

Intersectional barriers to rare conditions healthcare



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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 the members of the rare condition community most in need of support. Addressing these issues requires a collaborative effort, and we recognise the invaluable work of other experts, patient advocacy groups, and researchers who continue to highlight how wider societal disparities compound the difficulties of living with a rare condition.

“ There is a critical need to include more people from ethnic and racial minorities and their families in rare conditions 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, rare kidney conditions

“ Equity for the myasthenias means acknowledgement of the significant health, social, financial, and psychological hurdles patients need to overcome before and after diagnosis. Myasthenia is a disease that is misunderstood and misdiagnosed, and like many rare conditions, a person's journey is heterogenous. The difficulty and length of this journey is influenced by factors beyond their control - geography, socioeconomic background, race, and gender, to name a selection. ”

- Representative from myasthenia community

“ Equity is when 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



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

geneticalliance.org.uk/rdd26



◀ The Seven Anti-Racism Principles graphic produced by the NHS Race and Health Observatory and available on their website

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.

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.

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.



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 disease inequity: achalasia and delayed diagnosis

The report documents how patients 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 patients 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.

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