World Health Organization

Executive Board
156th session

Agenda item 6 EB156(15)

10 February 2025

Rare diseases: a global health priority for equity and inclusion

The Executive Board, having considered the report of the Director-General,[1]

Decided to recommend to the Seventy-eighth World Health Assembly the adoption of the following resolution:

The Seventy-eighth World Health Assembly,

Having considered the report by the Director-General;

Recognizing that a rare disease is often described as a specific health condition affecting 1 in 2000 individuals or fewer in general population, and that there are currently over 7000 known rare diseases impacting more than 300 million people globally, with 70% of these conditions starting in childhood;[2] and that, while the frequency of most rare diseases can be described by prevalence, some rare diseases can be more precisely described by incidence;[3]

Noting that rare diseases are often complex and multisystemic, affecting multiple organs and leading to comorbidities, and that many of these conditions are chronic, progressive and can consequently result in serious disabilities and premature death;

Recognizing that some persons living with a rare disease have disabilities, which may have a greater impact on their health, and that they may also face various barriers, which may hinder their full and effective participation in society on an equal basis with others;[4]

Recognizing also that, in addition to the physical impact, some persons living with a rare disease, their families and caregivers may experience discrimination and psychosocial consequences such as isolation, stigmatization and limited opportunities for social inclusion, which are often intensified by a lack of public awareness and knowledge, and the absence, limited scope or poor implementation of policies and social support;

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[1] Document EB156/6.

[2] The Lancet Global Health. The landscape for rare diseases in 2024. Editorial. 10.1016/S2214-109X(24)00056-1.

[3] Wang et al. Orphanet Journal of Rare Diseases. Operational description of rare diseases: a reference to improve the recognition and visibility of rare diseases (2024), pp. 19–334.

[4] Resolution WHA74.8 (2021).

Recognizing further that persons living with a rare disease (including those whose disease is undiagnosed), their families and caregivers may be psychologically, socially and economically vulnerable throughout their life course, facing specific challenges in several areas, including but not limited to physical and mental health, education, employment, financial well-being and leisure;

Emphasizing the importance of adopting a holistic patient-centred approach to address the needs of persons living with a rare disease, focusing on enhancing their functioning and working with society to remove, to the extent possible, the barriers they face in accessing health, education, employment and other domains of life;

Noting that the high prices of many health products for rare diseases, and inequitable access to such products within and among countries, as well as the financial hardships associated with their high costs, pose significant challenges for some persons living with a rare disease;

Recognizing from an equity perspective that women and children living with a rare disease encounter greater challenges in accessing care, including the late diagnosis, biases in symptom assessment, and reduced access to timely and appropriate treatment, which significantly impact their quality of life and overall health outcomes;

Recognizing also the importance of achieving universal health coverage, including for persons living with a rare disease and their families and caregivers, and that universal health coverage implies that all people have access, without discrimination, to nationally determined sets of essential quality health services, from health promotion to prevention, treatment, rehabilitation and palliative care, as well as essential, safe, affordable, effective and quality medicines, vaccines, diagnostics and health technologies, including assistive technologies, ensuring that the cost of using these services does not lead to financial hardship;

Recognizing further the importance of implementing integrated care, considering the health system along with social and community services, for enabling persons living with a rare disease to achieve optimal health and well-being;

Acknowledging that to enhance physical and mental health, well-being and life expectancy for everyone, it is essential to achieve universal health coverage, including persons living with a rare disease;

Recalling in particular the United Nations Sustainable Development Goal target 3.8 (Achieve universal health coverage, including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all), the United Nations political declaration of the high level meeting on universal health coverage (2019),[5] which includes rare diseases, and the political declaration of the high-level meeting on universal health coverage (2023),[6] reaffirming the commitment to ensure that no one is left behind, and other universally agreed resolutions and declarations;

[5] United Nations General Assembly resolution 74/2 (2019).

[6] United Nations General Assembly resolution 78/4 (2023).

Recalling also United Nations General Assembly resolution 76/132 (2021) on addressing the challenges of persons living with a rare disease and their families, which paved the way for greater integration of rare diseases into the agenda and priorities of the United Nations system;

Noting that reaching the correct diagnosis can take over five years, that many persons living with a rare disease never receive a timely or adequate diagnosis, although nearly half of genetic diseases start in childhood, and that insufficient screening programmes, including newborn screening, and unequal access to diagnostic services, infrastructure and expertise contribute to delayed diagnosis and management;

Noting also that for undiagnosed persons with a suspected rare disease, entering a coordinated diagnostic and research pipeline offers a unique hope to speed up diagnosis, as recognized by the International Rare Diseases Research Consortium;

Recalling resolution WHA76.5 (2023) on strengthening diagnostics capacity, which recognizes that diagnostic services are vital for the prevention, diagnosis, case management, monitoring and treatment of communicable, noncommunicable, neglected tropical and rare diseases, and which emphasizes equitable access to diagnostics for all, and highlights the importance of diagnostics for healthcare service delivery, ranging from prevention to treatment, as well as access to research projects on diagnostics;

Recalling also resolution WHA75.8 (2022) on strengthening clinical trials to provide high-quality evidence on health interventions and to improve research quality and coordination, in which the Health Assembly called on Member States, inter alia, “to encourage the targeting of clinical trials towards the development of health interventions that address public health priorities and concerns of global, regional and national importance, including communicable and noncommunicable diseases, with a focus on the health needs of developing countries, and that evaluate the safety and efficacy of health interventions, including having special regard to common diseases in low- and middle-income countries, unmet medical needs, rare diseases and neglected tropical diseases”;

Recalling further resolution WHA77.2 (2024) on social participation for universal health coverage, health and well-being, in which the Health Assembly urged Member States, inter alia, to strive “to ensure that social participation influences transparent decision-making for health across the policy cycle, at all levels of the system”;

Recalling resolution WHA77.5 (2024), in which the Health Assembly invited Member States, inter alia, to consider implementing a universal newborn screening programme, including comprehensive screening for congenital disorders; and recognizing the importance of early detection programmes, including those for prevention and mitigation of health conditions that may result in disabilities, while also addressing the specific needs and considerations for diagnosis, management, and long-term care that meets the needs of affected children;

Recognizing that early identification can prevent the onset of disease symptoms or delay the progression of both common and rare diseases, thereby reducing child mortality and morbidity, improving the quality of life of persons living with a rare disease and conferring significant benefits on them, their families, their caregivers and society as a whole;

Acknowledging the disparity of resources between rural and urban areas within and among countries, the limited availability and geographical dispersion of rare disease specialists and centres of expertise, along with the lack of patient pathways, referral systems and effective knowledge-sharing platforms, which hinders necessary consultations with specialists on diagnosis and optimal patient care, thereby resulting in suboptimal clinical management for persons living with a rare disease;

Noting that due, in part, to limited resources for research, diagnosis and treatment along with the insufficient equitable investment and financial incentives for drug development in rare diseases, more than 95% of rare diseases still lack an effective treatment;

Acknowledging that even when treatments and care are available, high costs may often lead to delayed, inconsistent and inequitable access;

Acknowledging also that rare diseases fall within the scope of the WHO's Fourteenth General Programme of Work, 2025–2028, as well as the WHO's efforts to achieve the goals outlined in its first strategic priority of extending universal health coverage to one billion more people as stated in the WHO's Thirteenth General Programme of Work, 2019–2025, and in alignment with countries' national context and priorities;

Acknowledging further that although each country, in line with its national context and priorities, faces unique challenges in meeting the needs of persons living with a rare disease, there are common issues, such as constrained health budgets and a shortage of specialized services, resources and expertise leading to health inequities within and among Member States, which collectively result in persons living with a rare disease worldwide often struggling to access the care and support they need;

Highlighting the WHO's commitment to promote health equity and support Member States in ensuring that all persons living with a rare disease, regardless of their condition, receive timely and appropriate healthcare services;

Emphasizing the critical need for global collaboration to tackle the unique challenges faced by persons living with a rare disease, and by their families and caregivers, especially mothers – including: the implementation of policies and programmes that prevent and combat stigma and social exclusion; accurate data collection; and increased awareness – in line with countries' national context and priorities;

Recognizing the need to foster innovation that promotes social cohesion and reduces inequalities and discrimination, and to enhance research efforts and develop innovative therapies for rare diseases;

Underscoring the need to address the root causes of inequality and discrimination faced by persons living with a rare disease, their families and caregivers, and in this regard recognizing that there is a need for health policies and programmes to foster inclusion and create an environment conducive to respect for their rights and dignity;

Noting that rare diseases may lead to disabilities, and in this regard, recalling the principles embodied in the Constitution of the World Health Organization and the Convention on the Rights of Persons with Disabilities, and stressing the importance of their implementation, including inter alia through relevant policies, programmes and strategies at

the national and international levels to promote inclusion and rights of persons with such disabilities;

Recalling United Nations General Assembly resolution 78/12 (2023) entitled "World Duchenne Awareness Day",[7] in which the General Assembly decided to designate 7 September, the current World Duchenne Awareness Day, as a United Nations Day, recognizing that Duchenne muscular dystrophy is one of the most common paediatric genetic rare diseases, and encouraging Member States to raise awareness on the specific challenges and needs faced by persons living with a rare disease, their families and caregivers through national campaigns, educational programmes and information dissemination, with the goal of fostering greater understanding and empathy towards those affected by rare diseases and promoting global solidarity,

1. URGES Member States,[8] taking into account national context and priorities:

(1) to commit:

(a) to providing appropriate support to WHO in developing a comprehensive global action plan on rare diseases;

(b) to integrating rare diseases into national health planning by developing and implementing national policies, effective programmes and actions, including developing primary and secondary evidence-based preventive actions and strategies aimed at preventing and improving healthcare services for persons living with a rare disease through an integrated approach, ensuring equitable access to timely, cost-effective and affordable, available, accurate diagnosis, particularly for newborns through universal screening programmes, and the necessary cost-effective treatment, social and healthcare services;

(c) to implementing effective programmes that promote mental health and psychosocial support for persons living with a rare disease, as well as policies and initiatives that enhance the well-being of their families and caregivers;

(d) to accelerating efforts toward achieving and extending universal health coverage by 2030, ensuring healthy lives and well-being for all individuals, including persons living with a rare disease, throughout their life course, in order to stop the rise and reverse the trend of catastrophic out-of-pocket health expenditure as appropriate, by re-emphasizing the commitment to progressively provide persons living with a rare disease with quality essential health products, healthcare services, and affordable medicines, diagnostics and health technologies by 2030;

(e) to strengthening health systems, particularly in primary healthcare, to ensure universal access to a wide range of affordable and high-quality healthcare services for persons living with a rare disease, especially children;

[7] United Nations General Assembly resolution 78/12 (2023).

[8] And, where applicable, regional economic integration organizations.

(f) to fostering the inclusion of relevant competencies in the pre-service education of students and lifelong learning of health workers in preventing, diagnosing, treating and managing rare diseases;

(g) to further increasing awareness and education initiatives about rare diseases among healthcare providers, policy-makers and the public in order to promote understanding of and support for affected individuals;

(h) to removing barriers that persons living with a rare disease, their families and caregivers face in accessing safe water, sanitation and hygiene, including addressing physical, institutional, social and attitudinal obstacles, promoting appropriate measures to ensure equitable access for these individuals, their families and caregivers in both rural and urban areas;

(i) to considering, as appropriate, the development and utilization of digital technologies,[9] including telemedicine and data-sharing platforms in order to improve access to specialists and treatments, especially in remote areas or those with limited medical resources, ensuring that technologies are accessible;

(j) to promoting the involvement of patient organizations, peer support groups, organizations of persons with disabilities, including groups led by persons living with a rare disease, in policy development to ensure that the voices of those affected by rare diseases are heard and incorporated into decision-making processes;

(k) to facilitating the establishment, as appropriate, of dedicated national task forces or coordination bodies to oversee the implementation of policies related to rare diseases, enhancing accountability and effective management;

(l) to encouraging the establishment of national, regional and international centres of excellence as specialized hubs for care, research and training for rare diseases;

(m) to encouraging the establishment of a national registry for rare diseases, or collaborating with existing international registries for rare diseases, as appropriate, to strengthen their capacity on data collection, analysis and disseminating disaggregated data on persons living with a rare disease, while respecting data protection and privacy, to achieve evidence-based decisions at all levels;

(n) to considering implementation of the eleventh revision of the International Classification of Diseases (ICD-11), and where appropriate, interoperable codification systems for rare diseases such as the Orphanet nomenclature of rare diseases, at their earliest possibility, and in accordance with their available resources, in order to enable the recording, reporting and monitoring of rare diseases at the national and international levels;

[9] Guided by, inter alia, WHO's global strategy on digital health 2020–2025.

(2) to encourage collaboration between policy-makers, governmental health and research authorities, academic institutions, clinicians, patient organizations, the private sector and civil society in order to foster innovation in research and innovative diagnosis and treatment that proactively address rare diseases;

(3) to support efforts to adopt innovative ways of funding and mobilize resources from all sources (for example, public and private funders) for integrated action on rare diseases, including research and innovation, and to consider expanding opportunities, with a focus on developing countries;

(4) to strengthen cooperation at the national, regional and international levels to promote equitable and timely access to affordable, safe, effective and quality medicines for all persons living with a rare disease across the world, leaving no one behind;

(5) to bring high-level attention to rare diseases and related aspects within multilateral forums, as appropriate, to help ensure sustained and concrete political visibility and momentum, and explore ways in which to integrate rare diseases into health policy and programmes reflecting the national strategies and priorities;

(6) to regularly assess, where applicable, the implementation of their national action plans for rare diseases and, to the extent possible, evaluate their contribution to the implementation of regional action plans related to rare diseases;

2. REQUESTS the Director-General:

(1) to develop – in consultation with Member States, and in collaboration with nongovernmental organizations including patients' organizations, academic institutions, in line with the Framework of Engagement with Non-State Actors, as applicable, and experts in rare diseases – a comprehensive 10-year draft global action plan for rare diseases, in alignment with the agreed strategic priorities of WHO and its Fourteenth General Programme of Work, 2025–2028, including all necessary preparatory work, and budgetary aspects, to be submitted for consideration by the Eighty-first World Health Assembly in 2028;

(2) to conduct preparatory work, including: mapping existing WHO standards, guidelines and protocols relating to rare diseases; providing an initial technical report on rare diseases; identifying technological innovation opportunities (including e-health, m-health, digital and artificial intelligence solutions) to centralize clinical health information for diagnostics and treatment;

(3) to establish a workstream promoting universal health coverage for persons living with a rare disease;

(4) to identify centres of excellence around the world that are able to cluster clinical work in certain rare disease groups and that can act as hubs to exchange experience and clinical knowledge and provide peer-to-peer medical reviews and advice, including across borders;

(5) to ensure that the global action plan for rare diseases encompasses, but is not limited to, the following key components:

- a comprehensive framework to foster equitable access to timely, cost-effective, affordable, available, accurate diagnosis and evidence-based treatments, and an adequate management of rare diseases, aligned with the principles of universal health coverage as outlined in the United Nations political declarations of the high-level meetings on universal health coverage of 2019 and 2023, and taking into account the social determinants of health;

- strategies for improving data collection, research and surveillance on rare diseases to enhance understanding, timely and confirmed early identification, including screening, diagnosis and treatment options in collaboration with Member States' national authorities, with the ultimate goal of sharing knowledge and data in the field and fostering investment in research;

- guidelines for the establishment of national and regional registries to facilitate the screening, monitoring and management of rare diseases;

- global targets and strategic objectives, along with clear guidelines to improve access to affordable and equitable healthcare services for persons living with a rare disease, the essential health products needed for accurate diagnosis and effective treatment for persons living with a rare disease, as well as an accompanying process for accountability and monitoring to track implementation progress, including at the national level;

(6) to support Member States, upon request, in the development of national policy and strategies to enhance the health of persons living with a rare disease, including addressing the social and financial implications of supporting persons living with a rare disease in a sustainable and inclusive way;

(7) to submit a draft global action plan on rare diseases for consideration by the Executive Board at its 162nd session, with the intention of submitting this draft global action plan to the Eighty-first World Health Assembly for adoption;

(8) to report on the implementation of this resolution to the Seventy-ninth World Health Assembly in 2026, through the Executive Board at its 158th session, and to submit progress reports to the Health Assembly in 2028 and 2030.

Eighteenth meeting, 10 February 2025
EB156/SR/18
