

Towards a World Health Assembly Resolution on Rare Diseases

Summary

“We are few. We are many.” Each rare disease may in and of itself be rare, but the 6,000+ identified rare diseases affect more than 300 million persons worldwide, a conservative estimate. Rare diseases are at a flex point. They were once thought of as a disparate assortment of isolated conditions believed to affect few people, often in a devastating way, that were considered either too difficult to address or not worth understanding, diagnosing, treating, and supporting. Today, thanks both, to the mobilisation of the civil society - patient and families empowerment, researchers and clinicians actions-, catalysing actions for each disease locally and internationally as well as across all rare diseases to address their common challenges, science is advancing, healthcare provision is improving, policies are emerging in more countries, translating into more people being diagnosed as well as longer survival, better care and well-being for an increasing number of conditions and persons.

Nevertheless, despite significant recent advances in health innovation, research and development, and progress towards Universal Health Coverage (UHC), rare diseases remain a significant unmet need in healthcare. For a broad range of reasons including limited health budgets, resources, and expertise, Persons Living with a Rare Disease (PLWRD) across the globe continue to face unique challenges in accessing appropriate care and support.

Many regions and countries have made commendable progress in addressing the needs of PLWRD, but this progress has not been consistent across all communities. To systemically and sustainably improve the rare disease ecosystem, collective action through a collaborative and multisectoral approach is crucial. To ensure that UHC indeed entails “health for *all*”, rare diseases need to be made a global health priority.

Member States have a unique opportunity to address an existing unmet need in healthcare by adopting a World Health Assembly (WHA) resolution on rare diseases. This action will help shed light on the challenges faced by the more than 300 million PLWRD and will lead to achieving significant returns on investment in science and technology: in policy guided by principles of equity and empowerment, in health systems strengthening for optimized access to diagnosis and care, and in social systems resourcing to support PLWRD and their families. The ideal outcome of a resolution would be for Member States to task the World Health Organization (WHO) with developing a global action plan on rare diseases. A global action plan on rare diseases is urgently needed to propose sustainable and systemic solutions for a better global ecosystem for rare diseases, to ensure greater equity for all PLWRD worldwide, to reach the Global Health 2035 Goals, and to make UHC a reality.

What is a Rare Disease?

Persons living with a rare disease face distinct and significant challenges that arise from the infrequency of their medical condition, such as a long diagnostic journey, inadequate clinical management, and limited access to effective treatments. The burden of rare diseases on patients, their caregivers and families, healthcare systems, and society overall, merits greater visibility and recognition.

A rare disease is a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2000 persons living in any World Health Organization-defined region of the world. Rare diseases include, but are not limited to, rare genetic diseases, rare cancers, rare infectious diseases, rare poisonings, rare immune-related diseases, rare idiopathic diseases, and rare undetermined conditions. Rare Diseases International, in collaboration with a global multi-stakeholder panel of experts and the WHO, led the development of the Operational Description of Rare Diseases as a common reference to inform decision-makers consistently across the world. It describes what diseases are considered rare, how many persons are affected, and why the rare disease community deserves specific attention¹.

The burden of rare diseases on society at large

Each rare disease affects only a small number of individuals. However, considering the 6000 to 10000 rare diseases collectively, a sizeable portion of the general population (estimated 3 – 8%) is affected: over 300 million people globally. Recognizing the breadth of rare diseases across the world and their impact on physical and mental health means that social care systems are key to prioritizing services and improving outcomes².

Many rare diseases are chronic, progressive conditions that result in debilitating impairments. Many are serious and life-threatening. Rare conditions are often described as *complex and multisystemic* because they may affect multiple organs and bring about comorbidities. Beyond the physical impact, PLWRD experience psychosocial consequences such as isolation, stigmatization, and discrimination.

The burden of rare diseases extends to families and caregivers, impacting society at large. Approximately 70-90% of rare diseases begin in childhood. Parents, especially mothers, often assume caregiver responsibilities, with negative consequences on their physical, psychosocial, and financial well-being. Workplace productivity loss for PLWRD and caregivers contributes significantly to the economic burden of rare diseases³⁻⁴.

¹ Available online: <https://www.rarediseasesinternational.org/wp-content/uploads/2022/08/INFOGRAPHIC-OPERATIONAL-DESCRIPTION-OF-RARE-DISEASES.pdf>

² Tisdale A, Cutillo CM, Nathan R, et al. The IDeaS initiative: pilot study to assess the impact of rare diseases on patients and healthcare systems. *Orphanet Journal of Rare Diseases* 2021; 16: 429.

³ Angelis A, Tordrup D, Kanavos P. Socio-economic burden of rare diseases: A systematic review of cost of illness evidence. *Health Policy* 2015; 119: 964–79.

⁴ EveryLife Foundation for Rare Diseases. *The National Economic Burden of Rare Disease Study*. 2021.

What are the challenges?

The diagnostic journey

PLWRD face distinct and significant challenges arising from the infrequency of their medical conditions. Due to the rarity of each individual disease and the wide dispersal of PLWRD across the globe, expertise and information are limited. PLWRD navigate an uncertain diagnostic journey because clinicians and healthcare systems often lack the awareness and experience to recognize rare conditions. From the very beginning, screening programs for asymptomatic individuals – such as newborn screening – are key, as the care and treatment of many rare diseases is more efficient when initiated as early as possible. Such screening programs have an important impact on patients' prognosis, but their availability varies widely from region to region. Even once an individual starts exhibiting symptoms of a rare disease, they often remain undiagnosed for a long time or may experience misdiagnoses, resulting in inappropriate treatment or irreversible disease progression. PLWRD wait up to five years on average before receiving a correct diagnosis for their condition⁵.

Even with a diagnosis, the clinical management of patients is often inadequate. Clinical experts on a given disease are often few and are dispersed around the world. The lack of knowledge sharing and exchange platforms makes it difficult for non-experts to consult experts on how to best care for their patients. Without sufficient knowledge about a rare disease, clinicians cannot assess its complexity, anticipate the disease's course, or standardize health management practices.

95% of rare diseases do not currently have an available treatment

The current trend of R&D investment into rare disease is significant and includes discovery, technologies, treatments and advanced therapies, systems and service delivery. However, this investment and any subsequent implementation are increasing inequity across the globe and there is still a lack of interest in research and development and a lack of financial incentives to develop drugs for the small and dispersed population of PLWRD. Additionally, conducting rigorous clinical trials poses methodological and ethical challenges for rare disease drugs. As a result, more than 95% of rare diseases do not currently have an available treatment⁶.

Access to treatment

The current cost of most treatment remains, and will continue to remain, out of reach for PLWRD, even in countries with strong healthcare systems. When treatment and care are available, high pricing often results in delayed, inconsistent, and inequitable access, and the benefits of the treatment are often uncertain due to limited clinical evidence. In Europe alone, the average cost burden of rare diseases was estimated at €107,000 per patient per year (PPPY), approximately 15 times greater than the average cost burden of high-prevalence

⁵ Available online: <https://www.eurordis.org/wp-content/uploads/2024/05/Diagnosis-printer-.pdf>

⁶ The Lancet Diabetes (2019). Spotlight on rare diseases. The Lancet Diabetes & Endocrinology, 7(2), 75. [https://doi.org/10.1016/S2213-8587\(19\)30006-3](https://doi.org/10.1016/S2213-8587(19)30006-3)

diseases, estimated at €7,000 PPPY⁷. While this is a direct cost of rare diseases, there are also indirect costs incurred by PLWRD, their families and society. The loss of productivity for both patient and caregiver has a detrimental impact not only on the family but also on the economy as a whole.

Access to care

In the absence of treatment, PLWRD often face challenges in accessing primary care services which can be limited and inadequately equipped to cater to the needs of rare disease patients. Studies provide evidence on the socioeconomic burden on families, communities, health systems and countries quantifying over the life course, the cost of none or limited care and treatment options. This research clearly underscores the return of investment in society achieved by increasing investment in preventative, early and holistic care for rare conditions⁴⁻⁶. There is an urgent need for healthcare systems to build on the experience gained over the last 20 years as well as to develop and implement innovative solutions for equitable and improved access to care and support for PLWRD.

Stigmatization

The challenges faced by PLWRD go far beyond health-related barriers. They experience stigmatization and inequities in other aspects of their lives, such as difficult inclusion and integration in mainstream education systems, discrimination and social exclusion, and a greater risk of impoverishment. This results in a significant social and financial burden for PLWRD and their families and caregivers, healthcare systems, and society overall, calling for the need to advocate for their cause and take actions to address their unique yet critical challenges. Recognizing the breadth of rare diseases across the world and their impact on physical and mental health means that social care systems are key to prioritizing services and improving outcomes⁸.

How can we catalyze national and local impacts by putting rare diseases on the global health agenda?

Each country and region faces unique challenges in addressing the needs of PLWRD in their community. However, all communities also face common challenges, such as restricted health budgets and limited availability of specialized services, resources, and knowledge. This results in patients around the world often struggling to access appropriate care and support.

Progress in improving the lives of PLWRD is being made across the globe at the regional and national levels; however, the wide range of challenges faced by PLWRD cannot be addressed equitably by a single actor. Collective action through a **collaborative and multisectoral approach** is the key to ensuring that the needs of patients and their families are met in

⁷ Andreu, P., John Atay N., Piccinini, E., Chiesi, G., & Cioffi, G. (2023). Rare disease burden of care and the economic impact on citizens in Germany, France and Italy. Chiesi USA. <https://chiesirarediseases.com/assets/pdf/rare-disease-burden-of-care-and-the-economic-impact-on-citizens.pdf>

⁸ Tisdale A, Cuttillo CM, Nathan R, et al. The IDEaS initiative: pilot study to assess the impact of rare diseases on patients and healthcare systems. Orphanet Journal of Rare Diseases 2021; 16: 429.

every part of the world. Making rare diseases a priority on the global health agenda will help ensure that PLWRD are included in crucial efforts toward achieving UHC for all.

Why a resolution on rare diseases for WHA 78?

By creating an international focus on rare diseases with a resolution at the WHA, the global health community can better integrate rare diseases into other key health issues such as primary healthcare, child health, non-communicable diseases etc. to leverage the progress made in addressing these issues in order to also benefit the rare disease community. A WHA resolution will also help mobilize support for greater investment in rare diseases and facilitate the pooling of resources and expertise, so that Member States and other stakeholders can maximize their impact in addressing the diverse needs of PLWRD and their families. Overall, by strengthening UHC for PLWRD not just nationally and regionally, but also globally, health stakeholders can solidify rare diseases as an international priority on the global health agenda and ensure a healthier future for rare disease patients worldwide.

Adopted in 2015, the WHO Sustainable Development Goal (SDG) 3.8 is to “Achieve UHC, including financial risk protection, access to quality essential healthcare services and access to safe, effective, quality and affordable essential medicines and vaccines for all”. In 2019, UN Member States reaffirmed this commitment when they adopted the United Nations Political Declaration on UHC, which includes rare diseases (Resolution A/RES/74/2, 2019⁹). This declaration was renewed in September 2023 with the Political Declaration of the High-Level Meeting on UHC (Resolution A/RES/78/4, 2023¹⁰). For UHC to be truly universal and to accelerate the progress towards achieving SDG 3.8, health stakeholders around the world need to put rare diseases on the global health agenda.

In December 2021, a major turning point in the global political landscape was achieved when the UN General Assembly unanimously adopted the UN Resolution Addressing the Challenges of Persons Living with a Rare Disease and their Families (Resolution A/RES/76/132, 2022)¹¹. The 2021 UN Resolution paves the way for greater integration of rare diseases into the agenda and priorities of the UN system. **However, a health-specific framework for coordinated action with all stakeholders and Member States is needed in order to solidify commitment and translate these commitments into action to overcome the barriers to accessing diagnosis, treatment, and care for PLWRD.**

Endorsing a WHA resolution on rare diseases aligns with countries’ commitments to achieving UHC and promoting health equity for all citizens and supports the 13 and 14 General Programme of Work of WHO, which include rare diseases within their scope. Governments can work towards ensuring that PLWRD have access to affordable, effective, and comprehensive healthcare services, regardless of the rarity of their condition.

⁹ Available on-line : <https://documents.un.org/doc/undoc/gen/n19/311/84/pdf/n1931184.pdf?token=uy2wWNqXRmv9i666L9&fe=true>

¹⁰ Available on-line: <https://documents.un.org/doc/undoc/gen/n23/306/84/pdf/n2330684.pdf?token=KCr4mLjen72fSsriu9&fe=true>

¹¹ UN General Assembly (76th sess.: 2021-2022). Addressing the challenges of persons living with a rare disease and their families: resolution / adopted by the General Assembly, A/RES/76/132.

What would a Global Action Plan on Rare Diseases address?

Many countries and communities have made significant strides in addressing the challenges faced by PLWRD in their regions. However, progress has not been uniform across the globe. A Global Action Plan on Rare Diseases will act as a catalyst for systemic change and will provide Member States with a tangible framework for action - adapted according to the level of advancement of each Member State - to support the development of national policy and strategies addressing the social and financial burdens of treating PLWRD in a sustainable and inclusive way.

A comprehensive global action plan on rare diseases would provide a clear and detailed roadmap for all stakeholders, promoting interdisciplinary and intersectoral approaches. This plan would establish global targets and strategic goals, along with specific actions and steps to achieve them. The guiding principles of this plan would include a common reference point for identifying rare diseases and a commitment to people-centered primary health care and universal health coverage. It would advocate for an integrated approach to care throughout an individual's life and ensure that policies and practices are informed by the best available evidence.

Moreover, the plan would outline a framework for intersectoral action, emphasizing the empowerment and involvement of PLWRD and their caregivers. To ensure the effectiveness of this global action plan, a robust process for accountability and monitoring would be established, allowing for the tracking of implementation progress. Additionally, a dedicated budget for the WHO would be allocated to support member states in executing the plan, ensuring that resources are available to facilitate these critical actions.

Conclusion

By adopting a WHA resolution on rare diseases, Member States have the opportunity to strengthen their positions as leaders in healthcare innovation and advocacy while reaffirming their commitment to leaving no one behind in the ultimate pursuit of health for all and to contribute their respective expertise and resources to improve the lives of PLWRD not only nationally, but globally.

The WHA resolution, and ideally the resulting global action plan, will be the catalyst that urges Member States to work together towards enhancing policy processes, advancing research, accelerating innovation, and improving access to care for PLWRD worldwide. This will ultimately ensure that PLWRD are included in the critical work being done to advance UHC across the globe.

About Rare Diseases International

[Rare Diseases International](#) (RDI) is the global alliance of PLWRD of all nationalities across all rare diseases, representing more than 100 member organizations across the 6 continents. RDI's mission is to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent its members and enhance their capacities.

RDI's main goal is to address inequities faced by PLWRD and to shape international policy on rare diseases, strengthening health systems to address rare diseases, and advancing Universal Health Coverage. RDI was successful in having rare diseases identified in the 2019 UN Declaration on Universal Health Coverage and again in the 2023 renewal. In 2021, RDI successfully advocated for the adoption of a United Nations General Assembly (UNGA) Resolution on Rare Diseases. RDI has had a Memorandum of Understanding with the WHO since 2019 and became a Non-State Actor in official relation with the WHO in 2024.