

What are the drivers of inequity for rare conditions?

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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. [Genetic Alliance UK's Equity for Rare: Delivering fairer healthcare systems for people with rare conditions report](#) explains 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.

England Rare Diseases Action Plan 2025 Health inequity scoping review

[Annex E of the England Rare Diseases Action Plan 2025](#) 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 and 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.

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



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

geneticalliance.org.uk/rdd26

This scarcity creates three key challenges:



Low priority

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.



Low clinical familiarity

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.



Low evidence

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.

“ Rare isn't taken seriously. Rare seems to be the continuing theme throughout 'it's a rare condition, so symptoms and next steps are rare too'. I also think the fact it's mainly invisible is a huge factor into why it's not taken seriously enough. I find it unfair because I'm supposed to trust these professionals with my care and wellbeing but am often met feeling the opposite. ”

- Person affected by PTEN/Cowden syndrome

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