https://doi.org/10.35845/kmuj.2025.24141

EDITORIAL

Invisible burden: addressing rare and genetic disorders in Pakistan's healthcare landscape

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THIS ARTICLE MAY BE CITED AS: Sherin A. Invisible burden: addressing rare and genetic disorders in Pakistan's healthcare landscape. Khyber Med Univ J 2025;17(3):249-50. https://doi.org/10.35845/kmuj.2025.24141

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Rare and genetic disorders remain largely invisible in Pakistan's healthcare landscape, yet they place a devastating burden on patients, families, and health systems. Their neglect in national health agendas contrasts sharply with global commitments under the UN 2030 Agenda, Universal Health Coverage (UHC), and the 2021 UN Resolution, which recognize equity for persons living with rare diseases as a priority.[1] However, the absence of a universally accepted definition, thresholds and criteria vary widely across countries, continues to hinder coherent policy and regulation.[2] Establishing a consistent definition is essential to guide strategies, prioritize research and drug development, and reduce inconsistencies shaped by demographic and socio-cultural contexts.[3] Rare diseases, though individually uncommon, collectively affect an estimated 3.5-5.9% of the global population, about 263-446 million people at any time. With nearly 72% of these conditions being genetic and about 69.9% often manifested in childhood, their burden is profound yet largely invisible within health systems.[4]

While global figures highlight the magnitude of rare and genetic disorders, the challenge is even greater in developing countries, where fragile health systems, scarce resources, and competing priorities limit progress.[5,6] In such contexts, the lack of clear definitions, inadequate diagnostic infrastructure, and weak policy attention widen inequities. Pakistan exemplifies these challenges, highlighting the urgent need to translate global commitments into locally responsive strategies.

Pakistan, as a member of the Undiagnosed Diseases Network International, is working to establish a framework for the detection and care of undiagnosed rare diseases. However, significant challenges persist, including a shortage of experts, limited funding, low awareness among both the general population and healthcare providers, restricted access to specialists and genetic testing facilities, weak coordination among healthcare institutions, and the absence of culturally appropriate care models. Contributing factors such as high rates of consanguinity, gender disparity, stigma, cultural and religious influences, and limited public understanding of the genetic basis of these conditions further compound the problem.[7,8]

To address these challenges, Pakistan requires a comprehensive response that aligns with international standards while remaining sensitive to local needs and cultural contexts. Notable progress has been made in building local infrastructure and expertise, with the establishment of the Rare Registry for inherited metabolic disorders by the Biochemical Genetics Laboratory at Aga Khan University serving as a key milestone in data collection and disease burden assessment.[9] However, there remains an urgent need for a coordinated, nationwide registry to capture the prevalence of rare and genetic disorders across all provinces.

Simultaneously, strengthening diagnostic infrastructure through the expansion of genetic laboratories and ensuring equitable access to early detection services must be prioritized. Addressing the critical shortage of trained professionals requires targeted capacity-building initiatives for clinicians, pediatricians, and genetic counselors. Public-private partnerships can also play a transformative role by reducing financial barriers, making advanced diagnostics and long-term care more accessible in low- and middle-income settings.[10]

Professional societies such as the Pakistan Society of Medical Genetics and Genomics (PSMG)[11] and the recently established Pakistan Society for Novel and Rare Diseases (SNARE)[12] are well positioned to advance the field of medical genetics and rare disease care. By fostering interdisciplinary collaboration, raising awareness among both professionals and the public, and supporting education and telehealth initiatives, these organizations provide vital platforms for advocacy and innovation. Their efforts are equally crucial for promoting the culturally sensitive integration of medical genetics into Pakistan's healthcare framework.

Pakistan stands at a pivotal juncture in addressing rare and genetic disorders. The path forward requires a coordinated national strategy that integrates registries, diagnostic infrastructure, workforce development, and sustainable financing into a cohesive framework. Aligning these efforts with global commitments under the UN 2030 Agenda and UHC will not only reduce inequities for patients and families but also strengthen the resilience of the entire health system. By leveraging professional societies, fostering public-private partnerships, and promoting culturally sensitive care, Pakistan can bring the "invisible burden" of rare diseases into the national spotlight. This approach can bridge science, policy, and compassion to create a more equitable and hopeful future for those affected.

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CONFLICT OF INTEREST

The author declared no conflicts of interest, financial or otherwise, that could compromise the integrity, objectivity, or validity of their opinions.

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