

RARE DISEASES INTERNATIONAL

RESOURCE MAPS

*A resource to understand the
global rare disease landscape*



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READER'S NOTE

Throughout 2022 and 2023, members of the Rare Diseases International (RDI) team recorded first-hand accounts about the rare disease landscape in various countries around the world, through conversations with patient representatives and medical experts across the six World Health Organization (WHO) regions.

These resource maps aim to collate information gathered to-date in an easy-to-access snapshot of the regional and national situation for rare diseases across the WHO regions. As such, these resource maps are living documents that will continue to be updated, strengthened, and refined with the help of our community. **If you see any information that is not accurate for your region, please raise this with our team and together we will address it.**

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We would like to extend our special thanks to all the patient leads, medical experts, and international federations who shared their knowledge and experiences allowing us to create these resource maps for our community.

INTRODUCTION

Understanding and strengthening existing networks and collaborations is essential to improve healthcare practices and support for persons living with a rare disease (PLWRD). This document serves as a resource map outlining the rare disease landscape by country. This includes valuable insights into the presence of patient organizations actively advocating for rare disease communities and existing political interventions in place to support these. This guide also details the existing networks and centers at both the national and regional levels, as well as current collaborations and referral systems for rare diseases.

Countries with available information have been grouped based on the World Health Organization (WHO) regional categorization¹ and listed alphabetically. The information is further broken down to highlight particularities of each political, patient advocacy and healthcare system within each country with relation to rare diseases. While some information might still be in progress or incomplete, this resource serves as a crucial starting point for understanding the current landscape of rare disease efforts across the globe.



By bringing to light the diverse range of national and regional networks, as well as encouraging collaborations and sharing knowledge, this resource aims to foster relationships and progress in the field of rare diseases.

	Population size (2020) In Millions ²	Estimated rare disease population prevalence (3.5-5.9%) in Millions ³
1. African Region	1,036.1	35.8 - 60.4
2. Region of the Americas	1,018.1	35.6 - 60.1
3. East Mediterranean Region	730.8	25.6 - 43.1
4. European Region	932.9	32.7 - 55
5. South-East Asia Region	2,021.4	70.8 - 119.3
6. Western Pacific Region	1,939.2	66.6 - 114.4
Total	7,678.5M	267.1M – 452.3M

¹ <https://datahelpdesk.worldbank.org/knowledgebase/articles/906519-world-bank-country-and-lending-groups>

² United Nations, Population Division. The World Population Prospects - the 2019 revision. New York, 2019.

³ Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0>

AFRICAN REGION (AFR)



Countries: Total of 47 Member States

Member States

Algeria, Angola, Benin, **Botswana**, **Burkina Faso**, Burundi, Cameroon, Cape Verde, Central African Republic, Chad, Comoros, Congo, **Côte d'Ivoire**, Democratic Republic of Congo, Equatorial Guinea, Eritrea, Ethiopia, Gabon, Gambia, **Ghana**, Guinea, Guinea-Bissau, **Kenya**, Lesotho, Liberia, Madagascar, Malawi, Mali, Mauritania, Mauritius, **Mozambique**, Namibia, Niger, **Nigeria**, Rwanda, Sao Tome and Principe, Senegal, Seychelles, Sierra Leone, **South Africa**, South Sudan, Swaziland, **Uganda**, United Republic of Tanzania, Togo, Zambia, **Zimbabwe**.

* Note: Countries in **BOLD** are those represented within this Resource Map.⁴

⁴ Image created with mapchart.net

AFR Rare Disease Landscape Overview

As reported by patient leaders and medical experts from thirteen countries in the region

Rare Disease Policy Landscape

- While rare diseases are increasingly becoming recognized across the sub-Saharan African region, governments are prioritizing and developing services to tackle competing health priorities, such as non-communicable diseases, overshadowing the needs of rare and congenital diseases.
- In some African countries, efforts towards developing universal health coverage (UHC) are advancing, e.g., a national health insurance fund exists in Kenya, Ghana, and South Africa. However, lack of rare disease data, policy, definition, NS legislation provide little leverage to call for improved health service and support investment in actions to tackle rare diseases in the region.

Rare Disease Health System Landscape

- While there is a growing awareness of the concept of rare diseases and a growing number of experts and specialists in many African countries, the general lack of public awareness around rare diseases leaves affected individuals exposed to cultural and social stigma.
- Diagnosis is one of the biggest barriers to care in Africa. Limited healthcare expertise, shortage of essential equipment and supplies, and inadequate infrastructure to diagnose congenital disorders and rare diseases result in many patients remaining undiagnosed or misdiagnosed.
- Pockets of expertise and specialists exist within the main academic teaching hospitals (e.g., South Africa, Ghana). However, most expertise and services are concentrated in capital or main cities and urban areas. With the majority of the populations living in rural areas with little specialized services, low income, poor infrastructure, and travel costs are common barriers for people to access care and treatment.
- Where public healthcare services are available (e.g., Botswana, Ghana and South Africa), there is access to general healthcare, under the developing UHC initiatives, but treatments for rare diseases are excluded from the “essential health package”. In countries with dual systems, only a low proportion of the population has private health insurance (e.g., 10% of the population in Kenya, 16% in South Africa), resulting in the majority of the population relying on inadequate state services or paying out-of-pocket for treatment.

Rare Disease Patient Organization Landscape

- Patient groups established in the region include national alliances for rare diseases, for example in South Africa, Ghana, Botswana, and Kenya. Patient support and advocacy groups play a critical role in coordinating and supporting families in accessing specialized services.
- National groups are collaborating across the region to address challenges associated with rare diseases under the Africa Rare Disease Alliance.

REGIONAL ALLIANCE

Africa Rare Disease Alliance (ARDA)

BOTSWANA

Rare Disease Policy Landscape

- In Botswana, there is lack of political awareness and support for rare diseases as a health priority. Access to general physical care and therapies is provided under the “essential health package” but there is nothing specific for rare diseases.

Rare Disease Health System Landscape

- Diagnosis is usually undertaken outside of Botswana and Africa. Even when a diagnosis is secured, patients cannot access support or treatments as there is a lack of funding. Healthcare workers often do not know how to manage the care of this population. This results in many people not seeing the benefit of being diagnosed.
- Nonetheless, the concept of rare diseases has started to emerge. The main academic teaching hospital in the capital has some expertise in rare diseases. This hospital is collaborating with other centers outside the country to improve diagnosis and care.
- Most of the population lives in rural areas resulting in barriers to access hospitals within bigger cities.

CENTERS

Princess Marina Hospital (Gaborone)

Rare Disease Patient Organization Landscape

NATIONAL GROUP

Botswana Organization for Rare Diseases (BORDIS)

BURKINA FASO

Rare Disease Policy Landscape

- There is no structured, state-supported organization for rare diseases.

Rare Disease Health System Landscape

- The private sector that is organized to care for rare diseases according to its capacity.
- There is an overall lack of health professionals who can detect rare diseases and refer patients; a lack of adequate medical and paramedical materials for examinations and treatments; and healthcare infrastructure is situated in unsuitable buildings.
- High costs of examinations and treatments is a significant barrier for patients to access care.
- Nevertheless, university hospitals are improving technical platforms to allow better diagnostic investigations and there is a training school for physiotherapists, social workers and psychologists to work towards comprehensive care.

CENTERS


Centre Hospitalier Universitaire Pédiatrique Charles De Gaulle (Ouagadougou)

Rare Disease Patient Organization Landscape



- The Fondation Internationale Tierno et Mariam, a nonprofit organization focused on human rights protection of children and women, intervenes in Burkina Faso as well as Guinea.

CÔTE d'IVOIRE

Rare Disease Policy Landscape

-  The Ministry of Health has not taken health measures to support people living with rare diseases.



Rare Disease Health System Landscape

-  The majority of this community does not have access to care due to the high cost of consultations and examinations.
-  There is a lack of rare disease centers and medical training on specific rare diseases. Expertise is based in Abidjan making it difficult for rural populations to access the hospitals. When hospital care is accessed, there is limited follow up after a diagnosis due to the lack of rare disease expertise.

CENTERS

Centre Hospitalier Universitaire de Yopougon (Abidjan)

Rare Disease Patient Organization Landscape

-  Patient groups work to increase awareness of rare diseases, provide training to doctors, and support free consultations for affected individuals. For example, patient support groups organised a conference with medical professionals on the impact of rare diseases and the opportunities for patients and their families to learn more about rare diseases.
-  Aux Pas du Coeur is a patient organization committed to raising awareness, help patients get diagnosed, and encourage doctors and the Ministry of Health to take an interest in rare neuromuscular diseases.

GHANA



Rare Disease Policy Landscape

- There is no rare disease policy, which affects the funding and reimbursement of care. However, there are criteria and procedures for designating and defining orphan drugs. A definition of rare diseases exists but no prevalence is specified.⁵

Rare Disease Health System Landscape

- The government provides most healthcare services; however, rare diseases are not included in the national health insurance system and there are no dedicated clinics for rare diseases. As rare diseases are not covered by national health insurance, families are required to finance the gap in coverage by paying for private care. Private and traditional (spiritual) medicine is available and accessed by a large portion of the population.
- The healthcare system has 5 levels of providers: primary care for rural areas, health centres and clinics, district hospitals, regional hospitals, and tertiary hospitals. Most of the care and expertise is concentrated in the capital, whereas most people in Ghana's 30 million population live in rural areas.
- Depending on disease type, there are some specialists who can manage care of the diseases, usually based in teaching hospitals. There are four tertiary hospitals with a fifth hospital being developed. All tertiary hospitals are connected to universities.
- Rare diseases are increasingly being recognized and health professionals are interested in learning more about these conditions.

CENTERS

-  Komfo Anokye Teaching Hospital (Kumasi)
-  Korle Bu Teaching Hospital (Accra)

Rare Disease Patient Organization Landscape

- Patient groups are active in supporting families coordinate care and connect them to specialized services around the country.

NATIONAL GROUP


Rare Diseases Ghana Initiative

⁵ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020 <https://doi.org/10.1016/j.jval.2020.06.020>

KENYA

Rare Disease Policy Landscape

- The government committed to Universal Health Coverage which is expected to raise standards of care in Kenya. However, the primary focus is infectious diseases and non-communicable diseases (NCDs)
- The country has a definition for rare diseases, a definition for orphan drugs and an official list of orphan drugs⁶

 RD Definition:
Less than 50,000



Rare Disease Health System Landscape

- Healthcare is reportedly fragmented with limited resources and high medical costs. There is a dual insurance system of public and private insurance. While the national insurance fund covers all diseases, the insurance is capped which is leading to an increasing use of private insurance to access healthcare. Only a low proportion of the population has private health insurance (about 10% of the population in Kenya) resulting in the majority of the population relying on inadequate state services or paying out-of-pocket for treatment (80%).
- There is general lack of awareness and understanding amongst doctors, policy-makers and the general public as to the needs of rare diseases. While awareness is building, rare diseases continue to face social and cultural stigma. Families affected are perceived as “being punished,” so families seek care outside of healthcare system, which is difficult to track.

NETWORKS & CENTERS

- There are six referrals hospitals in the public sector and teaching hospitals in the private sector; however, referral systems are poor. In most centres, there is no specific coordination of follow-up care. The patient has to centralize the information from different specialist services.

CENTERS

-  Kenyatta National Hospital (Nairobi)
-  Moi University and Referral Hospital (Eldoret)

Rare Disease Patient Organization Landscape


- Patient groups are filling gaps in care and services, e.g., providing emotional support, supporting referrals, and offering education on specific conditions.

NATIONAL GROUP Rare
Disorders Kenya



⁶ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020 <https://doi.org/10.1016/j.jval.2020.06.020>

MOZAMBIQUE

Rare Disease Policy Landscape

-  The Ministry of Health is engaging with patient organizations and supporting changes to policies to improve access and quality of healthcare.


Rare Disease Health System Landscape

-  The national health system is reported to have limited capacities to diagnose and treat people living with rare diseases. Even when the referral structure might be in place, scarcity of human resources and essential medicines limits diagnosis and treatment capacities.
-  Information and public awareness about rare diseases is limited, which can impact on delays to securing a diagnosis.

CENTERS

Hemophilia Unit, Hospital Central de Maputo (Maputo)

Rare Disease Patient Organization Landscape

-  Patient groups are mobilizing to develop and connect patients and experts under a national network for rare diseases.

NIGERIA

Rare Disease Policy Landscape

- The country's health priorities are communicable diseases such as malaria, tuberculosis, HIV/AIDs. Non-communicable diseases and rare diseases lack funding, attention, and awareness.

Rare Disease Health System Landscape

- There is still wide misunderstanding and lack of knowledge of rare diseases. Resources and information on some rare disease is also not available in Nigeria and emotional and psychological support systems are limited.
- Medical care for rare disease in Nigeria fails to meet the need of patients and high costs are a barrier to seeking medical attention. There is a lack of expertise and awareness of rare diseases among medical personnel, a lack of standard tests for diagnosing a rare disease, and examination and tests are very expensive, leading to rare diseases often being misdiagnosed in Nigeria.
- Treatment options are limited or not available which adversely affects the outcomes and quality of life of patients.

CENTERS

- Aminu Kano Teaching Hospital (Kano)
- Lagos University Teaching Hospital (Lagos)
- Gombe State University (Gombe)
- National Hospital (Abuja)
- University of Nigeria Teaching Hospital (Ituku-Ozalla)
- University of Port Harcourt Teaching Hospital (Port Harcourt)
- University of Ibadan (Ibadan)

Rare Disease Patient Organization Landscape

- Patient groups are active in raising awareness of rare diseases through health awareness campaigns via online platforms and public outreach programs. Patient groups also provide emotional and physiological assistance and provide financial aid through fundraising for patients who cannot afford to pay for health services.

SOUTH AFRICA

Rare Disease Policy Landscape

- There is no current standard definition of rare diseases. A rare diseases policy framework is under development to level the contributions across private and public systems and provide some certainty in terms of access.

Rare Disease Health System Landscape

- Healthcare is fragmented across the country and not accessible universally. South Africa has a dual healthcare system, with about 16% of the population accessing services through a private insurance system, and 84% of the population accessing services in the public sector. The last few years have seen a reduction in private insurance benefits, as companies attempt to exert pressure on the public systems to do more for high-cost diseases.
- South Africa has Prescribed Minimum Benefits for insurance providers, which establishes a minimum basket of care (based on what is available in state services) that can be offered for a list of 270 conditions. However, the list is outdated and generally not inclusive of current treatments.
- While access to rare disease treatments is slowly improving in the country, one of the biggest barriers the rare disease patient community faces in accessing treatment is the inflexibility of pricing models for high cost drugs as there is no exemption for orphan medicines.

NETWORKS & CENTERS

- Throughout the country, there are pockets of expertise in rare diseases. There are several academic hospitals, usually supported by genetic centres, however their coverage does not extend to rural populations.

CENTERS

- Bloemfontein Adult Haemophilia Treatment Centre, University of the Free State (Bloemfontein)
- Groote Schuur Hospital, Department of Haematology (Cape Town)
- Johannesburg Hospital, Comprehensive Haemophilia Care Clinic (Johannesburg)
- Steve Biko Academic Hospital (Pretoria)
- Donald Gordan Hospital (Gauteng)
- Albert Luthuli Hospital (Durban)
- Red Cross Children's Hospital (Cape Town)
- University of Cape Town Lung Institute (Cape Town)

Rare Disease Patient Organization Landscape

NATIONAL GROUP

Rare Diseases South Africa

UGANDA

Rare Disease Policy Landscape

- Once a condition is identified as rare, it is integrated into the already existing system of care. The system allows for multidisciplinary care to support patients, although rare diseases are given less attention and are not a health priority.

Rare Disease Health System Landscape

- The healthcare system suffers from challenges such as lack of public awareness, mismanagement of care due to limited knowledge among health workers, and hard to reach patients missing appointments leading to complications and premature deaths due to delayed treatment.
- The regional referral hospitals face challenges in providing essential care, including the lack of supplies and facilities; medical teams are often overstrained and lack knowledge of rare diseases.
- The greatest needs for people living with rare diseases are dedicated treatment centers, diagnostic services, and availability of essential medicines.

CENTERS

Mulago Hospital (Kampala)

Rare Disease Patient Organization Landscape

- Patient groups collaborate under the Non-Communicable Diseases Department.
- Disease-specific groups in the region include Haemophilia Foundation of Uganda

ZIMBABWE

Rare Disease Policy Landscape

- There is no government support for individuals affected by a rare disease. Most of the country's health funding and investments are focused on communicable diseases. This results in rare conditions not being prioritized when it comes to diagnosis and care.

Rare Disease Health System Landscape

- Due to the economic state of the country, and lack of information around rare diseases, most people living with a rare disease are undiagnosed or misdiagnosed.
- Children with rare diseases do not have to pay for hospital care in government facilities, however, the governmental hospital system is currently crippled due to inadequate funding.
- Medical health professionals do not have the necessary skills to diagnose, treat, and care for rare disease patients. However, some patients get first preference during emergencies and some rare diseases have special hospital services focused on their care.

CENTERS

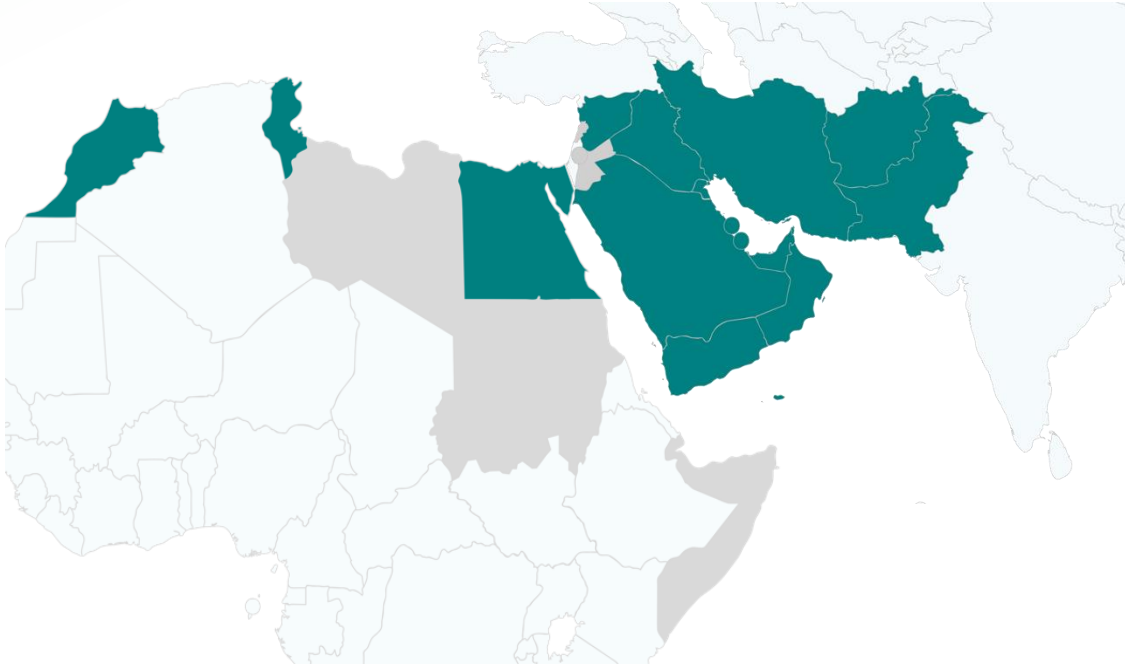
- Mpilo Central Hospital (Bulawayo)
- Parirenyatwa Group of Hospitals (Harare)

Rare Disease Patient Organization Landscape

NATIONAL GROUP

Child and Youth Care

EAST MEDITERRANEAN REGION (EMR)



Countries: Total of 23 Member States

Member States and Economic Areas

Afghanistan, Bahrain, Djibouti, Egypt, Iran (Islamic Republic of), Iraq, Jordan, Kuwait, Lebanon, Libya, Morocco, Oman, Pakistan, State of Palestine⁷, Qatar, Saudi Arabia, Somalia, Sudan, Syrian Arab Republic, Tunisia, United Arab Emirates, Yemen.

⁴Note: Countries in **BOLD** are those represented within this Resource Map.

⁷ United Nations recognised the State of Palestine in 2019

⁸ Image created with mapchart.net

EMR Rare Disease Landscape Overview

As reported by patient leaders and medical experts from eighteen countries in the region

Rare Disease Policy Landscape

- Across the region, rare diseases compete with other health priorities, such as chronic diseases, and are not recognized as a health priority in North Africa, the Levant or Arabian Gulf regions.
- The Gulf Cooperation Council (GCC), a Council of GCC Health Ministers coordinate joint health action in the Gulf States. The GCC charter sets the rights of GCC citizens to have equal access to national healthcare and treatment services in all Member States. In addition, the GCC member States are cooperating to obtain competitive and reasonable prices and supplies of medicines.

Rare Disease Health System Landscape

- In the North Africa region, healthcare for the more prevalent rare diseases, such as hemophilia, is reported to be good. There are centers of expertise in certain countries for hemophilia and thalassemia (Egypt, Morocco, and Tunisia). Experts collaborate internationally.
- In Levant region, the political changes, sanctions, instabilities, and conflicts, have impacted the availability of basic healthcare. This is especially true in post-conflict areas, where healthcare services were once well resourced and organized but have since been destroyed or significantly damaged, such as in Iraq, Afghanistan, and Syria.
- The Arabian Gulf States generally have well-resourced healthcare services, available therapies, and are investing in the development of genomic programs (Saudi Arabia, Oman, Qatar, Kuwait, UAE). Rare disease healthcare within the Arabian Gulf is mainly accessed through government-backed free or subsidized public hospitals. Experts in the region collaborate internationally.
- The Arabian population has one of the highest rates of genetic diseases in the world. Consanguineous marriages have been associated with increased likelihood of birth defects of autosomal recessive disorders.⁹ Pre-marital genetic screening is a standard offer in many Arabian Gulf States (UAE, Bahrain, Oman and Saudi Arabia). In Iran, genetic counselling services are well developed and available under the public health insurance and private clinics.

Rare Disease Patient Organization Landscape

- Patient groups are variable in number and organization across the East Mediterranean region.
- In the more affluent countries, well-developed public services and available therapies may be associated with lower levels of civic action. Nevertheless, patient communities are becoming more active in advocating for vulnerable groups (Kuwait, Saudi Arabia, and UAE).
- The Thalassemia International Federation and World Hemophilia Federation have member groups in most countries in the region. These Federations support national patient group members and local populations by providing humanitarian aid and purchasing therapies.

⁹ Heidari F, Dastgiri S, Tajaddini N, et al. Prevalence and Risk Factors of Consanguineous Marriage. European Journal of General Medicine [serial online]. December 2014;11(4):248-255. Available from: Academic Search Complete, Ipswich, MA. Accessed October 4, 2016.

AFGHANISTAN

Rare Disease Policy Landscape

- The WHO and UN agencies are helping the Afghan government implement the National Health Policy 2015-20 and Strategy 2016-20. In spite of these efforts, a significant amount of people continues to live with an undiagnosed rare disease and treatments for some rare diseases are difficult to access.
- Afghanistan has a definition for orphan drug and an official list of orphan drugs.¹⁰

Rare Disease Health System Landscape

- Conflict and instability in the country continues to impact healthcare availability. Care is heavily dependent on the support of humanitarian programs and international federations, who provide essential basic medicine.
- The distance between patients and available care in this vast country also poses a barrier to accessing care, treatment, and services.
- Two specialist centers exist for hemophilia and additional branches are being developed in additional provinces; care is coordinated by a lead center, who has a blood bank and clinical team. However, security continues to be a problem preventing the free movement of patients to access their treatment.



Rare Disease Patient Organization Landscape

- Patient groups work closely with international federations and local experts to organize care for people living with rare diseases as the healthcare infrastructure was destroyed due to conflict.
- The World Federation of Hemophilia is providing support through humanitarian aid and provision of factor treatment, nevertheless, some people can experience shortages of factor for up to six months.




¹⁰ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020
<https://doi.org/10.1016/j.jval.2020.06.020>

BAHRAIN



Rare Disease Policy Landscape

-  The Ministry of Health is generally supportive of rare disease and is planning to develop the Bahrain Genome Project.
-  Bahrain has a definition for orphan medicines.¹¹ Orphan medicines are funded by the government and treatments are available depending on approval and funding.


Rare Disease Health System Landscape

-  Patients must be referred to the governmental hospital for a clinical review in order for a funding request for treatment to be submitted to the government.
-  Similar to other countries in the region, consanguine marriages are culturally acceptable and there is little awareness of the risks of conception, birth defects and abnormalities and the increased risk of autosomal recessive disorders for the offspring. Pre-marital genetic screening is offered in the country. Genetic carriers are incidentally identified through fertility services.
-  The Al Jawhara Center is a leading genetics center and has been campaigning to raise awareness of rare diseases in Bahrain and the wider region. The AL Jawhara Center and Arabian University, in collaboration with the Ministry of Health, have been active in organizing events for Rare Disease Day since 2013, hosting educational and awareness events.

CENTERS

-  Al Jawhara Center for Molecular Medicine, Genetics and Inherited Disorders (Manama)
-  Salmaniya Medical Complex (Manama)

Rare Disease Patient Organization Landscape

-  There is no formal rare disease patient organization or group, however the Al Jawhara Center supports affected individuals and their families.

¹¹ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020 <https://doi.org/10.1016/j.jval.2020.06.020>

EGYPT

Rare Disease Policy Landscape

- The 2014 Constitution addresses health as a fundamental human right and has greatly improved healthcare in Egypt. Recently, the government began working towards universal health coverage.
- Therapies are evaluated by a committee of doctors approved by the Ministry of Health. However, there is a lack of knowledge and understanding of the different conceptualization needed in the evaluation of orphan medicines compared with routine therapies. Treatments provided by the Ministry of Health do not cover all needs, for example, surgical operation.
- Rare diseases compete against other health priorities, including cancer and chronic diseases such as diabetes and heart disease.

Rare Disease Health System Landscape

- Healthcare is delivered through private and public hospitals. Social insurance pays for care and treatment in governmental and university hospitals. The national institutes and university hospitals are well equipped but there is limited awareness and resources for rarer diseases. Moreover, there is a huge variation in the treatment patients receive, depending on the hospital.
- Nearly two-thirds of the population come from rural area in Egypt to seek care and advise in the main cities. As Egypt is situated at the heart of the East Mediterranean region, people also travel from countries in the North Africa, the Levant and Arabian Gulf to access care and treatment.
- The newborn screening program was established in early 2000s. The genetic counseling program started in 2002. Rare disease patients access care in private clinics for genetic tests and the samples are sent abroad.

NETWORKS & CENTERS

- Experts in Egypt actively network with other centers within the region and internationally. These experts participate in international networks, conferences, research, and registry activities.

CENTERS

- Human Genetics and Genome Research Division at the National Research Center (Giza)
- Ain Shams University (Cairo)


Rare Disease Patient Organization Landscape

- Patient organizations collaborate with the government to secure support and appropriate treatment for people living with a rare disease. Patient groups provide emergency support and in collaboration with international federations, provide humanitarian support to enable people to access operations and essential medicine when patients are not covered by the national system.
- The Egyptian Society of Medical Genetics organizes events to raise awareness of rare diseases for Rare Disease Day.

ISLAMIC REPUBLIC OF IRAN

Rare Disease Policy Landscape

- Rare diseases are defined within the country's policies and there is also a definition of orphan medicines.¹²
- Orphan medicines are available in the country and are reviewed for public system reimbursement by the Ministry of Health's health technology assessment body.
- It is reported that the country is working on developing a national plan for rare diseases.
- Iran faces political sanctions that may indirectly impact healthcare within the country as it is reportedly impeding greater international collaboration.

 RD Definition:
Less than 1 in 200,000

Rare Disease Health System Landscape

- Health insurance coverage varies. Complimentary insurance is taken out to supplement the basic medical care.
- Genetic counselling services across Iran are well developed and available under public health insurance and private clinics, except for whole genome sequencing (WGS). While the performance of WGS is sent abroad, the interpretation of the results is undertaken in Iran. The genetic clinics are connected to a network of specialists ensuring diagnosis and follow-up care is provided to affected individuals.

Rare Disease Patient Organization Landscape

- The Iranian population has a good level of awareness of rare diseases, due to awareness raising activities of the national alliance which include media coverage. Therefore, there is less social stigma for affected individuals and families.
- The national patient group, the Rare Disease Foundation of Iran (RADOIR) established a patient registry system (SABNA) to identify and support the registered rare patients with facilities and services.
- RADOIR is a national group developing a National Strategic Plan for rare diseases in collaboration with Tehran University of Medical Sciences (TUMS).¹³

NATIONAL GROUP
Rare Disease Foundation of
Iran (RADOIR)

¹² Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020 <https://doi.org/10.1016/j.jval.2020.06.020>

¹³ <https://radoir.org/en/wp-content/uploads/2021/08/Rare-Diseases-Foundation-of-Iran-RADOIR-2008-2020.pdf>

IRAQ

Rare Disease Policy Landscape

- Once an affluent country with well-organized healthcare services, Iraq now faces significant challenges due to the political situation, instabilities, and conflicts.
- Iraq has an official list of orphan drugs.¹⁴

Rare Disease Health System Landscape

- Healthcare and hospital infrastructure have been significantly damaged due to conflicts; healthcare is reliant on humanitarian organizations and non-governmental organizations who offer basic healthcare.
- Conflicts, violence and threats also results in a human resource shortage of healthcare practitioners.
- Genetic testing is expensive and not readily available in the country.
- There are periods of up to 6-7 months where there is a shortage of essential care, treatment and medicine. These shortages negatively impact the lives of people living with a rare disease and can cause disability or lead to life-threatening situations for this vulnerable population.
- Iraq took part in Rare Disease Day for the first time in 2020, with the pediatric hospital in Al Sulaymaniyah organizing an event with speakers and gifts for rare disease patients.

Rare Disease Patient Organization Landscape

- Patient groups are active, nationally and with international federations, to bring the community together and focus on improving awareness action with the government.
- Some international federations, like the World Federation of Hemophilia, provide meaningful support to patients, providing humanitarian aid and creating awareness.

¹⁴ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020
<https://doi.org/10.1016/j.jval.2020.06.020>

KUWAIT




Rare Disease Policy Landscape

- Public healthcare services for general care are divided across Kuwait's governments creating fragmentation of care and larger health inequities for rare disease communities.
- No orphan drug legislation or definition of rare diseases identified in the country.¹⁵

Rare Disease Health System Landscape

- Healthcare, including for rare diseases, is provided by public government-backed free hospitals, which centralize specialized healthcare. However, large patient volumes, negligible allocations specific to rare diseases, and short working hours, result in disparities in services leading families with financial means to turn to the private sector to fill in the gaps. The private sector does not necessarily provide specialized services but is more accessible.
- Healthcare is not institutionalized and is dependent on the doctor's capacity and willingness to continue to learn as the rare disease space and knowledge evolves rapidly.
- Kuwait initiated a Genome Project led by the Genatak Genomic Medicine Center to sequence individual genomes of the Kuwait population under three sub-groups: Saudi Arabian, Persian, and Bedouin. The Genome Project was established to support the implementation of personalized genomic medicine.
- Healthcare quality is highly correlated with patient self-advocacy and awareness of the disease and treatment updates.



CENTERS

-  Kuwait Medical Genetic Center (Shuwaikh Industrial)
-  National Bank of Kuwait Children's Hospital (Shuwaikh Industrial)
-  Genatak Genomic Medicine Center




¹⁵ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020
<https://doi.org/10.1016/j.jval.2020.06.020>

MOROCCO



Rare Disease Policy Landscape

-  No orphan drug legislation or definition of rare diseases identified in the country.¹⁶
-  The Ministry of Health is reportedly collaborating with patient organizations to develop a National Plan for Rare Diseases

Rare Disease Health System Landscape

-  The country is divided into 16 regions and so is the healthcare system¹⁷
-  Coverage of healthcare services, lack of resources, and high costs of medicines lead to a delay in securing a diagnosis and access to treatment. This is reported to be the main issues for patient groups in Morocco.
-  There are two national reference centers for hemophilia.

Rare Disease Patient Organization Landscape

-  The patient community is active and collaborates with the Ministry of Health.
-  The National Alliance of Rare Diseases (AMRM) has been active in supporting collaboration across families and professionals.

NATIONAL GROUP
National Alliance of Rare
Diseases (AMRM)

¹⁶ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020
<https://doi.org/10.1016/j.jval.2020.06.020>

¹⁷ https://www.who.int/workforcealliance/knowledge/PPE_Morocco_CaseStudy.pdf

OMAN

Rare Disease Policy Landscape

- The Ministry of Health is the sole regulator of healthcare and has established a high-quality public healthcare system in Oman. Rare diseases are not a health priority; most resources are allocated to common conditions like cancer and trauma.
- Orphan drugs are defined within the country's legislation.¹⁸ The Ministry of Health has an HTA committee to review new medicines, but there is a lack of understanding of the concept of rare diseases and orphan medicines.

Rare Disease Health System Landscape

- Healthcare is free for Omani citizens but not for non-citizens. Refugees are covered for medical tests and healthcare; however, cancer care is not covered in this care package.
- Oman has a genome program to build data and a gene profile for the Middle East, supporting the move to personalized medicine. However, all genome tests are sent abroad for processing which impacts access to diagnostic services due to costs.
- Premarital counselling and screening have been established to reduce the incidence rates of congenital and genetic disorders.

NETWORKS & CENTERS

- Healthcare is organized under 10 regions, with each region having a central regional referral hospital that is supported by a network of primary and secondary services.¹⁹ The two main specialist and tertiary hospitals, Royal Hospital and Sultan Qaboos University Hospital, are based in the capital city of Muscat. However, services are fragmented between the hospitals.
- The Ministry of Health has developed a clinical genetics service through the referral hospital in each region, connected to the Royal Hospital.
- A large proportion of the population lives in the rural areas, which makes it difficult to access specialist medical services based in the capital. However, diagnosis and treatment for many diseases can be provided locally.

CENTERS

- Royal Hospital (Muscat)
- Sultan Qaboos University Hospital (Muscat)

Rare Disease Patient Organization Landscape


- There is a lack of patient groups in the country. Nevertheless, Rare Disease Day has been celebrated since 2014.

¹⁸ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020 <https://doi.org/10.1016/j.jval.2020.06.020>




¹⁹ Grant CS, Al-Kindy N. Surgery in Oman. Arch Surg. 2005; 140:21-25

PAKISTAN





Rare Disease Policy Landscape

-  No orphan drug legislation or definition of rare diseases identified in the country.²⁰



Rare Disease Health System Landscape

-  Over the past 15 years, Rare Genetic Research has found that Pakistan has a high frequency of rare diseases; consanguineous marriages are one of the leading causes of rare genetic disorders in the population.
-  Less than 1% of newborns are currently being screened for rare diseases as only handful of hospitals (mainly private) offer these services.
-  The Department of Pathology and Laboratory Medicine in collaboration with Division of Women & Child Health, the Aga Khan University (AKU), organized a four-day conference in Karachi in March 2020. The conference brought together genetic researchers, pathologists, and child health specialists from public and private sector organizations across the country in one platform. The conference program focused on outcomes and challenges in management of inherited metabolic disorders, new-born screening, rare disorders case presentations, and good laboratory practices alongside the ethical challenges.

CENTERS

-  The Children's Hospital Lahore (Lahore)
-  Aga Khan University Hospital (Karachi)
-  Liaquat National Hospital (Karachi)
-  Pakistan Institute of Medical Science (PIMS) (Islamabad)

Rare Disease Patient Organization Landscape

-  Social stigma attached to patients with rare disease is also a hurdle in terms of social acceptability and integration.
-  Rare Disease Day has been celebrated since 2012. The day has been used to host educational seminars, poster presentations, and group discussions on rare diseases. For Rare Disease Day 2020, there were a number of educational events held around the country. Conference and webinar topics covered included bridging the gap between stakeholders and genetic testing. There were also literacy, education, and awareness programs that took place throughout Pakistan.

²⁰ <https://www.keionline.org/wp-content/uploads/KEI-Briefing-Note-2020-4-Defining-Rare-Diseases.pdf>

QATAR


Rare Disease Policy Landscape

- While Qatar is quite active in advocating for rare diseases in the international sphere, it does not have orphan drug regulations or policies specific to rare diseases or rare disease healthcare.²¹
- In September 2023, Her Excellency Minister of Public Health Dr. Hanan Mohammed Al Kuwari stated Qatar's commitment to universal health coverage and its awareness of rare diseases as a global concern. She explains that the scarcity of expertise and specialized centers and the related economic burdens necessitate cooperation and open communication with regional and international scientific institutions, to ensure equitable access to health care for those in need.²²

Rare Disease Health System Landscape

- Centralized and well-resourced healthcare system.
- In 2015, the Qatar Genome Program was launched to establish the path to personalized medicine and position Qatar as a pioneer in precision medicine. The aim of the Genome Program was to sequence over 10,000 whole genomes. The population-based project has developed a database of whole genome sequencing, omics, and phenotypic data in the 'Qatar Biobank'. The data is being used for research and to inform healthcare policy in the country.
- Academic and scientific research has been integrated into healthcare centers through the implementation of genomics into the clinical setting. Research is being completed under national research partnerships through the Qatar Genome Program research consortium with over 90 researchers, the Qatar National Research Fund (QNRF), and a national genome data network.
- The Hamad Medical Corporation (HMC) and the Heidelberg University Hospital (HUH) collaborated to establish in 2019 the National Centre for Rare Diseases to offer clinical care to rare disease patients, provide rare disease education to clinicians, and engage in rare disease research.²³ The center is visited by people with rare diseases from Middle Eastern countries.
- Qatar has a National Newborn Screening Program since 2003 that supports the diagnosis of rare genetic and metabolic disorders.²⁴

CENTERS

-  National Centre for Rare Diseases

²¹ <https://hukoomi.gov.qa/en>

²² <https://qna.org.qa/en/News-Area/News/2023-09/23/0040-qatar-participates-in-side-event-on-universal-health-coverage-for-people-with-rare-diseases>

²³ <https://www.isns-neoscreening.org/qatar-the-first-anniversary-of-the-establishment-of-the-national-centre-for-rare-disease/>

²⁴ <https://menafn.com/1099580614/Qatar-QNSP-diagnoses-over-1000-newborns-with-diseases-disorders>

SAUDI ARABIA

Rare Disease Policy Landscape

- Prescriptions for novel treatments are submitted via an application to the Ministry of Health for approval as the government covers the cost for rare disease therapies.

Rare Disease Health System Landscape

- Healthcare is provided either through governmental hospitals or private clinics and is funded by the state for Saudi citizens.
- There is a high level of competency and expertise in Saudi Arabia, normally based in the main hospitals in the main cities, but rural populations can struggle to access care in the cities.
- There is no structured primary pediatric care resulting in a significant delay in diagnosis for most patients with rare disease. Referral to tertiary care centers for genetic diagnosis is the only option for these patients and most testing is sent to external labs in EU or the US. Access to medicines and therapies for rare diseases depends on the prevalence of the disease and patient's access to a tertiary care center.
- Saudi Arabia has a high prevalence of severe inherited diseases, which is estimated to affect 8% of births, presenting a significant health burden on families and the national healthcare system. This incidence of rare disease is increased due to the social practices of inter-ethnic marriages (which can have a bigger impact in smaller communities). Pre-marital genetic screening is offered. The newborn screening panel covers many metabolic diseases where treatment is available.
- In 2013, Saudi Arabia launched the Saudi Arabia Genome Program to map 100,000 genomes and it is one of the world's top 10 genome programs. The Genome Program aims to identify the genetic bases of severe and common inherited disease in the Saudi population and establish a foundation for genomic medicine. The program intends to share data and expertise between the Arabian Gulf region and internationally.

CENTERS

- Eastern Region Business Unit
- King Abdulaziz City for Science
- Prince Mohammed Bin Fahd Hospital of Genetic Blood Disease (Al Qatif)

Rare Disease Patient Organization Landscape

- There are disease-specific patient organizations in the country. Rare disease day has been celebrated since 2013, and organized by patient groups and the local hospitals, providing lectures for health professionals and presentations of patient testimony and stories.

SYRIAN ARAB REPUBLIC

Rare Disease Policy Landscape

- There is a significant lack of rare diseases classification and integrated definition.

Rare Disease Health System Landscape

- As healthcare and hospital infrastructure has been significantly damaged due to past conflicts; healthcare is reliant on humanitarian organizations and non-governmental organizations who offer basic healthcare.
- Surrounding conflicts, violence and threats also results in a human resource shortage of healthcare practitioners.
- Healthcare for thalassaemia is provided in the Syrian Arab Republic under a national guideline and through nine specialized health units distributed across the country.

Rare Disease Patient Organization Landscape

- Syria has been raising awareness of rare diseases since 2018. School children in Syria learn about rare diseases and show their solidarity for those living with a rare disease and their families by taking part in the Rare Disease Day campaign.

TUNISIA

Rare Disease Policy Landscape

- No orphan drug legislation or definition of rare diseases identified in the country.²⁵ The list of covered diseases has not been updated in several decades.

Rare Disease Health System Landscape

- Rare diseases are not covered by the governmental healthcare system, except for a few diseases.
- Some patients receive treatment through humanitarian programs from pharmaceutical companies and others by participating in clinical trials.
- For hemophilia, there are three good multi-disciplinary treatment centers who are collaborating and deliver prophylactic therapy.
- Patients who do not live in the city have to travel long distances, up to 3-4 hours, to get treatment.

Rare Disease Patient Organization Landscape

- There are several active patient organizations: National Alliance of Tunisia, the Tunisian Association of Angelman and Rett Syndromes (ATSAR), the Tunisian Association of Intermediate Metabolism Diseases (ATMMI), the Tunisian Association for Lysosomal Diseases (ATML), the Tunisian Association of Hemophilia (ATH), the Tunisian Association of Patients with Primary Immunodeficiency Diseases (ATUN-DIP), DEBRA Tunisia and the Association to help children suffering from Xeroderma pigmentosum.

NATIONAL GROUP National Alliance of Tunisia

²⁵ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020 <https://doi.org/10.1016/j.jval.2020.06.020>

UNITED ARAB EMIRATES

Rare Disease Policy Landscape

- No orphan drug legislation or definition of rare diseases identified in the country.²⁶

Rare Disease Health System Landscape

- The United Arab Emirates has a well-resourced healthcare system and excellent infrastructure and global partnerships. The government is responsible for providing all available healthcare services for UAE citizens and if any services are not available, then the government is obliged to send patients abroad.
- There is a high incidence of genetic disorders, specifically thalassemia, muscular dystrophy, and kidney disease. Screening for thalassemia and other diseases before marriage is becoming mandatory and a mandatory neonatal screening program is offered free of charge by the government.
- There is, however, a lack of genetic counselling especially for the Arabic speaking. This is a barrier for communication and providing information.
- Research collaborations exist with Bahrain and Oman.

CENTERS

- Fujairah Thalassaemia Centre (Fujairah)
- Genetic and Thalassemia Centre (Dubai)
- Faculty of Medicine and Health Sciences (Al-Ain)
- National Screening Centre for Women and Child Health (Abu Dhabi)

Rare Disease Patient Organization Landscape



- The UAE Rare Disease Society is an active patient organization that partners with centers to actively raise awareness of rare diseases and educate the public through Rare Disease Day.

NATIONAL GROUP
UAE Rare Disease Society



²⁶ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020
<https://doi.org/10.1016/j.jval.2020.06.020>

YEMEN


Rare Disease Policy Landscape

-  Yemen is experiencing a humanitarian crisis, including many in the country facing severe poverty and malnutrition. The ongoing conflict, as well as the economic decline, continues to disrupt healthcare services.
-  Orphan drugs are defined within the country's legislation.²⁷

Rare Disease Health System Landscape

-  Healthcare services have been significantly affected since 2015 with only half of health facilities operational and with many facing shortages of basic equipment and health workers. The lack of availability of essential, basic treatments, continues to impact the health of vulnerable communities.
-  The healthcare system is further stressed by the increasing number of people emigrating to Yemen as the system cannot provide basic care for everyone in need.

Rare Disease Patient Organization Landscape

-  National thalassemia patient groups are calling for international action to provide essential therapies and services to patients in the country.

²⁷ Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis. Chan et al. 2020
<https://doi.org/10.1016/j.jval.2020.06.020>