includes diversifying decision-making structures to ensure they are fundamentally inclusive.
- **Collect and publish data**: Committing to the comprehensive collection and publication of race inequity data. This information should directly inform policy and strategy; where data is missing, policies must change to ensure its collection.
- **Identify racial bias**: Actively seeking out and identifying racial bias within organisational policies and decision-making processes.
- **Apply a race-critical lens**: Integrating a race-critical perspective when adopting new interventions, testing improvements, or designing and delivering services.
- **Evaluate and reflect**: Using specific metrics that recognise racism as a determinant of health to assess the effectiveness of interventions. These evaluations should be made public to share best practices across the NHS.

The NHS Race and Health Observatory website provides associated briefings and an infographic to help healthcare systems operationalise these principles, aiming to shift the dial on long-standing racial inequalities in healthcare access and outcomes.

England Rare Diseases Action Plan 2025 Health inequity scoping review

Annex E of the England Rare Diseases Action Plan 2025, published by the Department of Health and Social Care, summarises a scoping review examining health inequities experienced by people living with rare conditions in relation to diagnosis and access to health and social care services.

The review was commissioned by the UK National Institute for Health and Care Research (NIHR) Policy Research Programme to inform the UK Rare Disease Action Plan. It was conducted by researchers at the University of Exeter Medical School, alongside colleagues from the EPPI Centre at University College London.

Seventeen distinct types of inequity were identified, including delayed diagnosis, limited clinician knowledge, inadequate information provision, poor care co-ordination, and restricted access to mental health and other services. Inequities were also found along lines of race/ethnicity, gender, socioeconomic status, geographic location, age and disability.

The review's findings are being used to address inequities through the NHS England Core20PLUS Framework. It reinforces the case for improved data collection, greater clinician awareness, and more equitable service commissioning for people with rare conditions in England.

How the rare community experiences inequity

The diagnostic odyssey

The most recent data on time-to-diagnosis for people with a rare condition shows that 1 in 4 in the UK waited at least 3 years between the first symptoms showing and receiving a confirmed diagnosis. The same data indicates that for the vast majority (95%) of the 3 year wait, people were within the healthcare system i.e. they were actively seeking help from the health service.

Inequities experienced and reported by respondents to our survey include:

Knowledge gaps and lack of clinical curiosity

Healthcare professionals often lack the training to recognise rare presentations. Many settle for treating individual symptoms rather than investigating the root cause. Those without a formal label often face scepticism. This lack of 'clinical empathy' leaves individuals feeling without validation and carrying the emotional burden of their condition alone.

Rare conditions often remain invisible in healthcare systems because medical training and digital triage tools focus primarily on high-prevalence conditions. Furthermore, these conditions are frequently excluded from core curricula and strategic planning which leaves clinicians struggling to navigate rare conditions.

> **[Equity is] better training of healthcare professionals about rare conditions and how they present.**
> - Person affected by hypergamma globulin anaemia sometimes known as XLA or Bruton's disease

> **[Equity is] to have healthcare professionals who will listen to me, rather than dismiss my bleeding disorder, because women don't have bleeding disorders. I don't expect them to know about my bleeding disorder but I do expect them to listen to me and phone my consultants for advice, there is a 24 hour on call haematologist.**
> - Person affected by platelet function disorder TPM4 variant

Systemic referral barriers

People with rare conditions are often passed between specialists who fail to connect multisystemic symptoms, with many facing delays or barriers to accessing referrals for appropriate tests, services, or experts. This systemic failure is primarily caused by low clinical familiarity with rare pathologies across the medical profession. Many rare conditions do not have recognised care or diagnostic pathways and so the path to a diagnosis and subsequent specialist care frequently depends on 'luck' regarding a clinician's personal expertise or an individual's perseverance and advocacy, rather than a standardised right to access equitable healthcare.

> **I was diagnosed with Addison's following referral to a PoTS clinic which was not easy to access. The biggest problem is recognition of symptoms and referral to the appropriate doctors by a GP. It took over 20 years to get a diagnosis of EDS after having pain and dislocating joints dismissed for years.**
> - Person affected by Addison's disease and Ehlers-Danlos syndrome

SWAN UK: Hana's story

'It's been such a rough and tough journey. Since he was born people have been trying to piece bits together. Trying to diagnose. Test after test. Doctors coming up with their own hypothesis and then organising tests again and again.

Once we had the VUS (Variants of Uncertain Significance) it took a further two years for specialist review. And only then were we aware that this was relevant and thoughts were that this strongly felt that this is 'the most likely' cause of all his complexities. Then we were introduced to SWAN.

We feel that people seem to shy away from rare conditions, shy away from genetics as they are 'too costly'. We were only offered genetic testing 4 years down the road.

When our son has to go into A&E or urgent care, the professionals have no clue what being undiagnosed means. Or don't seem to have any interest in his genetics. It would be so nice if education is passed on to healthcare professionals about this area of care. It seems so far into the shadows of the unknown. It's so exhausting having to tell accident and emergency staff what this means and what it means to our son at that moment.

Equity is being able to participate in activities without judgement or fear. Being listened to fully. It would be so nice to one day not have to keep explaining over and over what this means to have an 'Undiagnosed' condition and that it is just as important as any other 'condition' or diagnosed illness.

Just because he doesn't have a 'label' he still has 'something' and this is important to allow him to feel like an individual with his condition, and in himself.'

SWAN UK: Educational resources and community support

SWAN UK is the only dedicated support community in the UK for families affected by syndromes without a name - a genetic condition so rare it often remains undiagnosed. To combat the isolation and uncertainty these families face, SWAN UK has developed a suite of resources.

Central to this work are a new series of videos designed to bridge the gap between families and healthcare providers and animations offering easy to digest information about having an undiagnosed genetic condition and the journey to diagnosis. These resources include:

- Videos featuring a nurse, clinical geneticist and family to help healthcare professionals better understand and support the undiagnosed community. These will be launching on the SWAN UK website in April.
- Animations produced in collaboration with SWAN families, offering practical information and reassurance to those navigating a suspected genetic condition without a diagnosis.
- A new leaflet created in collaboration with Rareminds to support emotional wellbeing.

By raising awareness and providing high-quality digital information, SWAN UK ensures that families feel less alone while empowering professionals to offer more informed care.

SWAN UK
syndromes without a name

Heart UK: Alison's story*

HEART UK is the UK's only cholesterol charity, providing support, information and services for families and health professionals.

Familial hypercholesterolaemia (FH) is a genetic condition that affects the liver's ability to process cholesterol, leading to elevated LDL (or 'bad' cholesterol). The condition is passed through families and can result in a higher risk of heart disease at an early age if untreated. Heterozygous FH [HeFH] (where a faulty gene(s) is inherited from one parent) affects about 1 in 250 people. Sadly, most people with FH in the UK remain undetected.

When both parents have FH, and pass the faulty genes to a child, the child may be homozygous FH (HoFH). This is much rarer, affecting 1 in 250,000 people. The signs and symptoms of homozygous FH are the same as heterozygous, but they appear earlier, and the disease progresses much more aggressively.

HEART UK
THE CHOLESTEROL CHARITY

Alison is mother to two boys, born two and half years apart. They were both born with xanthomas visible on the sacrum. Xanthomas are yellowish fatty skin deposits, which commonly appear in HoFH children.

When Alison presented her first born aged two and a half years to a doctor, the child was undiagnosed, she was told she was neurotic and advised to pierce the xanthoma. She then took her young child on a journey to see multiple specialists, including a dermatologist, and an eye specialist. Eventually, a paediatric dermatologist asked about cholesterol in the family. Alison advised that her mother-in-law had HeFH and that her husband had high cholesterol. At last, aged four, the child was given a blood test, which revealed extremely elevated LDL cholesterol of 14-15 mmol/L. The same proved true for her other son. A genetic test for all family members confirmed both Alison and her husband were HeFH, and their sons were HoFH.

Alison's sons faced further challenges, including the bumpy transition from paediatric to adult services, as well as health problems. It has felt that different parts of the health system have not always communicated effectively, which can impact care.

Alison feels that the system could be improved through greater awareness of FH. Given FH is readily treated once diagnosed, it would be sensible to include screening for FH at birth. From those index cases, genetic testing could then take place through the family, providing assurance and treatment for those who need it.

*Name changed to maintain anonymity.

Breaking Down Barriers: Sian's Story

Sian is a GP and person affected by a rare condition

'A 23-year-old woman presented with early fullness, severe abdominal, flank and pelvic pain and nausea, both of which were exacerbated by oral intake. This progressively worsened to the point at which she was unable to tolerate any oral intake. All investigations were normal, in addition the patient had diagnoses of hypermobile Ehlers Danlos syndrome, postural orthostatic tachycardia syndrome and new daily persistent headache; she had had a constant headache for 8 years. Patient was trailed on nasojejunal feeding but this was not tolerated, and therefore she was commenced on intravenous nutrition, TPN.

The patient advocated for further investigations in the form of a CT angiogram to rule out rarer causes of her symptoms. CTA showed superior mesenteric artery syndrome, renal nutcracker syndrome and pelvic congestion syndrome. She then underwent a gastrojejunostomy and left renal vein transposition which resulted in marked symptomatic improvement. The patient was able to eat normally 4 days post-op, abdominal, flank, pelvic pain and nausea completely resolved as did the patient's constant headache.

Following surgery, the patient went back into education having previously not completed school, and then to medical school. She subsequently had a further three open abdominal surgeries during this time, two of which due to failed surgical treatment of nutcracker syndrome, however she is now a qualified doctor working in the NHS.

Diagnostic and treatment pathways for the most common type of EDS, hypermobile EDS currently do not exist, and for vascular compressions syndromes care is even more limited. Care for these conditions, if anything has gone backwards in recent years, with now only one UK centre predominantly seeing these patients, instead of two. Lack of knowledge, understanding and treatment pathways for these conditions, combined with medical professionals preconceived opinions on EDS, often lead to diagnostic delay, and poor care for patients with these conditions, due to this, equity for these conditions in this country is limited.

To truly improve care for this patient population the root cause of patient's poor experience with these conditions and associated conditions, such as EDS needs to be addressed. A deeper understanding of these conditions will likely only come about from an improvement in medical education, without this, the cycle of poor care, diagnostic delay and patient trauma, due to outdated opinions of these conditions, which are not rooted in current evidence, will likely continue. Equity for these conditions would mean that patients are able to access timely care for them, and regardless of where they live and their financial status, without risk of further psychological harm'.

Care coordination

People living with rare conditions often have complex health needs that require support across multiple health services, making effective coordinated care a high priority. However, most individuals do not receive this support and are instead forced to coordinate their own care while managing their condition.

The absence of coordinators

The low priority and low clinical familiarity surrounding rare conditions mean that, unlike more common conditions, rare conditions rarely qualify for funded care coordinators. The administrative burden is often pushed entirely onto the family. Families are left to manage their own care, chasing test results and synchronising appointments, without any professional oversight. This lack of support often leads to fragmented care.

> **There is a lot of talk of care coordinators but no one I have spoken to with my conditions appears to have been assigned one. They seem to be available for people with cancer or more recognised conditions. This is why I mentioned the idea of availability of regional nurses specialising in rare conditions. It is interesting to note (for example) that in Australia where there are less people known to be living with scleroderma all have access to nurses specialising in scleroderma.**
> - Person affected by systemic sclerosis sine scleroderma; bronchiolitis obliterans; Non CF bronchiectasis

Poor communication and incompatible systems

Different hospital departments and specialists often fail to communicate with each other or with the person's GP. IT and record-keeping systems across different NHS services often do not 'talk' to each other, leading to lost information and repetitive testing. This leads to people with rare conditions and their families having to repeat their symptoms and explain their condition over and over.

> **[Equity is] better communication between GPs and hospitals in relation to diagnosis and management of conditions unrelated to rare conditions.**
> - Person affected by mitochondrial myopathy

Transition

The transition from paediatric to adult care for young people with rare conditions can be challenging, primarily driven by low clinical familiarity and the low priority assigned to transitional planning. In paediatric settings, care is often holistic and family-centred, but as young people move into adult services, this structured support is often lost. Because adult specialists may lack experience with conditions that were historically considered childhood conditions, the low evidence base for adult management leads to a fragmented experience where young people 'fall through the gaps' losing access to coordinated multidisciplinary teams and having to become responsible for their own care.

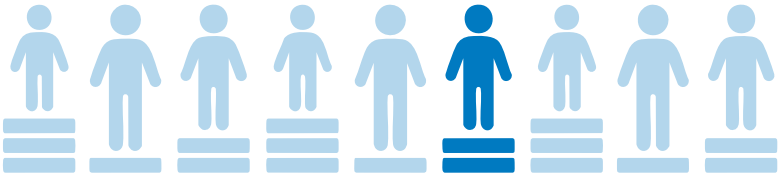
> **Moving from paediatric to adult care is poorly coordinated, leaving young people vulnerable at a critical stage. [Equity] is a smooth transition to adulthood, young people supported with clear pathways into adult healthcare, further education, and employment.**
> - Representative of congenital hyperinsulinism, chondrocutaneous branchial remnant (CCBR), Scheuermann's disease

Beyond healthcare

The impact of a rare diagnosis is rarely confined to a clinical setting, yet the broader infrastructure of education, social care, and the welfare system often fails to adapt. Due to low clinical familiarity and a lack of priority, these sectors frequently rely on rigid, 'one-size-fits-all' frameworks that do not account for rare conditions. This lack of integration means that individuals and their families are often forced to act as their own advocates, fighting to justify their needs to professionals who may have no prior knowledge of their rare condition.

The consequence of this systemic gap is a significant socio-economic burden on the person with the rare condition or their family member, parent or carer. Whether it is the struggle to secure tailored support in schools, the deprioritisation of social care funding due to low evidence of long-term outcomes, or the repeated denial of disability benefits by assessors, the result remains the same: a profound sense of isolation and financial strain. Without better recognition and a move away from standard templates, those with rare conditions remain effectively invisible to the services intended to support them.

> **"Equity looks like being given the help needed for me and my family to function in the same way as a family which does not have a rare condition, for example the provision of suitable housing, adaptations to my workplace so I can use my mobility aids without difficulty there, the provision of dropped curbs so I can get around my local area using mobility aids without having to go out of my way to find places I can cross the road and assistance with everyday tasks which I would normally struggle with."**
>
> - Person affected by Charcot-Marie-Tooth disease type 1A



Mast Cell Activation Syndrome - Jane's story*

People with mast cell conditions can have life-threatening symptoms triggered by anaesthetics and medicines commonly used during surgery and treatments for other conditions that should be urgently addressed. Careful planning can keep these people safe, but it can also slow down access to care that other people would get much faster from the NHS.

'It was exactly four months after a breast cancer diagnosis that I had a lumpectomy.

I saw a surgeon several times who promised an operation 'in four weeks' and then nothing. Every enquiry was met with 'we are waiting to hear from X and they would try and divert me to different places. I eventually tried a private hospital but they deemed me too high risk. They diverted me to an NHS hospital, but it was 150 miles away. Travel is really difficult for me because of the smells, vibration etc. It wasn't until I involved our local Patient Advice and Liaison Service (PALS) representative that a date quickly appeared.

Local hospitals MUST get to grips with MCAS. If I end up in an accident or emergency situation, they need to be able to manage my care.

Every part of my experience included delay and ignorance. I had an anaesthetist refusing to read the protocol written by his own Head Anaesthetist just three minutes before an operation, then riding roughshod over my subsequent reaction, using dressings I reacted to, being given endocrine treatment already known to be ill-advised. I then had horrible skin reactions after radiotherapy that took three months to heal and was told that 'this doesn't happen' when I requested help.

That being said, my post-operative nursing team was FANTASTIC. Taking time for actions like opening windows, removing packets of Clenil wipes from the ward and even a radiographer remembered to wipe down equipment with non-Clenil wipes for me.'

*Name changed to maintain anonymity.

Alstrom Syndrome UK: Sophie's story

Addressing equity in rare conditions means ensuring that people without genetic markers, defined NHS care pathways or pharmaceutical options are not excluded from research, care and compassion in healthcare. As genomic medicine and AI continue to shape healthcare, there is a risk that these existing inequalities will widen. The vascular compression community highlights this gap clearly.

Vascular compression syndromes are rare anatomical abnormalities in which blood vessels compress surrounding organs or structures, causing severe and often debilitating symptoms.

These conditions are not clearly genetic, currently have no definitive NHS diagnostic pathway and no approved pharmaceutical treatments. For most patients, the only potential intervention is complex, high-risk surgery, which may not even be accessible to them.

Symptoms frequently include severe digestive pain, difficulty eating, weight loss, fatigue, and systemic illness. These symptoms often mimic more common gastrointestinal and functional disorders, leading to repeated misdiagnoses, delayed recognition, and dismissal of a person's experience. As a result, people may cycle through healthcare services for years without answers, support, or validation, becoming increasingly unwell.

This example exposes a core equity challenge: how healthcare systems respond to ambiguity. People with rare conditions are clearly ill, yet the absence of adequate research, biomarkers and evidence-based treatments leaves them vulnerable to falling into a clinical and systemic 'black hole'.

To address this, equity in rare conditions must go beyond genetics and drug development. We need to redefine how we view treatment to include validation, symptom management, care coordination and sign posting to supportive services. We also need systems in place so that these people do not slip through the healthcare system due to uncertainty or lack of evidence.

Most critically, this requires a culture change where a person's suffering and pain is validated even when we don't have the answers. Without this shift, innovation risks benefiting only those whose conditions fit existing models of certainty, leaving others behind.

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 condition?

In 2023, Metabolic Support UK's Thoughts into Action project explored this question from the rare condition community's perspective. The research revealed that people with rare conditions 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 conditions not only survive but thrive.

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.

Access to specialist care, treatment and medicines

For people affected by rare conditions, accessing the right medications, expert care, and support services is essential for managing their condition and avoiding unnecessary treatments and referrals.

Difficulty accessing specialist care

Because rare conditions suffer from low clinical familiarity, expertise is rarely found in local hospitals. Consequently, care is concentrated in specialist centres or regional hubs, often located far from the person with the rare condition. This can create a physical and financial burden, as they must often travel across the country to see the only team in the UK with sufficient clinical evidence to treat them.

> **[Equity is] being able to get specialist care / treatment without having to fight for it and doing all the research myself.**
> - Person affected by neurofibromatosis type 1

Value of expert care

Despite the cost and distance involved in traveling to receive expert care, people with rare conditions will often accept this inconvenience if it means gaining access to expert care or receiving an appropriate care plan.

Workforce shortages

A chronic lack of specialised consultants and the resulting waiting times to access them was raised by survey respondents. For a person affected by a rare condition this delay often means a progression of symptoms without any clinical intervention, as there simply are not enough experts familiar with their specific needs to manage the caseload.

> **[Equity is] to have more consultants in the field. I know there is a huge shortage in neurologists. So training doctors need to know more about these conditions. It took forever to see a consultant, a physio, a urologist and a bowel specialist. We ended up going private to see the same specialists to reduce the wait from years to only 6 months.**
> - Person affected by hereditary spastic paraplegia

Postcode lottery access to medicines

Access to medicines for rare conditions is frequently dictated by regional funding rather than clinical need, a direct consequence of low priority within local NHS trusts. This can create deep inequality, where a person's location determines whether they receive a life-altering therapy or are denied it based on local reimbursement barriers that do not exist elsewhere. Some respondents noted differences between devolved nations.

Supply chain issues

Some respondents highlighted supply chain issues. For example, people affected by Addison's disease often face shortages of essential medications, such as hydrocortisone. These supply issues are frequently driven by low priority within the broader pharmaceutical market; because the patient population is small, manufacturing and distribution are not always robustly protected, leaving people in a state of constant anxiety about whether their medicine is available.

> **[Equity] means better treatment options beyond off-licence medications, which can be ineffective, carry severe side effects, and present difficulties in supply and access. For children and young adults, equity means having the confidence, knowledge, and resources to explain their condition, stand up for their rights, ask questions, and take an active role in shaping projects and resources that affect them.**
> - Representative of congenital hyperinsulinism, chondrocutaneous branchial remnant (CCBR), Scheuermann's disease

Access to non-medicine treatments

Respondents noted that access to interventions such as physiotherapy, speech therapy, and medical diets are often chronically underfunded for rare conditions. This is typically due to low evidence regarding long-term outcomes for small cohorts, leading commissioners to view these essential services as a lower priority for people with rare conditions than those with more common rehabilitation pathways.

> **[Equity is] acceptance that our specific dietary needs are a medical necessity, not a personal choice.**
> - Person affected by hereditary fructose intolerance (HFI)

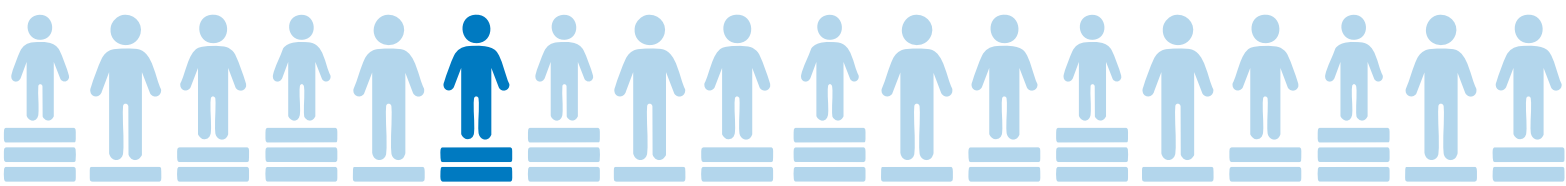
The National Society for Phenylketonuria (NSPKU): Chelsea's story

Chelsea Hodder lives in Merseyside with her husband and their seven-year-old daughter, Willow. Willow looks like any other bright, energetic child but behind the scenes, her life is defined by a rare genetic metabolic condition called phenylketonuria (PKU).

PKU means Willow cannot metabolise protein. 'What sounds manageable is, in reality, frightening. Willow can eat no more than three grams of protein a day without risking permanent damage to her brain. To put that into context, three grams is less protein than you'd find in a single slice of bread. Willow relies on prescribed medical foods and supplements and any normal food must be carefully weighed, measured, and planned. There are no quick snacks, no spontaneous treats, no carefree moments around food. For me and my husband, the constant monitoring is exhausting. We live with the knowledge that vigilance is not optional.'

Chelsea says it concerns her to see that Willow already feels the impact of being different. She notices when she can't eat what her friends eat, when she has to sit out of shared moments that most children take for granted. 'I am worried that as she gets older she will find it harder to maintain this rigid diet.'

Chelsea is backing the NSPKU's Hungry for Change campaign. 'Willow deserves more than a life of restriction and worry. We rely on specialist metabolic services to help Willow cope with her dietary treatment, but these services are under strain. We need prescribed foods to be accessible. There are also new pharmaceutical treatments coming for people with PKU, but the UK has a bad track record of making these available for people with PKU. Willow deserves a happy, hopeful future; and on Rare Disease Day, we are calling for change so that children like Willow can truly thrive.'



DEBRA UK: Abi's story

'I attended an epidermolysis bullosa (EB) mobile clinic local to me in Middlesbrough (NE England) as a representative of DEBRA UK and met people who had travelled several hours down from Scotland. Having to travel long distances for specialist care makes it harder to access, and even harder for those on lower incomes. Even the inequity is inequitable.

People living further from specialist centres may need to pay more to travel and more again to stay overnight when the distance there and back takes too long to travel in one day. Long journeys can be particularly physically difficult for people living with EB due to their more fragile skin. Where there are young children who must accompany their parent, the stress and cost of long distance travel is exacerbated. Single parent families and those with younger children may be particularly affected.

I listened to people living with EB describe how their primary care providers had asked questions demonstrating a lack of understanding of their condition. For example, the mistaken assumptions that their EB symptoms might be an allergic reaction. Additionally there is the risk that unrelated symptoms might be blamed on their rare condition and not properly investigated as a separate issue. Young children with EB will often have wounds that a healthcare professional might mistakenly see as the result of abuse. Increased general awareness of the condition is important to avoid generating anxiety in parents who might otherwise feel blamed for their child's wounds.'

Dr Abi Witherden is the Research Grants Manager at DEBRA UK.

Breaking Down Barriers: Mehreen's story

'As a mother supporting my child through the process of accessing care for median arcuate ligament syndrome (MALS), the overall experience has been prolonged, complex, and emotionally demanding. From the outset, care was slow to initiate, largely due to the highly specialised and rare nature of the condition. My child underwent multiple scans and investigations and was referred between numerous services before a clear diagnosis was considered. We were assessed twice by the CAHMS team ruling out any eating disorders such as Anorexia or Bulimia due to her significant weight loss. This resulted in significant delays and uncertainty, and the early stages of care felt fragmented and difficult to navigate.

On several occasions, we had to actively intervene to ensure referrals were followed through and concerns were addressed. Eventually, my child was referred to a specialised hospital in London, where we remained under their care pathway for almost two years. During this time, we believed progress was being made and that appropriate specialist treatment was imminent. However, we were later informed that this pathway would not continue, and my child was subsequently referred to another specialist centre in Birmingham. We are currently under their care and awaiting the next stages of treatment, with the hope that progress will now be more timely.

Overall, the journey to diagnosis and specialist intervention was lengthy, and the lack of early recognition significantly impacted my child's wellbeing and our family's emotional resilience. This experience highlights key issues surrounding equity and diversity in healthcare. Access to specialist services for rare conditions such as MALS often depends on geographical location, clinician awareness, and a family's capacity to advocate effectively. Families with fewer resources or lower health literacy may face greater barriers, leading to inequitable delays in diagnosis and care.

Achieving equity in healthcare requires consistent referral pathways, improved awareness of rare conditions, and fair access to specialist services for all people with rare conditions, regardless of background or circumstance.'

Access to research

Patients struggle to find available research opportunities

Respondents highlight that information on available trials is difficult to find. This is worsened by a lack of clinical support as healthcare professionals are often unaware of active trials. Often people with rare conditions have to self-navigate complex research without a clear referral pathway.

> **Equity for rare conditions is investment in research and long-term support, so that children and adults with rare conditions - many of whom have complex, lifelong needs - are not left behind or forgotten.**
> - Parent of a child with non ketotic hyperglycinemia

Geographical barriers to trials

Clinical trials are often held at specific international or national centres, requiring travel that many cannot afford or physically manage.

> **So far there have been no clinical trials for MdDS in the UK so if people want to donate their data by taking part in clinical trials they have to travel to Antwerp to do so and many of us can't afford to do that.**
> - Person affected by familial mal de debarquement syndrome

NIHR Rare Diseases Research Landscape Report (2023)

The NIHR Rare Diseases Research Landscape report provides a comprehensive mapping of the UK's research ecosystem between 2016 and 2021, revealing a total investment exceeding £1.1 billion. This funding is almost equally split between the public sector, with the NIHR and MRC contributing £627 million (representing 7% of their total joint portfolio), and the charitable sector, where 107 AMRC member charities provided £580 million across 2,600 studies. While the investment is substantial, it is often siloed by condition; for example, motor neurone disease received the largest share at 8%, followed by Huntington's disease at 5%.

Despite these figures, the report exposes a geographical 'postcode lottery'. Research activity is heavily concentrated in London-based institutions (overseeing 35.8% of all rare condition research awards). This centralisation leaves other regions behind, with Wales accounting for only 1.6% and the East Midlands just 1.4% of the national portfolio. This imbalance directly dictates patient access to life-changing opportunities. People living near centres of excellence in the South East have significantly higher recruitment rates for clinical trials. With almost half of studies being commercially funded, the report underscores the urgent need to decentralise infrastructure to ensure that the 3.5 million people in the UK living with a rare condition have equitable access to innovation, regardless of their location.

Case study: AMRC and ABPI Report: Achieving inclusivity in clinical research

A joint report by the Association of Medical Research Charities (AMRC) and Association of British Pharmaceutical Industry (ABPI), 'Achieving inclusivity in clinical research', addresses the critical gap between UK trial participation and the actual diversity of the patient population. It highlights that while interest in research is high, engagement is significantly lower among ethnic minority groups due to practical barriers like travel costs and a lack of community trust. To fix this, the report proposes a UK-wide strategy, the sharing of recruitment best practices, and a standardised system for reporting participant diversity by the end of 2025.

For rare conditions, the report stresses that inclusivity is a scientific necessity. Because patient populations are naturally small, every participant counts; excluding any group risks making data inaccurate or biased. It identifies that people with rare conditions from underserved backgrounds face compounded difficulties in accessing the few trials available. The report advocates for decentralised trial models, such as local clinics or home-based participation, to reach people who are geographically dispersed and highlights medical research charities as vital partners in building the trust needed to make these trials inclusive.

Breaking Down Barriers: Sharlene's story

Sharlene is an autistic young woman with multiple rare and complex physical health conditions who has experienced significant and ongoing barriers to equitable healthcare. Her clinical picture is complicated by hypermobile Ehlers-Danlos syndrome (hEDS) and multiple abdominal vascular compression syndromes, conditions that are poorly understood, rarely encountered, and lack clear diagnostic or treatment pathways within standard NHS care.

The rarity of these conditions has resulted in repeated dismissal of symptoms, prolonged diagnostic delays, and fragmented care across specialties. Clinicians have frequently expressed uncertainty or unfamiliarity, and in the absence of a clear pathway, concerns have been minimised rather than escalated. This uncertainty has too often defaulted to inaction.

Her autism significantly affects communication, sensory tolerance, and stress regulation. In clinical settings, she may struggle to articulate symptoms clearly, particularly when overwhelmed, fatigued, or in pain. These communication differences have led to her physical symptoms being misinterpreted or deprioritised, especially when clinical presentations do not conform to expected norms.

Her documented mental health history has further compounded this inequity. Rather than being recognised as a consequence of chronic illness, repeated medical trauma, and diagnostic uncertainty, it has frequently been used to frame her presentation as psychological in origin. This has resulted in physical symptoms being attributed to anxiety or distress, even in the presence of objective findings.

The intersection of autism, her rare condition, and mental health history has created a particularly high-risk scenario. She falls between services, with no single team taking overall responsibility for her care. Parental advocacy has been essential but has at times been perceived as problematic rather than protective.

The cumulative impact has been severe: delayed treatment, physical deterioration, repeated crises, and loss of trust in healthcare services. This case highlights the urgent need for autism-informed, trauma-aware, and rare condition-competent care, alongside clearer pathways and accountability. Equitable healthcare must recognise complexity and rarity as indicators for enhanced support, not dismissal.

Delivering a fairer healthcare system for people with rare conditions

To better understand what health equity looks like for people with rare conditions, Genetic Alliance UK consulted with the rare conditions community at the end of 2025. Through a programme of workshops and a survey which received 243 responses, we explored what equity looks like for people living with genetic, rare and undiagnosed conditions.

> **Equity for rare conditions means that no one is disadvantaged simply because their condition is unfamiliar, complex, or affects only a handful of people worldwide… Ultimately, equity looks like dignity, visibility, and meaningful inclusion, a system where being rare does not mean being forgotten.**
> - Person affected by neuroacanthocytosis (NA) syndromes: VPS13A disease (also known as chorea-acanthocytosis), XK disease (also known as McLeod syndrome)

The feedback gathered suggests that equity is not achieved until a person affected by a rare condition can navigate the healthcare system with the same ease and efficacy as a person with a common chronic condition. This is not to say that people with common chronic conditions have a smooth journey, but rather that the infrastructure for their care is more established, whereas those with rare conditions must often navigate a system not designed for their specific needs.

> **Recognition that one size healthcare for all does not reflect the specific needs of people with rare conditions.**
> - Person affected by primary immunodeficiency

What does an equitable experience look like?

Based on our community survey, an equitable system would be defined by:

- **Timely and accurate diagnosis**: Routine access to genomic testing and specialist pathways to end the diagnostic odyssey.
- **Informed clinical encounters**: A workforce trained to consider rare conditions as a possibility rather than dismissing atypical symptoms.
- **Coordinated care**: Dedicated care coordinators and unified digital records to ensure the system wraps around the person affected by a rare condition, rather than working against them.
- **Access to services, treatment, and medicines**: Fair and consistent availability of specialised therapies and healthcare support, ensuring that geographical location or cost do not act as barriers to essential care.
- **Research opportunities**: Inclusion in clinical trials and registries as a standard part of clinical care, ensuring every person has the chance to contribute to, and benefit from, medical breakthroughs.

Interestingly, the visible inequities described by respondents to our community survey, such as the diagnostic odyssey and poorly coordinated care, align closely with the four priorities identified by the UK Rare Diseases Framework.

It is important to note that The UK Rare Diseases Framework has been a fundamental driver of initiatives to address these inequities, notably:

- **National registries**: England, Wales, and Scotland have introduced rare conditions registries to improve data for research and treatments.
- **Healthcare professional resources**: The development of 'just-in-time' tools for healthcare professionals like the GeNotes platform, which provides clinicians with concise, actionable genomic information at the point of care, ensuring rare conditions are considered rather than dismissed.
- **Information hubs**: Scotland and Northern Ireland launched dedicated hubs to support healthcare professionals and people with rare conditons.
- **SWAN clinics**: Wales introduced nurse-coordinated care clinics specifically for undiagnosed genetic conditions.
- **Rare Disease Research UK**: Established 11 research nodes to foster collaboration between researchers, people with rare conditions, and charities.
- **Rare Diseases Research Landscape Project**: A comprehensive overview of rare condition research funded across the UK between 2016 and 2021 to support future policy and investment decisions.
- **Quality standard for rare conditions**: An Independent Advisory Group was formed to develop a Rare Disease Quality Standard for high-quality, cost-effective care.
- **Health inequity scoping review**: A project that published evidence on health inequities experienced by the rare community with regards to receipt of a diagnosis and access to health and social care services.

By driving forward initiatives such as quality standards, registries, and integrated research platforms, the UK Rare Diseases Framework has been instrumental in starting to address the core challenges of low priority, low evidence, and low clinical familiarity that lead to inequity for rare conditions.

To maintain this momentum, the governments of the UK must utilise this one-year extension to co-produce a successor to the UK Rare Diseases Framework that is permanent, sustainable, and fully resourced.

NICE

The Rare Disease Quality Standard: A tool for equity

People people affected by rare conditions often experience significant inequities in care, including delayed diagnosis and limited coordination across pathways. While the UK Rare Diseases Framework set an important strategic direction, there were no measurable, nationally recognised standards against which progress could be tracked or services held to account.

The NICE Rare Disease Quality Standard (QS) has been developed to address this gap.

Led by a UK-wide independent steering group, the initiative brought together people with lived experience, support organisations, clinicians, researchers, and system partners including NICE and NHS England. The initiative was led by the Rare Autoimmune Rheumatic Disease Alliance (RAIRDA) with Sue Farrington, the Alliance's Co-Chair, acting as project sponsor and Principle Consulting, the Secretariat for the Alliance, providing project support. The aim was to define a clear, shared minimum standard of care for what people with rare conditions should expect from the health system.

The Quality Standard consists of a set of concise, evidence-informed statements covering key domains that matter most to people with rare conditions, including timely diagnosis, coordinated care, mental health support, and access to information and specialist expertise. The statements were developed through a rigorous process with engagement from across the rare community. It included a scoping review, surveys, and a formal consensus workshop, before the draft statements were taken to a NICE committee for approval. The project marks the first time a NICE quality standard has been developed this way.

As a tool for equity, the Quality Standard serves several important functions. For people with rare conditions and families, it provides clarity and empowerment - setting out the minimum standard of care they should expect and supporting informed conversations with healthcare professionals.

For clinicians and service leaders, it offers a consistent reference point to guide service design and quality improvement. At a system level, it introduces transparency and accountability, enabling progress to be monitored and variation to be identified and addressed.

The Quality Standards development demonstrates how person-led, collaborative approaches can translate lived experience into practical tools that support more equitable, consistent care for people living with rare conditions across the UK.

Equity is...

- A timely, accurate diagnosis.
- Healthcare professionals being trained to identify rare conditions
- Better screening programmes to identify rare conditions earlier
- Access to the medicine you need through a simple, timely process regardless of where you live in the country
- A personalised care plan to help join-up services and reduce the burden on families
- Seamless communication between specialists, hospitals, and GPs
- Access to clinical trials and removal of financial and geographic barriers to participation
- Supporting people with rare conditions and their families to attend appointments that are far from home
- Access to psychological support, social care, and education
- Having a care coordinator
- Healthcare systems providing specialist diets and non-medicine treatment where necessary
- Receiving treatments in the most accessible and manageable form possible
- Prioritising research into rare conditions
- Access to the appropriate level of specialist care regardless of where you live

The findings from our survey and series of workshops were designed to specifically capture the lived realities of those affected by rare conditions. By exploring their experiences and listening to views of people with rare conditions, carers, and clinicians, we sought to distill the core elements that define an equitable healthcare system for rare conditions. The insights gathered from these diverse perspectives have been developed into a series of equity statements that serve as a reflection of our findings.

These equity statements highlight what is needed for people affected by rare conditions to feel they are receiving equitable care. It is important to recognise that there are a significant number of existing initiatives and tools, such personalised care plans and 'just-in-time' resources for healthcare professionals, that have begun to make the experience of care more equitable for some. There has been excellent work driving these initiatives forward under the UK Rare Diseases Framework, and it is essential that this momentum continues.

However, to create a healthcare system that is equitable for all, we must also take bolder, more decisive steps to address what is driving these visible inequities experienced by the rare conditions community.

Our five calls for action

To address the systemic inequities caused by each rare condition having inherently small populations, specifically the challenges of low prevalence, low clinical familiarity and low evidence, Genetic Alliance UK are calling on the governments of the UK to:

1. **Deliver a UK-wide map of rare conditions**: The four UK nations must urgently fund comprehensive rare condition registries and work in partnership to map all 7,000 rare conditions. Robust national data infrastructure is not optional, it is the foundation of equitable service planning, resource allocation, and healthcare delivery.

2. **Close the evidence gap through fair research investment**: Government and research funders must correct the imbalance in research investment by directing funding towards rare conditions. Decision-makers must also reform evidence standards to recognise that uncertainty is an inherent feature of rare conditions and should not be used as a barrier to access or innovation.

3. **Mandate system-wide accountability for rare conditions**: Healthcare systems must stress-test policies, commissioning decisions, and service delivery frameworks against the realities of low-prevalence conditions. This must include systematic auditing to identify gaps, eliminate inequities, and enforce consistent standards of clinical accountability across all services.

4. **Embed rare conditions into mainstream healthcare delivery**: Current reforms across the UK healthcare system present an opportunity to fully integrate rare condition care into routine service provision. Failure to act now will entrench existing health inequalities for the 1 in 17 people affected by a rare condition in the UK.

5. **Commit to a bold successor to the UK Rare Diseases Framework**: UK governments must commit to a long-term successor to the UK Rare Diseases Framework that delivers measurable improvements for people with rare conditions. This successor must set clear targets, and the nations must respond with adequate funding, directly addressing the structural drivers of inequity identified in this report.

1. Deliver a UK-wide map of rare conditions

While 3.5 million people in the UK live with a rare condition, many remain functionally invisible within national health data, planning, and commissioning frameworks. The UK currently lacks a comprehensive, unified map of prevalence, geographic distribution, and healthcare utilisation for rare conditions.

Healthcare systems cannot accurately prioritise what they cannot measure. This data deficit ensures that rare conditions are routinely sidelined in service design, workforce planning, and resource allocation. The consequences for people affected by rare conditions are profound, contributing to diagnostic odysseys, fragmented care, and barriers to accessing specialist care, medicines and research.

Genetic Alliance UK: Stats Behind the Stories (2024)

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 8 in 10 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.

Genetic Alliance UK Stats Behind the Stories (2024) report highlights the shocking lack of data held on rare conditions. By auditing 163 of the most prevalent rare conditions we found a lack of formal support structures:

- Less than 1 in 3 (44) of the studied conditions are supported by NICE guidance. Even then, most are subsumed into broad categories, erasing the specific clinical nuances required for effective treatment.
- Of the 79 conditions with a defined commissioner, only 9 benefit from a nationally commissioned specialised service. 44 conditions have no specialised service at all, forcing people with rare conditions to rely on generalist care.

Genetic testing is available for less than half (49%) of these conditions.

A national prevalence audit and mapping exercise would represent a foundational shift. By systematically assessing the prevalence and distribution of all rare conditions across the UK, the government and the NHS would gain the evidence base needed to identify unmet need, regional disparities, and pressure points across the care system. Crucially, this would enable rare conditions to be meaningfully integrated into population health planning, rather than treated as marginal or exceptional cases.

This would be a first step toward equity. Only by making rare conditions visible in data can the NHS design services that are proportionate, targeted, and fair, ensuring that people with rare conditions are no longer left behind simply because they are few in number.

Rare condition registries

Rare condition registries are centralised, secure databases that aggregate longitudinal medical, genetic, and demographic data, providing a framework for consolidating otherwise fragmented information. In the UK, these vital functions are managed by NCARDRS (England), CARDRISS (Scotland), and CARIS (Wales), with development currently underway to formalise similar structures in Northern Ireland (NIRADCAR).

By precisely quantifying the prevalence and geographical distribution of rare conditions, these registries enable the NHS to transition from reactive care to proactive, data-driven service planning. They allow for the strategic allocation of funding and the efficient design of specialist services. They facilitate the study of a condition's natural history, allowing clinicians to observe the condition's progression over decades. This collective intelligence identifies clinical patterns that would be invisible in isolation, directly supporting the clinical understanding that can reduce the diagnostic odyssey for people affected by rare conditions.

The value of rare condition registries include:

- **Clinical best practice**: Because each rare condition affects a small patient population, a healthcare professional might only see a small number of cases in their entire career. Registries pool data from every case nationwide, creating a large enough dataset to understand how to manage these conditions. Without a registry, treatment often depends on the specific knowledge of a local consultant. Registries allow experts to draft clinical guidelines to support standardised care, reducing inequities in the care people with rare conditions receive.
- **Addressing postcode lotteries**: By improving understanding of rare conditions and enabling the development of national standards, registries help address geographical inequities.
- **Understanding natural history**: To treat a rare condition we first need to understand its natural history. Registries track how a condition evolves over time, allowing clinicians to intervene at the exact moment it will be most effective to do so.
- **Supporting the life sciences sector**: Registries are indispensable for the life sciences sector. They identify eligible cohorts for clinical trials that would otherwise be non-viable due to participant scarcity.
- **Visibility and priority**: They grant individuals with rare conditions formal visibility within the healthcare system, ensuring they are seen by policymakers and providers alike and accounted for when making service planning decisions.

Despite their transformative potential to address the main challenges driving inequity for rare conditions, UK rare conditions registries have historically been hampered by a lack of funding and uncertainty caused by short-term funding cycles. To realise the potential of registries and use them effectively to address the inequities faced by rare conditions, governments must:

1. Commit to a sustainable funding model that enables ring-fenced, multi-year investment to ensure the long-term security and evolution of these data assets.
2. Ensure transnational data interoperability across the UK. With over 7,000 known rare conditions, no single nation holds a statistically significant population for most rare conditions. Data must be interoperable across the four nations and international partners, exemplified by the EUROCAT model, to empower global research and clinical collaboration.
3. Facilitate genomic integration: Given that 8 in 10 of rare conditions are genetic in origin, registries must be integrated with genomic medicine services. A 'unified genomic record' would allow for real-time data synchronisation, enabling rapid re-analysis as new variants are discovered.

2. Close the evidence gap through fair research investment

The inequitable distribution of research funding directly impacts people with rare conditions and contributes to their experiences of delayed diagnosis and fragmented care. It causes a significant loss of scientific innovation. Rare conditions often serve as blueprints for human biology and defunding them stalls breakthroughs that could benefit more common conditions.

According to the National Institute for Health and Care Research (NIHR) Rare Diseases Research Landscape Report (2023) the distribution of research activity across the thousands of known rare conditions is uneven. While there are over 7,000 rare conditions, the report reveals that research focus is heavily concentrated on a small minority. Just 23 rare conditions, representing less than 1% of all known rare conditions, account for the majority of research activity identified within the UK's funding portfolios. This imbalance means that while these conditions benefit from significant academic and industrial attention, the vast majority of rare conditions remain under-researched, contributing to the fact that only 1 in 20 rare conditions have effective treatment.

When comparing rare conditions to non-rare (common) conditions, the 2023 NIHR Research Landscape report highlights a significant funding gap. Rare conditions receive a disproportionately smaller share of available research funding compared to high-prevalence conditions.

The primary disparity lies in how research funding is distributed across the UK's total health research portfolio. The NIHR and MRC (Medical Research Council) spent approximately 7% of their total research budget on rare conditions between 2016 and 2021. In contrast, non-rare conditions, such as cancer, cardiovascular disease, and respiratory illnesses, account for the remaining 93% of the portfolio.

The current funding disparity is a systemic failure that traps thousands of rare conditions in a state of low priority and low evidence. While these conditions represent a vast range of human biology, they receive only 7% of the UK's health research budget. This lack of investment prevents the gathering of data, creating a self-fulfilling cycle where a lack of evidence results in continued underfunding.

Rectifying this imbalance is a strategic necessity for the entire healthcare system. Rare conditions serve as essential biological blueprints; the insights gained from studying them frequently lead to breakthroughs in common conditions like cancer and heart disease. By failing to address these inequities, we forfeit vital innovation and stall the progress of precision medicine. Investing in these under-researched areas is not merely a matter of fairness, but a prerequisite for medical advancements that benefit the whole of society.

> **"There is a critical need to include more ethnic and racial minorities people and their families in rare condition clinical trials. There is also a need to train clinicians in rare conditions, and to create more user-friendly and culturally-competent educational resources."**
> - Researcher of rare kidney conditions

> **"[Equity is] access to clinical trials/ research, I don't know of any clinical trials for this condition with very poor prognosis. Awareness, research, clinical trials, gene therapies potentially but most importantly that structure in England once clinical trials have been proven to appraise new therapies is not fit for purpose for expensive drugs for rare conditions."**
> - Parent of a child with DNM1L variant

LifeArc and Genetic Alliance UK: Accelerating R&D for rare disease in the UK: An opportunity to change millions of lives (2025)

'Accelerating R&D for rare disease in the UK: An opportunity to change millions of lives', addresses the systemic barriers preventing 3.5 million people in the UK from accessing life-changing treatments. The report provides a strategic roadmap to overhaul the UK's rare disease research and development landscape, ensuring that scientific breakthroughs are efficiently translated into clinical action.

To achieve this, the report details 10 specific recommendations:

Diagnosis and data

1. Make sure rare diseases are included in plans for the Health Data Research Service, which will aim to transform how NHS data is used.
2. Set clear standards for rare disease data collection and storage – and adequately support existing registries to maintain, streamline and unify their data.
3. Make whole genome sequencing easier to access and improve medical training, so that more doctors can spot rare diseases and understand the value of a diagnosis.

Market access: making drugs available

4. Health departments should invest in generating evidence that shows the real costs of rare diseases to all areas of society.
5. Regulatory and assessment bodies, responsible for approving treatments, must align their evidence requirements and tailor them to rare diseases

Support for innovators developing drugs

6. Create a support service that guides researchers through the complex drug development and approval process.
7. The next UK Rare Disease Framework must come with funding to map the full journey of rare disease research, so that researchers can plan efficiently and get treatments to people, faster.

Crucial drivers of change

8. A UK-wide champion and coordinator for rare diseases is appointed to lead and coordinate efforts across the UK.
9. Ministers from all four UK nations commit to a refresh of the 2021 UK Rare Diseases Framework, reflecting today's challenges and opportunities.
10. There is a review of the methods that Government could use to encourage more research into rare diseases in the UK

The report serves as an urgent call to action for the UK government and health leaders to prioritise rare diseases within national policy. By adopting these recommendations, the UK can secure its position as a global leader in life sciences while providing hope and tangible health outcomes for millions of people.

3. Mandate system-wide accountability for rare conditions

When decision-makers reach an evidence gap, they have a number of options. They could postpone their decision as they wait for evidence to be generated, and potentially commission work to fill the evidence gap. Or they could lower their evidence requirement threshold and manage risks through other means. Where it is impossible to fill evidence gaps - for example because populations of people affected by a rare condition are so small that it is not realistic to expect samples to be large enough for certain statistical tools - the choice is either - not making a decision or making a negative decision - or lowering or adapting evidence thresholds.

Some UK decision-making processes only accommodate the first option, applying frameworks designed for common conditions to rare conditions where the required evidence cannot exist.

Safety and access decisions

Traditional marketing authorisation depends on statistical significance requiring sample sizes that rare conditions often cannot meet. In the UK, the Medicines and Healthcare Products Regulatory Agency (MHRA) has two existing approaches can manage this challenge:

- Marketing authorisation under exceptional circumstances: This applies to medications where complete data collection is impractical due to the rarity of the condition or because gathering full information would be impossible or ethically problematic.
- Conditional Marketing Authorisation (CMA): When a medicine addresses an unmet need, the MHRA can issue a CMA in advance of the full clinical data becoming available, where there is reasonable expectation that this information will soon be available.

However it is well understood that gaps still exist. To address this MHRA have a programme of work to make decisions specifically for treatments for rare conditions where existing pathways are not appropriate, this could lead to a new pathway to an 'Investigational Marketing Authorisation'.

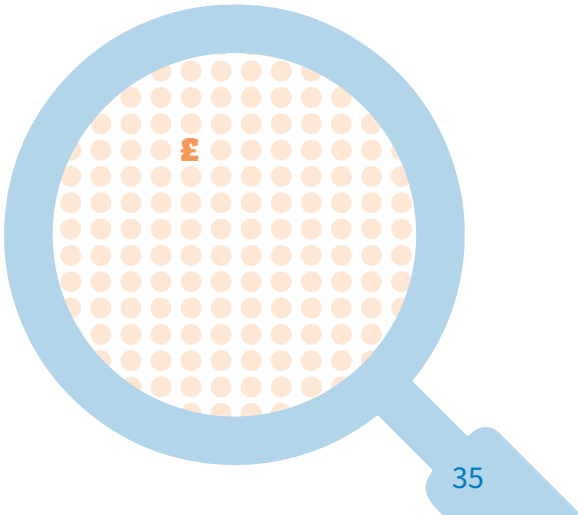
Funding decisions

The UK's cost-effectiveness decision-making architecture creates a 'missing middle' for rare conditions by channeling all treatments into two distinct pathways.

Both NICE (whose decisions apply in England, Wales and Northern Ireland) and the Scottish Medicines Consortium (SMC) have developed pathways to adapt their decision-making to allow for the challenges in assessing treatments for very rare conditions.

NICE's Highly Specialised Technologies (HST) pathway serves only ultra-rare conditions affecting fewer than 300 patients in England, with the headline adjustment in process being a raised threshold of £100,000 to £300,000 per QALY which allows for higher costs and/or greater uncertainty.

SMC's Ultra Orphan Pathway is broader in scope, including conditions affecting fewer than 1 in 50,000, and facilitates early access to treatments while additional evidence is gathered.



Rare conditions that are more prevalent than these thresholds must be assessed using standard routes. Rare conditions that affect between 300 and 29,000 people (the approximate maximum population of people living with a rare condition in England) have no adjustments written into NICE's Single Technology Appraisal methodology. Rare conditions affecting between 1 in 50,000 and 1 in 2,000 people in Scotland are assessed through SMC's 'standard route', though, medicines for rare conditions may be eligible for a Patient and Clinician Engagement (PACE) meeting if the initial assessment by SMC's New Drugs Committee is 'not recommended.

Therapies for these conditions are appraised against benchmarks never designed for small patient populations, making positive recommendations extremely difficult. The result is systematic inequity: people with all but the least prevalent rare conditions are most likely to be denied access to potentially life-altering treatments.

A robust audit with international comparison is necessary to understand whether our decision-making pathways are fit for purpose. This needs to be based both on treatments' progress through our systems, and those that may not be launched in the UK.

Planning and funding of services for rare conditions

Services for small populations are called 'specialised services', these services include those for rare conditions. Specialised services are organised by medical discipline or hospital department, not according to how rare conditions affect people or how care might best be deployed across their journey. While many individual approaches work well, there is no programme assessing the services from the perspective of people with rare conditions and there is no process to identify and fill gaps.

Each of the four UK nations maintains its own framework for designating, funding and delivering specialist services for rare conditions. Cross-border healthcare arrangements exist to enable people with rare conditions to access services beyond their home nation, typically from Scotland, Wales and Northern Ireland to specialist services in England. However, these pathways are often unclear and require additional referral processes that can result in some people with rare conditions failing to access the care they need. Cross-border treatment also imposes significant travel costs and time burdens on people with rare conditions, creating a practical barrier that compounds the administrative friction of navigating between different national systems.

The NHS can assess whether specialised services perform well against their own specifications, but there is no review process to assess whether these are the right services for the overall population of people living with rare conditions. Without an overarching equitable process to identify rare conditions without services and incentivise the development of services to treat them, inequitable service provision for rare conditions will persist.

In England a significant amount of rare condition commissioning has been delegated to 42 individual Integrated Care Boards (ICBs). This fragmentation renders people living with rare conditions statistically invisible, their numbers never reach the threshold needed to compete with high-volume local priorities. There is particular concern that ICBs may not prioritise rare conditions, especially neurological services where many rare conditions cluster. Specialist expertise concentrates in major metropolitan hubs, imposing a significant 'travel tax' on people from rural areas. Lack of integration between the four UK nations creates administrative friction for cross-border referrals, meaning a person's postcode determines their access to expertise.

There are clear opportunities to conduct meaningful audits of rare conditions care. Lists of rare conditions could be systematically matched against service specifications to identify coverage gaps. For example the panels of genes specified by the NHS England national genomic test directory for rare conditions are functionally a list of rare conditions that can be diagnosed through genetic testing in England. These could be mapped against clinical pathways to reveal where diagnostic capability exists without corresponding treatment pathways, or vice versa. These approaches would provide, for the first time, a national picture of the rare pathway and how equitable it is for the whole community of people living with rare conditions. Currently, bodies with oversight responsibility focus on ensuring quality of services that are commissioned, there is much less analysis of gaps in the coverage of these commissioning decisions.

Establishing oversight processes with responsibility for auditing whether service structures appropriately meet the needs of people with rare conditions is necessary. This should include systematic mapping of rare conditions against service specifications and clinical pathways to identify and address gaps, ideally by comparing pre-existing information. All rare conditions should have clearly defined commissioning routes with stated rationales, whether this is condition-by-condition, or organised by rare condition groupings. Commissioning decisions, both positive and negative, should be transparently recorded and easily accessible to people with rare conditions, healthcare professionals and support organisations.

MND Association: Inequitable access to treatment for people with SOD1 MND

Motor Neurone Disease (MND) is a rapidly progressive condition affecting the brain and spinal cord. There are around 5,000 people in the UK with the condition at any one time, with six people diagnosed and six dying each day. It leads to loss of movement, speech and, eventually, breathing, with over half of people dying within two years of diagnosis.

For the 2% of people with the genetic SOD1 form of MND a new treatment, the first MND treatment in nearly 30 years to be licensed by the MHRA, has been shown to significantly slow, and in some cases halt, disease progression. This is completely unprecedented in MND care.

However, access to this drug is deeply inequitable. While we await NICE approval, the drug is provided free through an Early Access Programme. However, NHS Trusts must fund the resources needed for the monthly lumbar punctures required to administer. Some trusts cannot meet the costs, while others refuse to engage in the EAP process. This has created a postcode lottery where access depends on local capacity rather than clinical need.

At least four people have died waiting for treatment, while others have permanently lost function.

This case exposes a systemic failure: lifesaving medicines can exist, be free, and be clinically endorsed yet remain out of reach for the people who need them.

The MND Association is calling for:

- National intervention and emergency funding to support delivery of promising, lifesaving treatments ahead of NICE appraisal.
- Establish clear eligibility criteria (including severity of disease, effectiveness of treatment, and likelihood of a positive NICE decision) to guide when exceptional medicines merit early national support.
- Immediate investment in the neurological workforce and infrastructure to ensure equitable rollout of current and future precision medicines.

4. Embed rare conditions into mainstream healthcare delivery

The UK's current healthcare reforms offer a rare opportunity to move complex conditions from the margins to the centre of national strategy. If the NHS is equipped to support those with rare conditions, it becomes more resilient for all.

The 10-Year Health Plan for England offers a transformative vision for the NHS, yet there remains significant uncertainty about implementation and timelines for delivery remain unclear. Simultaneously, Scotland has signaled a fundamental shift in approach to healthcare delivery, with work underway to develop a Long Term Conditions Framework. As elections approach in Scotland and Wales, this changing political landscape adds further ambiguity to long-term healthcare priorities across the devolved nations.

Recognising that while there is this uncertainty, this period of transition, aligned with the one year extension of the UK Rare Diseases Framework, presents a vital opportunity for the four nations to co-produce a resilient healthcare model for rare conditions. By embedding rare condition expertise into the system by design, we can ensure that they remain a foundational priority.

The strategic shift toward community-based care, digital transformation, and prevention offers a unique chance to embed rare conditions into the broader healthcare infrastructure. Rather than treating these conditions as isolated 'niche' cases, they should be viewed as the ultimate test for a modernised health service. For example:

- Utilising the proposed shift from hospital to community to mainstream genomic medicine brings the opportunity to improve diagnosis.
- The move toward a unified genomic record and the expansion of the NHS App allows for seamless data flow. This ensures that data about rare conditions is accessible at every point of care.

Treating rare conditions as a core priority is essential to preventing a two-tier healthcare system. Without this structural integration, we risk formalising health inequities and ensuring that medical innovation only reaches common conditions, leaving those with rare conditions behind.

Future of specialised commissioning position statement: Specialised Healthcare Alliance, Genetic Alliance UK and Neurological Alliance

The 'Future of specialised commissioning' position statement was developed by the Specialised Healthcare Alliance (SHCA) in collaboration with The Neurological Alliance and Genetic Alliance UK.

The statement outlines a unified vision for future commissioning, emphasising that reforms following the abolition of NHS England must prioritise people over administrative processes. The alliances collectively advocate for maintaining national service standards and clinical leadership while addressing systemic gaps, such as the lack of coordination between physical and mental health services.

By joining forces, the organisations call for increased transparency, regular engagement with support organisations, and clear accountability mechanisms. Their shared goal is to ensure that the delegation of services to local Integrated Care Boards (ICBs) does not result in a fragmented 'post-code lottery' for specialised care.

5. Commit to a bold successor to the UK Rare Diseases Framework

The current UK Rare Diseases Framework has been a catalyst for delivering a more equitable healthcare system for people with rare conditions. Significant work has been undertaken to address the inequities experienced and often perceived by the rare conditions community. Through a variety of initiatives, such as the establishment of dedicated information hubs, resources for healthcare professionals, and pilot care coordination initiatives such as the Syndromes Without A Name (SWAN) clinic in Wales, certain inequities have been addressed. While these initiatives are excellent examples of tools designed to tackle specific barriers, they primarily improve the experience of equity for certain segments of the rare conditions community. They do not, however, resolve the fundamental systemic challenges inherent in delivering truly equitable healthcare for all rare conditions.

However, the Framework has been instrumental in taking steps to address the three fundamental challenges to equitable care: low priority, low clinical familiarity, and low evidence. This has been achieved through several strategic interventions.

- **Robust data infrastructure**: National rare disease registries have been implemented across England, Wales, and Scotland to capture high-quality longitudinal data for research and treatment development. Beyond this, these platforms hold the potential to accelerate diagnosis by enabling the identification of rare clinical patterns more rapidly through centralised data. They also offer data to improve service planning. By mapping people with rare conditions, health authorities can more effectively commission specialist centres and ensure resources are strategically allocated.
- **Collaborative research ecosystems**: The establishment of Rare Disease Research UK has formalised collaboration between academia, people with rare conditions, and the third sector. With 11 specialised research nodes focusing on both specific and cross-cutting themes, complemented by a comprehensive landscape review commissioned under the England Action Plan, the UK is better positioned to understand what is needed to improve rare conditions research in the UK.
- **Quality standard for rare conditions**: To ensure consistency and cost-effectiveness in service delivery, an Independent Advisory Group was convened to develop the Rare Disease Quality Standard. This framework provides a benchmark for excellence, ensuring that high-quality care is delivered for people affected by rare conditions.

The one-year extension to the UK Rare Disease Framework is a welcome step, providing essential continuity in the context of widespread NHS reform in England and the devolved nation elections in Scotland and Wales in 2026. However, it is vital that a successor policy is secured with a clear, long-term commitment from all four nations.

Allowing the Framework to lapse in January 2027 without a formal successor would jeopardise the considerable momentum built over the last five years. There is a significant risk that progress will plateau just as the broader healthcare landscape undergoes its most substantial digital and structural transformation in a generation. To ensure the rare conditions community benefits from these systemic changes, and is not further disadvantaged by them, a permanent and evolved policy successor to the Framework is necessary.

It is only with a successor framework that real inroads can be made to tackling the three challenges to delivering an equitable healthcare service for rare conditions .

Conclusion

We recognise that this report is not the definitive answer to the vast challenge of health inequity. Our focus has been deliberately specific, discussing the systemic challenges that arise from the immutable fact of small population sizes inherent in rare conditions.

While the statistical reality of these small populations cannot be changed, it must never be used as a justification for systemic neglect. We have sought to demonstrate that although we cannot change the number of people affected by a rare condition, we can and must change how the system perceives them. By implementing smarter data-sharing frameworks, flexible clinical trial models, and heightened clinical awareness, we can ensure that rare does not mean invisible.

We also know that inequity for people with rare conditions is often layered. Factors such as ethnicity, socio-economic background, and geography intersect to create unique barriers and profound disparities. Vital work is being done across the sector to explore these intersectional inequities, and it is essential that this evidence is used to drive concrete policy changes.

This is why we require a robust, coordinated four nations approach. We must ensure that this broader body of research is not only heard but that its recommendations are actioned. Only by addressing both the systemic challenges of scarcity and the complexities of intersectional inequity can we begin to deliver a fair healthcare system for every person living with a rare condition.

Equality is one thing, equity is another

Equality, is one thing,
Equity, is another
This short, little rhyme
May help others to discover
Equality actually means,
We're all treated the same
But with our rare conditions
We are playing a different game
Equality is all well and good
To many, it may seem fair
But I can tell you this
It's useless if you're rare

Equality, treats us all alike
And that may well seem fine
For an equal chance to achieve
We need a tailored package to align

Equity, is about fair play
Unequal treatment can make things just
We need bespoke support,
Specialist care, usually, a must

Equity is, a big step forward
In giving us what we need
It's what rare conditions require
And hardly a sign of greed

Equity, is more than we can imagine
More than a dream in a pipe
It is within our grasp
I can sense it, as I type

Equity then, not equality
The message we wish to convey
Let's get right behind it on
Rare Disease Day

Equality is one thing, equity is another

Poem by Anthony Heard, Reading (immune thrombocytopenia and fibromyalgia)

Anthony kindly provided this poem for our Rare Disease Day report. You can find more of his work and many other creative pieces from the rare community in 'More than you can image: an anthology of rare experiences'

GENETIC ALLIANCE UK