

Delivering a fairer healthcare system for people with rare conditions

Funding statement: Our Rare Disease Day 2026 campaign is delivered by the Genetic Alliance UK team and funded via sponsorships and grants. Thank you to our industry supporters. These organisations have had no involvement in the development of the campaign, or any of its content. Without their support our campaign would not have been possible. **A full list of funders can be found at the end of this information sheet.**

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.

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.



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

geneticalliance.org.uk/rdd26

“ 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)

“ We've had to rely on our own skills as highly educated individuals to essentially project-manage the entire process. From researching specialists, coordinating appointments, and pushing for tests to secure a diagnosis and then appropriate care. This experience felt deeply unfair because the system placed the burden on us rather than providing clear pathways or proactive support. It highlighted how those without similar resources or knowledge would likely face even greater barriers, making equitable access almost impossible. We are so thankful we have the resources to do this for our daughter. ”

- Parent of a person affected by SYNGAP1

Equity is...

A timely, accurate diagnosis.



Supporting people with rare conditions and their families to attend appointments that are far from home



Healthcare professionals being trained to identify rare conditions



Access to psychological support, social care, and education



Better screening programmes to identify rare conditions earlier



Having a care coordinator



Access to the medicine you need through a simple, timely process regardless of where you live in the country



Healthcare systems providing specialist diets and non-medicine treatment where necessary



A personalised care plan to help join-up services and reduce the burden on families



Receiving treatments in the most accessible and manageable form possible



Seamless communication between specialists, hospitals, and GPs



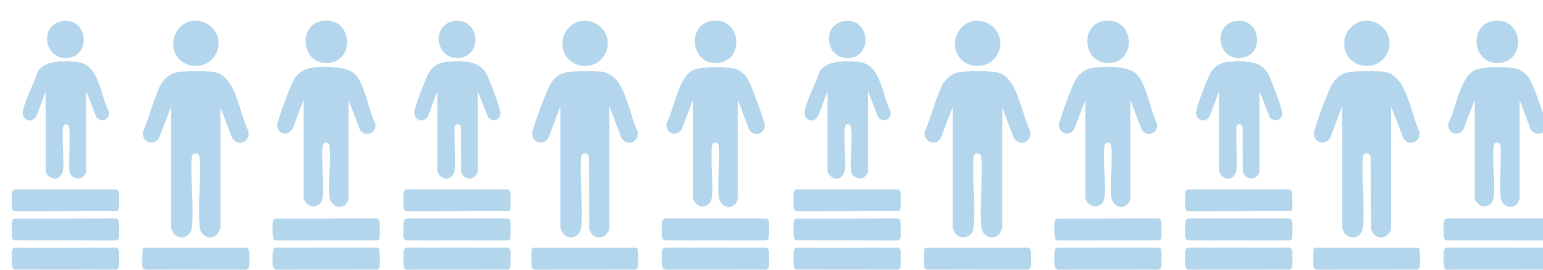
Prioritising research into rare conditions



Access to clinical trials and removal of financial and geographic barriers to participation



Access to the appropriate level of specialist care regardless of where you live



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.

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 conditions.
- **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 disease 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 disease community with regards to receipt of a diagnosis and access to health and social care services.

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.

“ **Equity is being seen as a person not a problem.** ”
- Person affected by a mitochondrial disease

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.’

The Rare Disease Quality Standard: A tool for equity

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.

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](#).

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