RETHINKING RARE DISEASE SYSTEMS FOR WOMEN

A Call to Action from the Society for Women's Health Research

Society for Women's Health Research

BACKGROUND

A rare disease is defined as a condition or disease that affects fewer than 200,000 people. There are about 10,000 known rare diseases, impacting an estimated total of 30 million Americans.[1] Women are disproportionately impacted by a number of rare diseases; fibromuscular dysplasia (FMD) and lymphangioleiomyomatosis (LAM) are just two examples, and women account for 60% of autoimmune patients – many of which are considered rare.[2,3,4] Yet, a comprehensive understanding of the impact of rare diseases on women remains severely deficient, as women have been only marginally considered in rare disease research, policy, and care.

Across conditions, women experience unique biological factors that influence their diagnosis, treatment, and long-term health. Women's life stages – from menarche and reproductive years to menopause – introduce additional considerations for rare disease detection and management. Hormonal fluctuations, reproductive health considerations, and sex-specific disease expression must be accounted for in research and approaches to screening, diagnosis, and health care.

At the same time, gender roles and societal factors also influence the rare disease experience for women. For example, women are more likely to encounter bias within the health care system. Their symptoms may be dismissed, minimized, or misattributed, contributing to a longer diagnostic odyssey. A 2024 study found that women with rare diseases wait an average of 5.4 years for diagnosis, compared to 3.7 years for men.[5] Such delays lend to missed treatment windows and avoidable physical, emotional, and financial burdens.

CALL TO ACTION

While individual diseases may be uncommon, the category of rare disease is not. Women living with rare diseases are too often navigating systems not designed for them. As policymakers, regulators, researchers, health care professionals, and patient advocates engage in solutions related to rare disease, there is ample opportunity to center women and their unique needs. In fact, solutions developed for rare diseases can serve as a model for broader care transformation. By designing a medical research and health ecosystem that is patient-centered, we can improve outcomes not only for individuals with rare disease, but across the entire patient population.

The Society for Women's Health Research (SWHR) emphasizes the following priority areas to support women living with rare disease and their caregivers.

- **Close Evidence Gaps to Enable the Development of Therapies in Rare Disease.** A strengthened and cross-cutting research agenda is essential to closing gaps in understanding and treating rare diseases. With an estimated 80% of rare diseases having a genetic component,[6] there is opportunity to better understand genetic and molecular mechanisms, including why certain conditions disproportionately impact women. Adherence to the National Institutes of Health (NIH) sex as a biological variable (SABV) policy across study

design and analysis is critical in this work. Similarly, all federally-funded medical research should prioritize embedding sex- and gender-informed approaches. Doing so will improve the evidence base that is foundational for diagnostic accuracy, illuminating sex-specific, clinical considerations, and advancing innovative treatments like genetically targeted therapies.

- **Improve Insurance Coverage Policies and Coordinate Care Across the Lifespan.** Women with rare diseases often experience fragmented care across the lifespan. Expanding access to care and strengthening health insurance coverage will improve timely evaluation, personalized care, and sustained treatment for women living with rare diseases. Passing legislation to address step therapy (otherwise known as "failure first") protocols can bridge barriers in patient access to appropriate treatments. Other federal and state policy efforts – particularly those focused on Medicaid eligibility and benefit design – are important opportunities to improve care access for rare disease patients, especially for those with complex, chronic, and disabling rare conditions who need access to multidisciplinary and long-term care, which are often excluded or fragmented under standard benefit structures.

  Furthermore, coverage policies should support integrated care models that connect specialties such as genetics, obstetric-gynecology, endocrinology, and mental health, and ensure seamless transitions between pediatric, adult, and geriatric care. This can be achieved, in part, by addressing prior authorization burdens and coverage discontinuities that extend the time to diagnose, delaying and interrupting treatment for patients with rare diseases.

- **Generate Lifespan- and Sex-Informed Rare Disease Evidence Through Modern Data Infrastructure.** Unlocking the full value of patient information to accelerate rare disease research and clinical care requires modernizing data infrastructure. This includes investing in interoperable systems that enable secure data sharing across providers, researchers, and systems. It may also include supporting patient-centered registries and platforms that allow patients to contribute longitudinal health data. Finally, policy efforts should explore ways to improve rare disease representation in current medical coding and classification systems, including ICD diagnosis codes, to better capture the complexity of rare diseases and sex-specific differences which are often underrepresented in existing datasets. Strengthening the rare disease evidence base will improve understanding of sex-specific and time-dependent disease trajectories and inform more precise treatment pathways.

- **Recognize and Support Caregivers – Without Overlooking Women as Patients**. Women disproportionately shoulder caregiving responsibilities, yet many women are operating not only as part of the "sandwich generation" of caregivers balancing caregiving for both children and aging parents, but are also in a dual capacity as both caregivers and patients – a role that is often overlooked. While expanding access to caregiver education, financial supports, respite services, and community-based resources are essential (alongside greater recognition of unconventional caregivers, such as children and siblings caring for parents or siblings), policy must also ensure that caregiving status does not obscure women's clinical needs or delay their own evaluations and treatment. This means strengthening systems that identify and support caregivers early, streamlining federal assistant processes to reduce administrative burdens associated with complex paperwork and eligibility processes, and explicitly integrating safeguards that ensure women in caregiving roles are still recognized, assessed, and treated as patients in their own right.

1. U.S. Food and Drug Administration. (2024). Rare diseases at FDA. FDA.
2. McCarthy, C., Gupta, N., Johnson, S., et al. (2021). Lancet Respir Med.
3. Khoury, M. H., Hershey, S., & LeLeiko, R. M. (2024). US Cardiology.
4. Abend, A. H., He, I., Bahroos, N., et al. (2024). J Clin Invest.
5. Faye, F., Crocione, C., Anido de Peña, R., et al. (2024). Eur J Hum Genet.
6. Gürkan, H., & Bilge Satkın, N. (2025). Balkan Med J.