Rare Diseases: A Global Health Priority for Equity and Inclusion

The Draft resolution proposed by the Arab Republic of Egypt and Spain, and co-sponsored by Brazil, Chile, China, Ecuador, France, India, Iraq, Jordan, Kuwait, Luxemburg, Malaysia, Panama, Palestine, Pakistan, Philippines, Qatar, Romania, Somalia and Vanuatu

The Executive Board
Recommends to the Seventy-eighth World Health Assembly the adoption of the following resolution

(PP1) *Recognizing* that a rare disease is often described as a specific health condition affecting 1 in 2,000 individuals or fewer in general population, and that there are currently over 7,000 known rare diseases impacting more than 300 million people globally, with 70% of these conditions starting in childhood[1], While the frequency of most rare diseases can be described by prevalence, some rare diseases can be more precisely described by incidence[2];

(PP2) *Noting* also that rare diseases are often complex and multi-systemic, affecting multiple organs and leading to comorbidities, and that many of these conditions are chronic, progressive, and consequently can result in serious disabilities and premature death;

(PP3) *Recognizing* that some persons living with a rare disease (PLWRD) 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[3];

(PP4) *Recognizing* also that, in addition to the physical impact, some PLWRD, 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, knowledge, and the absence, limited scope or poor implementation of policies and social support;

(PP5) *Recognizing* also that PLWRD (including undiagnosed PLWRD), 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;

(PP5 bis) *Emphasizing* the importance of adopting a holistic patient-centered approach to address the needs of PLWRD, 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;

(PP5ter) *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 PLWRD;

(PP6) *Recognizing* also 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;

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[1] The Lancet Global Health. The landscape for rare diseases in 2024. Editorial. 10.1016/S2214-109X(24)00056-1.
[2] Wang et al. Orphanet Journal of Rare Diseases. Operational description of rare diseases (2024), pp. 19-334
[3] WHO Resolution WHA 74.8 on The highest attainable standard of health for persons with disabilities.

(PP7) *Recognizing* also the importance of achieving universal health coverage (UHC), including for PLWRD, their families, and caregivers, and that UHC 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, health technologies, including assistive technologies, ensuring that the cost of using these services does not lead to financial hardship;

(PP7 bis) *Recognizing* also the importance of implementing integrated care, considering the health system along with social and community services, for enabling PLWRD to achieve optimal health and well-being;

(PP8) *Acknowledging* that to enhance physical and mental health, well-being, and life expectancy for everyone, it is essential to achieve UHC, including PLWRD;

(PP9) *Recalling* in particular the UN Sustainable Development Goal (SDG) 3.8 "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", the United Nations Political Declaration of the first high level meeting on UHC (A/RES/74/2, 2019)[4] which includes rare diseases, and the Political Declaration of the second High-Level Meeting on UHC (A/RES/78/4, 2023)[5] reaffirming the commitment to ensure that no one is left behind, and other universally agreed resolutions and declarations;

(PP10) *Recalling* also the UN Resolution (A/RES/76/132, 2021)[6] "Addressing the Challenges of Persons Living with a Rare Disease and their Families", adopted in December 2021 by the UN General Assembly, which paved the way for greater integration of rare diseases into the agenda and priorities of the UN system;

(PP11) *Noting* that reaching the correct diagnosis can take over five years, that many PLWRD never receive a timely or adequate diagnosis, though 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;

(PP11bis) *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 diagnostic, as recognized by the International Rare Disease Research Consortium;

(PP12) *Recalling* also Resolution (WHA76.5, 2023) "Strengthening diagnostics capacity" which recognizes that diagnostic services are vital for the prevention, diagnosis, case management, monitoring and treatment of communicable, non-communicable, 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;

(PP13) *Recalling* also Resolution (WHA75.8, 2022) "Strengthening clinical trials to provide high-quality evidence on health interventions and to improve research quality and coordination" calling on Member States "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 non-communicable 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,

[4] Available on-line: https://documents.un.org/doc/undoc/gen/n19/311/84/pdf/n1931184.pdf
[5] Available on-line: https://documents.un.org/doc/undoc/gen/n23/306/84/pdf/n2330684.pdf
[6] 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.

rare diseases and neglected tropical diseases";

(PP13bis) *Recalling* also Resolution (WHA77.2, 2024) "Social participation for universal health coverage, health and well-being" calling on Member States "to strive to ensure that social participation influences transparent decision-making for health across the policy cycle, at all levels of the system";

(PP14): *Recalling* also Resolution (WHA77.5, 2024) which invites Member States to consider implementing a universal newborn screening programs, including comprehensive screening for congenital disorders; and, recognizing the importance of early detection programs including 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;

(PP14 bis) *Recognizing* also 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 PLWRD deriving significant benefits to them, their families, their caregivers and society as a whole;

(PP15) *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 centers 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 PLWRD;

(PP16) *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[1];

(PP17) *Acknowledging* that even when treatments and care are available, high costs may often lead to delayed, inconsistent, and inequitable access;

(PP18) *Acknowledging* also that rare diseases fall within the scope of the WHO's fourteenth General Programme of Work (GPW14), as well as the WHO's efforts to achieve the goals outlined in its first strategic priority of extending UHC to one billion more people as stated in the WHO's thirteen General Programme of Work (GPW13), and in alignment with countries' national context and priorities;

(PP19) *Acknowledging* also that although each country, in line with its national context and priorities, faces unique challenges in meeting the needs of PLWRD, 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 states, which collectively result in PLWRD worldwide often struggling to access the care and support they need;

(PP20) *Highlighting* the WHO's commitment to promote health equity and support Member States in ensuring that all PLWRD, regardless of their condition, receive timely and appropriate healthcare services;

(PP21) *Emphasizing* the critical need for global collaboration to tackle the unique challenges faced by PLWRD, their families and caregivers, especially by mothers, including the implementation of policies and programmes that prevent and combat stigma and social exclusion, accurate data collection, increased awareness and in line with countries' national context and priorities;

(PP22) *Recognizing* the need to foster innovation that promotes social cohesion, reduces inequalities and discrimination, and the need to enhance research efforts and develop innovative therapies for rare diseases;

(PP23) *Underscoring* the need to address the root causes of inequality and discrimination faced by PLWRD, their families, and caregivers, and in this regard recognizing that there is a need for health policies and programs to foster inclusion and to create an environment conducive to respect for their rights and dignity;

(PP24) *Noting* that rare diseases may lead to disabilities, and in this regard recalling the principles embodied in the WHO Constitution and the Convention on the Rights of Persons with Disabilities and stressing the importance of their implementation, including inter alia through relevant policies, programs and strategies at national and international levels to promote inclusion and rights of persons with such disabilities;

(PP25) *Recalling* also the UN Resolution (A/78/L.12, 2023)[7] "World Duchenne Awareness Day", which designate 7 September as the World Duchenne Awareness Day, and recognizing that Duchenne muscular dystrophy is one of the most common pediatric genetic rare diseases, and encourage the raising awareness on the specific challenges and needs faced by PLWRD, 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.

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

(OP1.1) to commit:

(a) To provide appropriate support to the WHO in developing a comprehensive Global Action Plan on Rare Diseases;

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

(c) To implement effective programs that promote mental health and psychosocial support for PLWRD, as well as policies and initiatives that enhance the well-being of their families and caregivers;

(d) To accelerate efforts toward achieving and extending UHC by 2030, ensuring healthy lives and well-being for all individuals including PLWRD 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 PLWRD with quality essential health products, healthcare services, and affordable medicines, diagnostics, and health technologies by 2030;

(e) To strengthen health systems, particularly in primary health care, to ensure universal access to a wide range of affordable and high-quality healthcare services for PLWRD especially children;

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

(g) To further increase awareness and education initiatives about rare diseases among healthcare providers, policymakers, and the public to promote understanding and support for affected individuals;

[7] https://documents.un.org/doc/undoc/gen/n23/376/83/pdf/n2337683.pdf
[8] and where applicable, regional economic integration organizations

(h) To remove barriers that PLWRD, 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 consider, as appropriate, the development and utilization of digital technologies[9], including telemedicine and data-sharing platforms to improve access to specialists and treatments, especially in remote or limited medical resources areas ensuring technologies are accessible;

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

(k) To facilitate 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 encourage the establishment of national, regional and international Centers of Excellence as specialized hubs for care, research and training for rare diseases;

(m) To encourage the establishment of a national registry for rare diseases, or collaborate with already existing international registries for rare diseases, as deemed appropriate, to strengthen their capacity on data collection, analysis, and disseminate disaggregated data on PLWRD, while respecting data protection and privacy, to achieve evidence-based decisions at all levels; and

(n) To consider implementation of ICD-11, and where appropriate, interoperable codification systems for rare diseases such as ORPHA codes, at their earliest possibility, and in accordance to their available resources, in order to enable the recording, reporting and monitoring of rare diseases at national and international levels.

(OP1.2) To encourage collaboration between policy-makers, governmental health and research authorities, academic institutions, clinicians, patient organizations, private sector, and civil society in order to foster innovation in research and innovative diagnosis and treatment that proactively address rare diseases;

(OP1.3) To support efforts to adopt innovative ways of funding, mobilize resources from all sources (e.g. public, private funders) for integrated action on rare diseases including research and innovation, and consider expanding opportunities, with a focus on developing countries;

(OP1.3 bis) 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 PLWRD across the world, leaving no one behind;

(OP1.4) To mobilize high-level attention to rare diseases and related aspects within multilateral fora, 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 programs reflecting the national strategies and priorities;

(OP1.5) 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.

(OP)2. REQUESTS the Director-General:

[9] Guided by, among others, the WHO Global Strategy on Digital Health

(OP2.1) To develop in consultation with Member States, and in collaboration with nongovernmental organizations including patients' organizations, academic institutions, in line with FENSA, as applicable, and experts in rare diseases, a comprehensive 10-year Global Action Plan for Rare Diseases, and in alignment with the agreed-upon strategic priorities of the WHO and its GPW14, including all necessary preparatory work, and budgetary aspects, to be submitted to the 81st World Health Assembly in 2028.

(OP2.1bis) To conduct preparatory work, including mapping of 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 AI solutions) to centralize clinical health information for diagnostics and treatment;

(OP2.1ter) To establish a workstream promoting UHC for PLWRD;

(OP2.1 quater) To identify centers of excellence around the world that are able to cluster clinical work in certain rare disease groups which can act as hubs to exchange experience and clinical knowledge and provide peer-to-peer medical reviews and advice, including across borders;

(OP2.1 quinquies) The Global Action Plan may encompass the following key components, but not limited to:

- 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 UHC as outlined in the 2019 and 2023 UNGA Political Declarations 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 to share knowledge and data in the field and fostering the 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 PLWRD, the essential health products needed for accurate diagnosis and effective treatment for PLWRD, as well as an accompanying process for accountability and monitoring to track implementation progress, also at the national level;

(OP2.2) To support Member States, upon request, in the development of national policy and strategies to enhance the health of PLWRD, including addressing the social and financial implications of supporting PLWRD in a sustainable and inclusive way;

(OP2.3) To submit a draft Global Action Plan for consideration by the Executive Board at its 162nd session, with the intention of submitting this Global Action Plan to the 81st World Health Assembly session for adoption.

(OP2.4) To report on the implementation of this resolution to the Seventy-Ninth (79th) World Health Assembly in 2026 through the Executive Board at its 158th session, and to submit progress reports to the World Health Assembly in 2028, and 2030.