

A Comprehensive Framework for Rare Disease Management in Primary Care

Mike George ^{1,*}

¹Indiana University Bloomington U.S.

Abstract

Rare diseases, though individually uncommon, collectively affect millions globally, presenting unique challenges in healthcare. This paper proposes a comprehensive framework for primary care practitioners, focusing on the diagnosis, management, and advocacy for rare disease patients. By synthesizing international strategies and local practices, this approach aims to enhance patient outcomes and support families effectively. JRC D 2024; 4(8): 163–165

key words: rare diseases, primary care, general practitioners, diagnosis delays, orphan drug act

Introduction

Rare diseases, defined as conditions affecting a small percentage of the population, collectively impact millions worldwide. While each rare disease may be uncommon individually, their combined prevalence represents a significant public health concern. Estimates suggest that between 6% and 10% of the population are affected by a rare disease at some point in their lives, translating to over 1.2 million US, 30 million Europeans, and 25 million Americans living with such conditions. Despite this notable prevalence, rare diseases often remain underrecognized and inadequately addressed within healthcare systems [1].

For patients and their families, rare diseases pose unique challenges, including prolonged diagnostic delays, frequent misdiagnoses, and limited access to specialized care and treatments. These issues often result in inappropriate medical interventions, heightened psychological stress, and substantial financial burdens. In particular, the lack of cohesive national strategies in countries like further exacerbates these difficulties, leaving patients to navigate fragmented care pathways [2].

Primary care practitioners, as the first point of contact for most healthcare needs, are uniquely positioned to address many of these challenges. General Practitioners (GPs) not only play a critical role in early diagnosis but also provide holistic, relationship-based care that extends beyond medical management to include patient advocacy and family support. However, the absence of a

structured approach to rare disease management within primary care settings often limits the effectiveness of this role [3].

How Common Are Rare Diseases in the USA?

Rare diseases are collectively significant in the United States, with estimates indicating that approximately 25 million Americans live with a rare condition. This prevalence translates to about 8.4% of the population, based on a national population of around 298 million at the time of the estimate. These figures highlight that, while individual rare diseases are uncommon, their cumulative impact is comparable to that of other major health concerns, such as type 2 diabetes [3].

In the U.S., the National Organization for Rare Disorders (NORD) and the National Institutes of Health (NIH) Office of Rare Diseases (ORD) play central roles in raising awareness and supporting patients with rare diseases. These organizations have driven initiatives like the Rare Diseases Act (2002) and the Orphan Drug Act (1983), which aim to enhance research, drug development, and patient care for rare conditions [4]. Despite these efforts, many rare diseases remain underdiagnosed or misdiagnosed due to their low prevalence, lack of specialized knowledge, and limited resources within the general healthcare system.

The statistics underline the necessity of a coordinated healthcare approach that integrates primary care and specialized services to address the unique challenges

faced by individuals with rare diseases in the United States.

Varied Conditions, Shared Challenges

Rare diseases encompass a wide spectrum of conditions, ranging from genetic disorders like Marfan syndrome and Huntington's disease to acquired conditions such as Guillain-Barré syndrome and listeriosis. Despite the diversity in their etiology, presentation, and management, individuals with rare diseases often share remarkably similar challenges and experiences within the healthcare system [5].

One of the most prevalent issues is the delay in diagnosis. Studies indicate that patients frequently wait years—from 5 to 30 years in some cases—to receive a definitive diagnosis. Misdiagnoses are also common, with many patients undergoing inappropriate treatments, surgeries, or psychological interventions before the correct condition is identified. For instance, one survey found that 40% of rare disease patients were initially misdiagnosed, leading to unnecessary surgeries (16%), incorrect medications (33%), or unwarranted psychological care (10%) [6].

Beyond medical challenges, patients and their families face substantial social and emotional burdens. Common difficulties include limited access to accurate information, lack of knowledgeable healthcare providers, and high treatment costs. Many families struggle with the social consequences of a rare disease, such as isolation, financial strain, and difficulties in accessing education or employment opportunities for affected individuals [7].

These shared experiences highlight the systemic gaps in healthcare that disproportionately affect rare disease patients. The lack of a coordinated approach exacerbates these issues, leaving many patients to navigate complex healthcare systems without adequate support. Addressing these common challenges through a unified, patient-centered strategy can significantly improve outcomes and quality of life for individuals living with rare diseases and their families [7].

International Strategies for Rare Disease Management

Addressing the multifaceted challenges of rare diseases requires robust, coordinated strategies at the national and international levels. Several countries and regions have implemented pioneering initiatives to improve diagnosis, treatment, and support for individuals with rare diseases. These strategies highlight the importance of policy-driven approaches, patient advocacy, and collaboration across sectors.

United States: Legislative and Advocacy Framework

The U.S. has been at the forefront of rare disease management with landmark legislation such as the Orphan

Drug Act (1983) and the Rare Diseases Act (2002). The Orphan Drug Act incentivizes pharmaceutical companies to develop treatments for rare diseases by offering tax credits, market exclusivity, and grants for clinical research. This has led to the development of numerous "orphan drugs," providing life-saving treatments for conditions that would otherwise be neglected due to their lack of commercial viability.

The National Institutes of Health (NIH) established the Office of Rare Diseases (ORD) to enhance research and information dissemination. The ORD works closely with the National Organization for Rare Disorders (NORD) to connect patients with resources, promote awareness, and support disease-specific advocacy groups.

European Union: Coordinated Policies and Resources

In Europe, the establishment of Eurordis (European Organisation for Rare Diseases) in 1997 marked a significant step in uniting patient advocacy groups across the continent. Eurordis collaborates with the European Union to improve rare disease management through initiatives such as Orphanet, a comprehensive web-based database providing information on rare diseases, centers of excellence, and patient-support networks.

European countries have also implemented national strategies, with France's National Plan for Rare Diseases (2005-2008) being a notable example. This plan emphasizes equitable access to diagnosis, treatment, and care while integrating rare disease education into all levels of medical training.

Canada and New Zealand: Patient Advocacy Focus

Canada and New Zealand have prioritized patient advocacy by establishing national rare disease organizations: the Canadian Organization for Rare Disorders (CORD) and the New Zealand Organisation for Rare Disorders (NZORD). These groups focus on connecting patients with support networks, advocating for policy changes, and improving access to orphan drugs.

Global Collaboration

Internationally, rare disease organizations are increasingly collaborating to address shared challenges. Initiatives such as the International Rare Diseases Research Consortium (IRDiRC) aim to accelerate research and improve treatment accessibility worldwide. By pooling resources and expertise, these collaborations strive to overcome the limitations of individual countries' efforts.

These international strategies demonstrate the power of coordinated action, legislative support, and patient advocacy in transforming rare disease care. They provide valuable lessons for countries, like Australia, that are

still developing comprehensive approaches to address the unique challenges faced by rare disease patients.

Strategies for Rare Disease Management in the United States

The United States has made significant strides in rare disease management, but opportunities remain to enhance its framework further. A key priority is strengthening the epidemiology and data infrastructure for rare diseases. Expanding national registries and leveraging real-time data collection through electronic health records can improve the tracking of rare disease trends and inform policy-making. Additionally, integrating rare disease training into medical education and utilizing advanced diagnostic tools, such as genomic sequencing and AI-based technologies, can address delays in diagnosis. Establishing more centers of excellence will provide patients with specialized multidisciplinary care, accelerating both diagnosis and treatment.

Expanding access to treatments and orphan drugs is another critical area for improvement. While the Orphan Drug Act has incentivized pharmaceutical innovation, many patients still face significant barriers due to high costs and limited availability. Fast-tracking drug approvals and promoting public-private partnerships can bring more therapies to market while reducing financial burdens on patients. Concurrently, empowering patients and families through comprehensive support networks, self-care training, and legal advocacy will ensure they have the tools and protections needed to navigate the challenges of living with a rare disease.

Collaboration and awareness are essential to driving systemic change. Establishing a national task force for rare diseases can align stakeholders across federal, state, and local levels, while fostering global partnerships with organizations like Eurordis can enhance knowledge-sharing. Increasing public awareness through campaigns and media engagement can help reduce stigma and encourage early intervention. Coupled with increased federal funding for research and translational initiatives, these strategies will create a robust foundation for improving rare disease care in the United States.

Conclusion

Rare diseases, though individually uncommon, collectively represent a significant challenge for healthcare systems worldwide, including in the United States. Addressing the unique needs of rare disease patients requires a multifaceted approach that integrates robust data infrastructure, early diagnostic tools, accessible treatments, and comprehensive patient support. By building on existing frameworks like the Orphan Drug Act and fostering collaboration among stakeholders, the U.S. has the potential to lead the way in rare disease management.

Empowering patients and families, enhancing public awareness, and prioritizing funding for research are critical steps to closing the gaps in care. The development of specialized training for healthcare providers and the establishment of centers of excellence will further strengthen the system's ability to deliver timely and effective interventions. These efforts must be underpinned by a national commitment to coordinated strategies and equitable resource distribution.

Through a unified and proactive approach, the United States can not only improve the quality of life for individuals living with rare diseases but also set a global standard for rare disease care. With patients at the center of these efforts, the path forward holds the promise of better health outcomes, greater equity, and a more inclusive healthcare system for all.

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