Why Invest in Rare Diseases?

Ensuring UHC for Persons Living with a Rare Disease

Side-Event co-hosted by Rare Diseases International and France and Malaysia

Wednesday, 29 May 2024
18:00 – 21:00 (CET) - Restaurant Vieux Bois, Geneva

Background

On the occasion of the 77th World Health Assembly (WHA), Rare Diseases International, in collaboration with France and Malaysia will host a side event, titled "Why Invest in Rare Diseases?", on May 29, 2024, from 6:00 pm to 9:00 pm at the Vieux-Bois Restaurant in Geneva.

The event aims to underline the critical importance of investing in Rare Diseases to ensure greater equity for all Persons Living with a Rare Disease (PLWRD), contribute to reaching the Global Health 2035 Goals, align with the WHO Global Programme of Work 14 and make Universal Health Coverage (UHC) a reality.

The purpose of our event goes beyond merely discussing the importance of investing in rare diseases; it serves as a critical platform for advocating for a World Health Assembly resolution on rare diseases in 2025. Securing international commitment and a dedicated resolution on rare diseases at the World Health Assembly is essential to prioritizing rare diseases on the global health agenda and improving access to diagnosis, treatment, and support services for affected individuals worldwide.

Member States have a unique opportunity to be leaders in addressing an existing unmet need in healthcare by shedding light on the challenges faced by the more than 300 million PLWRD and proposing solutions for a better ecosystem for rare diseases. This can lead to significant returns on investment in science and technology, policy guided by principles of equity and empowerment, the strengthening of health systems for increased access to diagnosis and care, and improved resources for the social systems that support PLWRD and their families.

Despite considerable progress, including the recognition of rare diseases within the United Nations (UN) 2030 Agenda for Sustainable Development, which seeks to promote health equity by addressing the needs of the most vulnerable populations, PLWRD continues to face substantial challenges globally, such as delays in diagnosis,

difficulties in accessing treatments and services, social stigma, and financial burdens. Currently, 6,000+ rare diseases have been named and described. Of these 6000 diseases, the 400 most prevalent affect an estimated 90% of the population living with a rare disease; there is a very long tail of extremely rare diseases.[1-2]

In 2019, UN Member States adopted the United Nations Political Declaration on UHC, which includes rare diseases (Resolution A/RES/74/2, 2019). This declaration was renewed in September 2023 with the Political Declaration of the High-Level Meeting on UHC (Resolution A/RES/78/4, 2023).[3]

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). The 2021 UN Resolution paves the way for greater integration of rare diseases into the agenda and priorities of the UN system. However, a framework for coordinated action with all stakeholders and Member States is needed to translate these commitments into action and overcome the barriers to accessing diagnosis, treatment, and care for PLWRD to ensure no one is left behind in the pursuit of UHC.

The vast challenges of treating PLWRD cannot be resolved by a single country or entity. They can only be addressed by a global, multidisciplinary, and collaborative approach. “Leaving no one behind” can become a reality if WHO, in line with the 13 and 14 General Programme of Work, along with Member States and relevant stakeholders, adopts a resolution at the 78th World Health Assembly. This will secure rare diseases as a priority on the global health agenda and emphasize to Member States the importance of addressing the social and financial burdens of treating PLWRD in a sustainable and inclusive way so that people with rare diseases are not left behind in the work on UHC.

Event Objective and Target Audience

The objective of the event is to engage Ministers of Health, WHO representatives and rare diseases organizations in a discussion on why it is critical to invest in rare diseases. The event also aims to explore how to address an existing unmet need in healthcare by calling attention to the challenges faced by PLWRD and their families

[1] Nguengang Wakap, S., Lambert, D. M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., ... & Rath, A. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. European Journal of Human Genetics, 28(2), 165-173. https://doi.org/10.1038/s41431-019-0508-0
[2] Haendel, M., Vasilevsky, N., Unni, D., Bologa, C., Harris, N., Rehm, H., ... & Oprea, T. I. (2020). How many rare diseases are there?. Nature reviews drug discovery, 19(2), 77-78. https://doi.org/10.1038/d41573-019-00180-y
[3] 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.

and showcasing successful initiatives developed worldwide, and to emphasize the importance of a WHA resolution in 2025.

The following stakeholders form the target audience for the event:

- Representatives of Geneva permanent missions and Ministry of Health
- Policy Makers
- Global health organizations
- Private sector
- Representatives of RDI members who are participating in WHA 77
- Patient Advocates

Agenda

Time	Topic
6:00pm-6:10pm	Welcoming the participants on-site
6:10pm-6:15pm	Opening Remarks: Durhane Wong Rieger – Chair, Rare Diseases International Welcome and Introduction and Setting the Context: Importance of Universal Health Coverage (UHC) for PLWRD
6:15pm-6:35pm	Keynote Addresses introduced by Alexandra Heumber Perry, CEO of RDI - Dr Ruediger Krech, Director of Health Promotion, World Health Organization (WHO) - Antoine Saint-Denis, Director for European and International Affairs, French Ministry for Labour, Health and Solidarities - Datuk Dr Muhammad Radzi Abu Hassan, Director General of Health of Malaysia - Dr.Mohamed Hassany, Assistant Minister for Public Health Initiatives, Egyptian Ministry of Health
6:35pm-7:00pm	**Panel Discussion 1: Global Policy and UHC for PLWRD** Moderator: Alexandra Heumber Perry – CEO, Rare Diseases International Panellists: Dr Ruediger Krech, World Health Organization (WHO) Antoine Gliksohn, Global Alliance for Albinism Dr Androulla Eleftheriou, Thalassemia International Federation Anne-Sophie Chalandon, IFPMA Rare Diseases Working Group

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7:00pm-7:30pm	**Panel Discussion 2: Regional and National Approaches and Collaborative Solutions** Moderator: Durhane Wong Rieger, Chair of RDI Council Panellists: Dr Roberto Giugliani, Casa Hunter, Brazil Nadiah Hanim Abdul Latif, Malaysian Rare Disorders Society Trudy Nyakambangwe, Child and Youth Care Zimbabwe Yann Le Cam, EURORDIS- Rare Diseases Europe Juan Carrión, ALIBER and FEDER
7:30pm-7:35pm	Closing Remarks: Alexandra Heumber Perry – CEO, Rare Diseases International Call to Action: A WHA Resolution on Rare Diseases
7:35pm-9:00pm	Networking Cocktail Reception

RARE DISEASES INTERNATIONAL

Fact Sheet

Why Invest in Rare Diseases?

WORLDWIDE, THERE ARE

300 MILLION

PEOPLE LIVING WITH A RARE DISEASE

approximately

5% of the population will be diagnosed with a rare disease over their lifetime

OVER **7,000**

RARE DISEASES HAVE BEEN IDENTIFIED

These conditions can be chronic, progressive, degenerative, disabling and frequently life threatening. The 149 most frequent of these diseases account for 80% of rare disease diagnoses.

85% of RARE DISEASES are extremely rare (<1/1,000,000)

People living with an ultra-rare disease face even more barriers to diagnosis, treatment and care due to the low frequency of their conditions

72% of RARE DISEASES are genetic

70% of these diseases start in childhood

95% of RARE DISEASES have no available treatment

73% of PLWRD have been MISDIAGNOSED at least once

On average, people living with a rare disease in HICs wait **6 years** for a diagnosis

Diagnosis takes even longer in the case of ultra-rare diseases and in LMICs.

The financial burden of RARE DISEASES is **15 times higher** than that of common diseases

In addition to direct costs, PLWRD face indirect costs such as loss of income and barriers to education and employment due to stigma. The task of caring for family members with rare diseases falls most often upon women, amplifying gender inequity.

RARE DISEASES INTERNATIONAL

Fact Sheet

Ensuring UHC for PLWRD: Shift the Paradigm

Make Rare Diseases a HEALTH PRIORITY

We are few. We are many.
There are over 300 million Persons Living with a Rare Disease (PLWRD) around the world. To ensure that they receive the services they need, **we need to prioritise and integrate rare diseases within regional and national UHC and primary health care plans.**

Improve SERVICES for Persons Living with a Rare Disease

Persons Living with a Rare Disease have complex needs, beginning with the diagnostic journey, which can last 5-8 years - or in some cases, continue indefinitely. By strengthening primary healthcare and referral pathway capacities, we can ensure the timely diagnosis of rare diseases. After diagnosis, PLWRD need services that encompass a multidisciplinary approach. **By developing sustainable programs dedicated specifically to rare diseases, we can enable rapid and equitable access to diagnosis, treatment and social support.**

Reduce the FINANCIAL BURDEN of Rare Diseases

Persons living with a Rare Disease face financial burdens due to the high cost of treatments and medications. They must often travel long distances to access specialized care. PLWRD and their families also face high indirect costs, such as loss of income. Women are disporportionately affected, often taking responsibility for caring for a family member with a rare disease. **By investing in rare diseases, we can lower costs in the long term and improve equity for PLWRD and their families.**

Enhance KNOWLEDGE SHARING on Rare Diseases

International collaboration is key to improving treatment and knowledge of rare diseases. Ethical and responsible international data sharing can support diagnosis, increase clinical collaboration, facilitate research and accelerate treatment of undiagnosed and rare conditions. By increasing education and awareness of rare diseases, we can reduce the stigma and discrimination faced by PLWRD.

The vast challenges of treating PLWRD cannot be resolved by a single country or entity. They can only be addressed by a global, multidisciplinary, and collaborative approach.
A WHA Resolution: a catalyst for systemic and sustainable change to improve the lives of Persons Living with a Rare Disease.