

VERONICA  
LOPEZ GOUSSET

MATT  
BOLZ-JOHNSON

GLOBAL NEEDS ASSESSMENT STUDY EXTRACT

# COMMON NEEDS OF THE RARE DISEASE POPULATION IDENTIFIED GLOBALLY

OCTOBER 2021



RARE  
DISEASES  
INTERNATIONAL

# Common Needs of the Rare Disease Population Identified Globally

By Veronica Lopez Gousset, RDI Programme Manager and Matt Bolz-Johnson, RDI Programme Director

Through conversations with patient representatives and medical experts across the six World Health Organization (WHO) regions, members of the Rare Diseases International (RDI) team identified and prioritised ten common areas of need impacting the diagnosis, care, and treatment of the rare disease community globally. This work was done as part of a more extensive global Population Needs Assessment Study, conducted by RDI to substantiate the need and propose a framework for developing a Collaborative Global Network for Rare Disease.

This report examines the top ten common areas of need identified. These areas are all interrelated and generally stem from a lack of awareness and understanding of rare diseases at different levels of society, including policymakers, healthcare providers, and the broader community. Increased awareness of the burden that rare diseases represent for individuals, families, and healthcare systems leads to dedicated policy frameworks, specialised budgets and workforce, a focus on availability and coverage of specialised care, and community empowerment, among other elements. While these needs are common across all regions, there is variability in political support, healthcare system structure, and rare disease communities, resulting in these needs impacting different populations and diseases to different extents. Nevertheless, increasing awareness results in more significant commitment and ultimately more concrete action at the local level to benefit persons living with a rare disease (PLWRD) and their families.

## Methodology

An exploration of the global rare disease population needs was performed by collecting first-hand accounts of leading patient representatives and medical experts from multiple countries within each WHO region. As proxies for their populations, it is estimated that these individuals represented 83.4% of the global rare disease population. These interviews focused on capturing patients' needs, identifying population-specific considerations, and mapping the maturity and characteristics of healthcare systems for rare diseases. The experiences and needs reported were documented, and content analysis was undertaken through a qualitative review of the insights to determine key concepts, themes, and trends. The results were then validated, enhanced, and prioritized through Focus Groups organized by WHO region as follows (1) African region, (2a) North & Central America sub-region, (2b) South America sub-region, (3) East Mediterranean region, (4) European region, (5) South East Asia region, (6a) Western Pacific WPR A and (6b) Western Pacific WPR B.



Overall, patient representatives and medical experts from around the world participated in this evaluation. The findings of this exercise are summarized in this report.

## Results

The following ten areas were identified as the top cross-cutting needs across all rare disease populations globally. Prioritization of these needs varied by country and by rare disease community depending on the robustness of healthcare systems, the specific needs of the rare condition, and the maturity of the advocacy community.

1. Social and cultural acceptance, equality, and inclusion of Persons Living with a Rare Disease (PLWRD)
2. Systematic, standardised data collection and data sharing
3. Political recognition and dedicated policy framework and budget for rare diseases
4. Availability, affordability, and coverage of rare disease tests and medicines
5. Focus on prevention and screening
6. Widespread availability of expertise, specialised services, and standards of care
7. Coordination of care across devolved or fragmented healthcare systems
8. Geographical and cultural considerations
9. Supporting technology infrastructure and use of telemedicine
10. Empowerment of patients and families to self-care and advocate

### **1. Social and cultural acceptance, equality, and inclusion of PLWRD**

PLWRD and their families are exposed to social and cultural stigma and experience discrimination and exclusion in society due to a lack of awareness and understanding. At the health system level, this translates into the exclusion of affected individuals from care and lack of coverage of essential services, medicines, and treatments. At the community level, the physical appearance of some PLWRD and those with disorders impacting behaviour result in discrimination and the isolation of affected individuals and their families. Some rare diseases, including genetically linked diseases, can present a social taboo and carry a stigma that affects individuals' access to care and inclusion in society.

It is critically important to address the stigma associated with rare diseases and help families access the care and psychological support they need. Rare diseases affect all aspects of an individual's life, including education, employment, community inclusion, and participation. Reducing exclusion from society is a human rights issue that urgently needs to be addressed for this vulnerable community.

### **2. Systematic, standardised data collection and data sharing**

There is a lack of patient data and epidemiologic studies to understand the prevalence and incidence of rare diseases, thereby preventing governments from accurately assessing the significant hidden burden represented by rare diseases and congenital disorders.

Data are required to support advocacy efforts and enable evidence-based decision-making. Data highlight the true burden of rare diseases in each region and lead to relevant actions (such as developing new-born screening programmes and identifying expert centres) and accompanying resource allocations.

Given the rarity of these conditions, genetic databases and cross-system, cross-border data sharing are also essential to better understanding and diagnosing rare diseases. Registries are beneficial in helping to gather relevant data.

### **3. Political recognition and dedicated policy framework and budget for rare diseases**

While the European region and certain other countries (for example, Australia) have successfully developed rare disease policies, they are the exception and not the rule. Most countries and regions neither have nor are developing an officially approved dedicated policy framework for rare diseases (Africa, parts of the Americas, East Mediterranean, South East Asia, and some parts of Western Pacific and Europe). Where policies do exist, they are often not supported by a dedicated budget to enable proper implementation.

The lack of adequate political recognition, prioritisation, and support for rare diseases directly results in a lack of dedicated funding. It ultimately impacts these vulnerable communities' access to healthcare and the availability of medicines.

### **4. Availability, affordability, and coverage of rare disease tests and medicines in health systems**

Access to affordable diagnosis, care, and treatment was reported as one of the highest priorities for all patient communities, irrespective of location. Access to diagnostic services depends on the availability of genetic tests for screening and diagnosis. Genetic tests vary significantly in price (compounded by the lack of trained geneticist workforces) and Health Technology Assessment (HTA) processes that do not always apply specific considerations needed for value assessment in rare diseases.

Access to care depends on individual insurance schemes or geographical location. The biggest challenge is the degree of inclusion (or exclusion) of rare diseases in health insurance systems. In some countries, health insurance coverage is minimal or restricted (meaning it excludes rare diseases), leading to substantial financial burdens for PLWRD and their families. Lack of a national policy and recognised expert centres results in families forced to access specialists from the private sector, which is frequently not covered by public health insurance. Accessing specialists and expert centres can therefore generate significant expenses that families must pay out-of-pocket. In some regions, families are forced to choose between accessing treatment or feeding their families, particularly in certain countries in the African, East Mediterranean, and South East Asia regions.

Significant health inequalities within countries (between provinces and states) and across all the WHO regions make it difficult for PLWRD to access the health system equitably. Even in sub-regions supported by political commitment, for instance, in the Arabian Gulf, refugees, migrant workers, and foreign residents still face challenges in accessing diagnosis, care, and treatment. In the European Union, where citizens can theoretically access cross-border care, there remain significant administrative and financial barriers that prevent patients from exercising their rights to access cross-border care.

Lack of policy and legislation also impacts the availability of orphan medicines. Even in regions with a centralised designation of medicines, registration can be delayed many years due to the system's lack of readiness to include these medicines into reimbursement schemes. Small populations (markets) are particularly affected where companies lack incentives to register medicines, reducing the availability of drugs, devices, and consumables that meet international care standards. It is essential to work with all stakeholders - HTA agencies, regulators, companies, and patient groups - to develop access pathways to lifesaving, transformative, and potentially curative medicines. Certain countries

are leading the way in making potentially transformative medicines available (China, Czech Republic, Germany, USA, etc.).

## **5. Focus on prevention and screening**

Screening is a crucial method for reducing time to diagnosis. Programmes such as pre-marital, prenatal, and new-born screening, should be expanded in certain regions to reduce the incidence and eliminate the diagnostic odyssey and its associated financial burden on the health system. Perceptions and prejudices associated with genetic diagnosis (e.g., societal exclusion, impact on insurance coverage) can lead many patients to avoid seeking genetic testing. Community and political awareness-raising are essential for reducing these barriers to accessing accurate diagnosis.

Health promotion and education measures could significantly reduce the prevalence of rare diseases, for example, in areas with high consanguinity rates, such as in the East Mediterranean region.

## **6. Widespread availability of expertise, specialised services, and standards of care**

The availability of trained specialists is a core requirement for vulnerable communities to secure culturally appropriate care and support. Lack of specialists has been reported as a common barrier in all regions, especially geneticists and genetic counsellors (Africa, East Mediterranean, South East Asia). Lack of awareness and training of primary care physicians in rare diseases has also been highlighted as a barrier in accessing care for PLWRD and families. Physicians who are untrained and not sensitised to rare diseases in their medical curriculum or during induction into practice often overlook and miss ‘red flag’ symptoms and vital signs that could facilitate early diagnosis.

Even when it exists, access to expertise can be a challenge for PLWRD if specialists and centres are not formally recognised by the government nor sufficiently mapped in all regions. This scenario typically translates into specialist advice and treatment not covered by the public health insurance schemes or needing to be provided by the private sector. A vital goal of any rare disease policy is to centralise care and knowledge in expert centres, thereby increasing the experience and expertise of these centres, which ultimately improves the outcome for patients. However, even in countries with a national policy in place, not all provide for the centralisation of care; local autonomous regions and devolved budgets weaken or even undermine patients’ possibilities to access safe and effective care despite being approved by the national authorities.

In addition, the lack of national guidelines and treatment protocols - including those for essential medicines, medical devices, and life-altering drug therapies - can impact vulnerable populations. Lack of standardized care can directly lead to an increase in co-morbidities and a decrease in quality of life (e.g., access to prophylaxis treatment for rare blood disorders), leading to an increased burden on healthcare resources. International standards of care would help to promote and enhance the push towards internationally agreed-upon best practices. Earlier diagnosis, care, and treatment prescribed in line with evidence-based guidelines have been demonstrated to be less costly and more clinically effective, resulting in better outcomes for patients and decreases in all subsequent costs.

## **7. Coordination of care across devolved or fragmented healthcare systems**

Fragmentation of the healthcare system and the lack of centralised expertise and care were common barriers reported across regions. While healthcare is provided on a local level, rare diseases, given their low prevalence, need a national or even regional/international approach and strategy. Systems

that are devolved can therefore hinder and fragment care and follow-up for rare disease populations. Fragmentation can be due to devolved budgets between provinces or states (Australia, Italy, India, Spain, United States, etc.); lack of medical knowledge and adapted facilities or patients not knowing how to reach care (South East Asia); or once well-resourced healthcare systems damaged or destroyed due to current or past conflicts (Afghanistan, Iraq, Syria, Yemen). Political issues in some regions have also been reported to impact the access to essential services of the most vulnerable populations directly and indirectly (Iran).

There is also a reported lack of holistic, person-centred, and multi-disciplinary care across all regions. This could be caused by several factors: poor communication and information between health professionals and patients; a lack of coordination of care between centres and local services; poor transition from paediatric to adult care; or a lack of specialised clinical centres and medical knowledge (Africa, Europe, South East Asia, Western Pacific). Inadequate referral systems have also been reported as barriers, especially for rural communities, to accessing care (Africa and East Mediterranean), resulting in patients being lost to follow-up.

## **8. Geographical and cultural considerations**

A significant proportion of the world's population lives in rural communities, particularly in countries with vast geographical territories (Argentina, Brazil, China, India, Russian Federation, United States, and Saudi Arabia), resulting in physical barriers to accessing care services. Those who live in rural areas often have the most disadvantages, both in terms of access and referrals to expert centres or specialists. Travel burden, security, and costs create difficulties for patients. It can take a long time for PLWRD to access care when the major hospitals are located primarily in urban areas. This also contributes to prolonging the diagnosis odyssey.

The rural-city divide becomes more pronounced when there are little pockets of expertise, increasing the barriers for vulnerable persons accessing care and diagnosis. Even in cities and countries with a high level of specialist centres, insufficient knowledge and understanding of health professionals in frontline services can significantly impact people seeking to secure a timely and accurate diagnosis.

Cultural appropriateness is also essential, as ethical and practical considerations need to be observed when providing care to specific populations. It has been reported that barriers to accessing care and treatment can significantly increase for rare diseases and disproportionately affect women and minorities in some regions.

## **9. Supporting technology infrastructures and use of telemedicine**

Lack of investment in technologies to assist service delivery (South East Asia, Western Pacific) or the underutilization and recognition of telemedicine and assistive technologies (Africa, Americas, East Mediterranean, Europe, South East Asia, Western Pacific) are also barriers to care, particularly for rural communities.

Online consultation is not a widely available option across all regions as the technology is new to many countries and people. However, in some areas, telehealth has already changed the current practice of providing and monitoring care (Argentina, Chile, Israel). Cultural distance and demographic considerations also play a role as not all communities embrace the increased use of technology, e.g., in indigenous populations. Technology should be leveraged to bridge geographical and cultural distances without increasing health inequalities due to a widening digital divide.

## **10. Empowerment of patients and families to self-care and advocate**

Health literacy can be low in some regions, with many people unaware of rare diseases, leading to families having multiple children with the same condition. Prenatal counselling, new-born screening, and awareness amongst the general population can be very low in certain regions. Healthcare can be highly dependent on families' and patients' health literacy and the maturity of patient advocacy groups to support families in navigating complex healthcare systems and connecting with the right expert centre.

Education and support for families to learn how to self-care are essential, particularly in rural communities where expertise is less accessible. Trust must be built among doctors in the ability of parents and patients to self-care and advocate for appropriate care. In this context, patient groups are critical in supporting their communities in accessing care.

## Conclusion

While certain needs are more relevant based on each country's unique political, socio-economic situation and the complexities of each rare condition, this report presents a consolidated view of the areas of highest need as identified by the rare disease patient and medical communities globally. Addressing these needs begins with greater education and awareness at all levels of society and leads to appropriate access to early diagnostic and treatment and holistic healthcare and social services for the estimated 263-446 million<sup>i</sup> PLWRD and their families around the world.

---

<sup>i</sup> Wakap S, Lambert D, Olry A, Rodwell C, Gueydan C, Lanneau V, Murphy D, Le Cam Y, Rath A. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* 2020 Feb;28(2):165-173. doi: 10.1038/s41431-019-0508-0. Epub 2019 Sep 16. PMID: 31527858; PMCID: PMC6974615.





## 10 CROSS-CUTTING NEEDS

1. Social acceptance, equality, and inclusion
2. Systematic and standardised data collection and data sharing
3. Political recognition and policy frameworks and resources
4. Availability, affordability, and coverage of diagnostic tests, treatment and care
5. Focus on prevention and screening
6. Availability of expertise, specialised services, and standards of care
7. Coordination of care across healthcare systems
8. Greater geographical and cultural consideration
9. Technology infrastructure and use of telemedicine
10. Empowerment of patients and families for self-care and advocate