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Editorial

The Society for Novel and Rare Diseases (SNARE)

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Introduction

In an era of rapid medical advancements, individuals suffering from rare diseases often remain overlooked. These orphan diseases, affecting only a small fraction of the population, are frequently deprioritized by pharmaceutical companies, medical researchers, and healthcare policymakers. The lack of awareness and resources dedicated to rare diseases leaves countless patients undiagnosed and untreated, leading to significant social and medical challenges[1].

In Pakistan, the situation is even more dire due to cultural and social factors. The widespread lack of awareness and prevalence of consanguineous marriages, particularly in rural areas, contribute significantly to the high burden of rare genetic disorders. However, these issues remain largely unaddressed, compounding the struggles of affected families[2].

Across Pakistan, physicians regularly encounter unexplained symptoms and complex cases that pose significant diagnostic difficulties. Identifying a rare disease requires clinical persistence, multidisciplinary collaboration, and specialized expertise. Unfortunately, many patients are misdiagnosed with psychosomatic conditions or dismissed as malingerers, forcing them to seek treatment from quacks and fraudulent practitioners—further exacerbating their suffering and delaying proper care[3,4].

The Society for Novel and Rare Diseases (SNARE) is a duly registered, non-profit, multidisciplinary organization committed to advocacy, research, and patient support for those affected by rare diseases. SNARE's mission is to bridge the gap between patients, researchers, and policymakers, ensuring that rare diseases receive the attention and resources they deserve. In the United States, a disease is classified as rare if it affects fewer than 200,000 people. In the European Union, it is defined as affecting fewer than 1 in 2,000 individuals. Despite their low individual prevalence, over 7,000 rare diseases collectively impact approximately 300 million people worldwide[5]. In Pakistan, the definition and data surrounding rare diseases remain unclear, highlighting the urgent need for comprehensive research and national-level policy frameworks—a challenge that SNARE is committed to addressing.

Patients with rare diseases in Pakistan face delayed diagnosis, limited awareness, and scarce access to specialized testing and treatment. Even when a diagnosis is established, treatment remains a formidable challenge due to financial constraints, lack of infrastructure, and limited pharmaceutical interest. This frequently results in social isolation, psychological distress, and diminished quality of life.

To amplify its impact, SNARE is dedicated to establishing Centers of Excellence in collaboration with leading hospitals and universities, ensuring that rare disease patients receive specialized care from expert physicians and researchers. The organization also advocates for sustainable screening programs to prevent rare genetic disorders, engages with policymakers to promote legislative reforms, funding, and healthcare initiatives for rare diseases, and collaborates globally with similar organizations to advance research, treatment options, and patient support networks.

Organizations like SNARE serve as a lifeline for millions worldwide, amplifying the voices of patients, driving medical breakthroughs, and ensuring that no individual is neglected due to the

rarity of their condition. However, we cannot do this alone. We urge governments, pharmaceutical companies, researchers, and the general public to join hands in supporting the rare disease community. Whether through donations, volunteering, or policy advocacy, everyone has a role to play in advancing rare disease awareness and treatment.

As medical science progresses, SNARE is committed to ensuring that no disease is too rare to matter, and no patient is left behind. Through our collective efforts, we move one step closer to a world where every patient, regardless of how rare their condition is, receives the care and support they deserve.

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