

How women are revolutionising rare disease

Women have played a crucial role in rare disease awareness and better care, overcoming gender biases and systemic challenges to drive progress in this often-overlooked field.



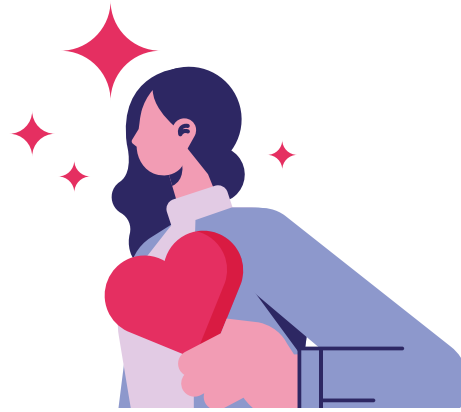
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Historically, women have been marginalised with their needs often being unmet as compared with men — a situation mirrored to a greater degree in the rare disease sector as compared with common conditions. This field grapples both with gender disparities and the broader challenge of gaining recognition for rare diseases within healthcare systems.

Women leading rare disease advocacy

Women, who traditionally take on caregiving roles, have been at the forefront of rare disease advocacy. Empathy and strong communication skills make them effective as both caregivers and advocates. These roles are often assumed out of necessity when they notice symptoms in their children — a common situation as 70% of rare diseases first present in childhood.¹ Women utilise various



approaches to raise awareness, from 'kitchen table' support groups to larger awareness campaigns to lobbying governments.

Female struggles in rare disease diagnosis

The diagnostic journey for rare diseases is notably lengthy, often taking four to eight years or more.² Women face additional delays due to gender biases in the healthcare system, with symptoms often being unrecognised or misattributed to psychological issues. They frequently encounter dismissive attitudes from clinicians, longer diagnostic journeys compared to men and poor socioeconomic support, resulting in substantial psychological, social and financial burdens.

Industry challenges women encounter

Women's lived experiences and the challenges they face in caregiving

roles empower them to be empathetic and effective leaders in the rare disease space. Their advocacy has led to increased awareness and funding for rare disease coordination and drug development.

However, across industries, including the pharmaceutical industry, leadership roles are predominantly occupied by men. Women in this field often struggle to advocate for themselves and achieve recognition. This is problematic since only with leadership diversity can we ensure comprehensive decision-making that reflects the population's needs.

Empowerment through education

With the efforts of advocates, clinicians are increasingly aware of the rare diseases they may encounter. However, they continue to struggle with recognising when a patient may have a rare disease; which diagnostic tests to run; where to refer them; and how best to treat them.

Medscape, a global leader in medical news, references and education, is tackling this issue by providing the largest collection of accredited rare disease education — free for clinicians — in collaboration with rare disease societies, advocates and key opinion leaders. This educational resource centre, Pathways in Rare Disease, was designed based on feedback from almost 1,000 clinicians across 16 different specialties.³

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References

1. Nguengang Wakap S, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* 2020;28:165-173.
2. The Lancet Global Health. The landscape for rare diseases in 2024. *Lancet Glob Health.* 2024;12:e341.
3. Rohani-Montez SC, et al. Educational needs in diagnosing rare diseases: A multinational, multispecialty clinician survey. *Genetics in Medicine Open.* 2023;1: 100808.

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