RARE DISEASES INTERNATIONAL

RESOURCE MAPS

A resource to understand the global rare disease landscape
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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 categorization\(^1\) 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.

<table>
<thead>
<tr>
<th>Population size (2020) in Millions(^2)</th>
<th>Estimated rare disease population prevalence (3.5-5.9%) in Millions(^3)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. African Region</td>
<td>1,036.1</td>
</tr>
<tr>
<td>2. Region of the Americas</td>
<td>1,018.1</td>
</tr>
<tr>
<td>3. East Mediterranean Region</td>
<td>730.8</td>
</tr>
<tr>
<td>4. European Region</td>
<td>932.9</td>
</tr>
<tr>
<td>5. South-East Asia Region</td>
<td>2,021.4</td>
</tr>
<tr>
<td>6. Western Pacific Region</td>
<td>1,939.2</td>
</tr>
<tr>
<td>Total</td>
<td>7,678.5M</td>
</tr>
<tr>
<td></td>
<td>267.1M – 452.3M</td>
</tr>
</tbody>
</table>

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\(^1\) https://datahelpdesk.worldbank.org/knowledgebase/articles/906519-world-bank-country-and-lending-groups


SOUTH-EAST ASIA REGION (SEAR)

Countries: Total of 11 Member States

<table>
<thead>
<tr>
<th>Member States</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bangladesh, Bhutan, Democratic People’s Republic of Korea, <strong>India</strong>, <strong>Indonesia</strong>, Maldives, Myanmar, <strong>Nepal</strong>, <strong>Sri Lanka</strong>, <strong>Thailand</strong>, Timor-Leste</td>
</tr>
</tbody>
</table>

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

4 Image created with mapchart.net
SEAR Rare Disease Landscape Overview
As reported by patient leaders and medical experts from six countries in the region

**Rare Disease Policy Landscape**
- The region has variations in the level of recognition and political support for rare diseases at country levels, as well as in the recognition and support of different types of rare diseases. For example, hemoglobinopathies tend to have strong political support and are covered by public insurance, while other rare diseases remain neglected in many countries. Nevertheless, an increased number of governments are recognizing the burden of rare diseases on their populations and committing to tackling rare diseases as a public health priority.
- Lack of uniform legislation, policy and regulation has repercussions within healthcare systems, creating either lack of services or variations in quality, patient rights and reimbursement.
- The Asia Pacific Economic Council (APEC) has a Rare Disease Strategy in Support of the ‘Healthy Asia Pacific 2020’ Initiative and advocates for better access and care through a Rare Disease Action framework and the formation of a Rare Disease Expert group. There is also the ASEAN+ Rare Disease Network: Unifying Voices of Patients, which was formed in 2017.

**Rare Disease Health System Landscape**
- Across the 11 countries in the region, dramatic differences are noted in the organization and maturity of the healthcare systems, especially for rare disease management, between countries and between states. Healthcare coverage is dependent on the public or private insurance schemes which range from free public health (Sri Lanka) to partial coverage, with some financial support available for certain rare diseases (Thailand, Nepal, India). The limited coverage of rare diseases results in families having to pay out-of-pocket to cover costs of care.
- Pockets of expertise exist with a growing number of centers of excellence for rare diseases, mostly located in urban cities across the region. In Thailand, seven Rare Disease Centers have been appointed since October 2019 as part of the Thai Universal Coverage Scheme. In India, Centers are informally recognized in Bengaluru, Chandigarh, Hyderabad, Kolkata, Mumbai, New Delhi, and Lucknow. In Sri Lanka, the two pediatric hospitals in Colombo and Peradeniya treat PLWRD.
- Access to innovative medicines is a challenge as there is no unifying regulatory mechanism in the region, and each country must individually negotiate with industry for regulatory approvals.
- Most countries do not have registries that capture the existing patients being treated.
- Inadequate information and awareness amongst health professionals, regulators/government agencies, and the public remains a key issue in securing improvement to care and patients’ rights.

**Rare Disease Patient Organization Landscape**
- The rare disease population in the region remains largely invisible and without a voice. However, engagement from the patient community is growing across the region and the patient voice is becoming stronger.
INDIA

Rare Disease Policy Landscape
India has a National Policy for Rare Diseases adopted in 2021. The New Drug and Clinical Trials Rule 2019 makes special provisions for orphan drug approvals. A national rare disease treatment policy launched in 2018 has not been formally accepted due to lack of funds and it is incomplete as it does not address all rare diseases. This policy includes considerations around disease categorization and planned funding mechanisms. Announced July 2023, a sum of $15 million USD was allocated to Rare Disease Centers of Excellence.

Rare Disease Health System Landscape
India has a very diverse and complex healthcare system with a three-tiered public health system and a multi-layered, private health system with varying capacity. The government health insurance schemes cover primary healthcare. Rare diseases have been traditionally excluded from public insurance schemes as they fell under an exclusion criterion covering genetic disorders. This resulted in out-of-pocket spending making the cost of care prohibitive for many PLWRD. However, this is now changing; some genetic disorders are now included in public health insurance schemes following the advocacy action of the Insurance Regulatory and Development Authority (IRDAI) that proposes providing some financial support for rare disease treatments to patients.
India is one of the largest producers of generic medicines in the world. Medicines for rare diseases which have a generic version available are considerably cheaper. However, many orphan drugs are not yet available as generics. This results in high prices, unaffordable to most citizens.
Significant variations exist between India’s states in the recognition and support of rare diseases. Some States are more advanced in their offer of treatment, newborn screening, and expert centers are located in less than half of the 28 states. There are 542 medical colleges in India but less than 50 have a functional genetic department and even fewer have a comprehensive team for managing rare diseases. There are more than 100,000 private hospitals and nursing homes in India but only a small subset of these have the expertise to manage a range of rare diseases. Some private diagnostic centers maintain a genome database, but this is not easily accessible.
Referral systems in India are weak as appointments can take place at a tertiary center for the first visit and with no follow up, many patients drop out of the system after learning the diagnosis as either no treatments are available, or they are unaffordable. Patients also migrate between various systems of medicine making it hard to track and follow up.
There are pockets of medical expertise and genetic services in most major city hospitals, with a network of over 40 genetic consultants. Still, expertise is fragmented and concentrated primarily in capitals and main cities whereas most of the population resides in rural areas.

NETWORKS & CENTERS
India has a national registry instituted by the Indian Council of Medical Research to collect data from 37 outreach centers; however, India still lacks a national rare diseases registry and data collection has been a major challenge as private hospitals are not mandated to report, and public hospitals have not yet established a mechanism to do so.
Active medical collaborations exists between India and countries such as US, Nepal and Sri Lanka

5 https://main.mohfw.gov.in/sites/default/files/Final%20NPRD%2C%202021.pdf
7 http://bmi.icmr.org.in/irdr/index.php
Patient communities are crucial to supporting access to care for families and affected individuals.

Well-established diagnosis and surveillance guidelines have been organized through the advocacy work of patient organizations. The aim is for patient groups to support dissemination and implementation so guidelines are better defined and followed.

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**Rare Disease Patient Organization Landscape**

Patient communities are crucial to supporting access to care for families and affected individuals.

Well-established diagnosis and surveillance guidelines have been organized through the advocacy work of patient organizations. The aim is for patient groups to support dissemination and implementation so guidelines are better defined and followed.

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**CENTERS**

- All India Institute of Medical Sciences (New Delhi, Jodhpur)
- Maulana Azad Medical College (New Delhi)
- Sanjay Gandhi Post Graduate Institute of Medical Sciences (Lucknow)
- Post Graduate Institute of Medical Education and Research (Chandigarh)
- Center for DNA Fingerprinting & Diagnostics (Hyderabad [diagnostic and research])
- King Edward Medical Hospital (Mumbai)
- Institute of Post-Graduate Medical Education and Research (Kolkata)
- Center for Human Genetics (CHG) with Indira Gandhi Hospital (Bengaluru [twin center])
- Sir Ganga Ram Hospital (New Delhi)
- Institute of Genomics & Integrative Biology (New Delhi [Research])
- Nizam Institute of Medical Sciences (Hyderabad)
- Center for Cellular & Molecular Biology (Hyderabad)
- L. V. Prasad Eye Institute (Hyderabad)
- Institute of Child Health (Chennai)
- Sri Ramachandra, Institute of Higher Education and Research (Chennai)
- Manipal Hospital (Bengaluru)
- Kasturba Hospital (Manipal)
- Christian Medical College (Vellore)

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**NATIONAL GROUPS**

- Organization for Rare Diseases India (ORDI)
- Indian Organization for Rare Diseases (IORD)
INDONESIA

**Rare Disease Policy Landscape**
No official definition or legislation for rare disease or orphan drugs.
Nationwide newborn screening policy has not been established yet, except for congenital hypothyroidism.

**Rare Disease Health System Landscape**
In 2017, Indonesia opened its first center of excellence for rare diseases in Cipto Mangunkusumo Hospital in conjunction with Human Genetic Research Center and Indonesia Medical Education and Research Institute Universitas Indonesia (IMERI UI), and patient organization to support families affected by rare diseases.⁹

**CENTERS**
National Center for Rare Disease Cipto Mangunkusumo Hospital (CMH)

**Rare Disease Patient Organization Landscape**

**NATIONAL GROUP:** Yayasan MPS dan Penyakit Langka Indonesia (the Indonesian MPS and Rare Diseases Foundation)

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⁹ [https://www.penyakitlangkaindonesia.org/events/official-opening-indonesia-center-excellence-rare-diseases](https://www.penyakitlangkaindonesia.org/events/official-opening-indonesia-center-excellence-rare-diseases)
NEPAL

Rare Disease Policy Landscape

Despite having a prevalence of less than 5/10,000, rare diseases still present a major public health problem in the country. Some diseases, which are considered rare diseases based on the global burden status, may have a higher burden in certain areas of the country.

There is a national program for control and elimination of a few rare diseases, which includes better management and surveillance systems. More knowledge about rare diseases and their burden is needed to increase the priority of targeting all rare diseases for the government.

Rare Disease Health System Landscape

Diagnosis and treatment are free for some rare diseases within the country’s national program but comprises primarily rare infectious disease, e.g., diagnostics (Leishmaniasis, Malaria), treatments (Leprosy, Leishmaniasis, Malaria), vaccines (Rabies, Japanese encephalitis, Measles, Rubella), preventive chemotherapy (Lymphatic Filariasis). Most genetically inherited rare diseases, e.g., neurofibromatosis, tuberous sclerosis, epidermolysis bullosa, are paid out of pocket by individuals. There is a provision for some financial support of select rare diseases, including some blood disorders and malignancies, cardiac problems, and neurological disorders; however, it does not cover the full treatment cost.

Although public health system is strong, the lack of clinical expertise at all levels of healthcare is still a major challenge.

NETWORKS & CENTERS

Expert centers in Nepal collaborate in the region with rare disease experts in India and Sri Lanka, including Human Genetics Unit (Faculty of Medicine, University of Colombo) in Sri Lanka.

CENTERS

National Academy of Medical Sciences, Bir Hospital (Kathmandu)

Rare Disease Patient Organization Landscape

National alliances exist as well as disease-specific organizations, building multi-stakeholder collaboration and working with governments.

Rare Diseases Society of Nepal (RDSN) is the only umbrella organization dedicated to improving the standard of care for patients diagnosed with rare diseases in Nepal.

NATIONAL GROUP

Rare Diseases Society of Nepal
SRI LANKA

Rare Disease Policy Landscape
In Sri Lanka, the government provides free healthcare to all citizens including rare disease coverage. According to the WHO, Sri Lanka has achieved 75% universal health coverage.

Rare Disease Health System Landscape
The network of national hospitals comprises teaching hospitals in major cities, provincial and district hospitals in towns, and base hospitals in villages. Every citizen has direct access to the hospital of their choice. Referrals to a provincial and/or teaching hospital are made by specialists when required. Doctors have been trained in sub-specialties such as Pediatric Endocrinology, Pediatric Nephrology, Pediatric Hematology. There are also Pediatric Geneticists in training. There is a database of rare diseases compiled by the College of Pediatricians.¹⁰

NETWORKS & CENTERS
Sri Lanka also has specialized centers. There is a network of two pediatric diagnostic centers, based in Colombo and Peradeniya, that treat PLWRD and connect them with additional treatment centers. Experts in the country collaborate in the region with rare disease experts in India and Nepal.

CENTERS
- Human Genetics Unit, Faculty of Medicine, University of Colombo

¹⁰ https://drive.google.com/file/d/1DMwENj0mTVhZoZDjrbnuiFlHuH3Kx8T/view
THAILAND

Rare Disease Policy Landscape

- 24 rare diseases are included as part of the National Health Security Office universal health coverage scheme; a Rare Disease Committee holds a budget meant to cover this pre-determined list. The number of diseases covered, are expected to progressively increase.
- Patient groups are in good relationship with the government. There is a patient-supported national registry that has enabled data collection on disease, unlocking budgets and securing funding.

Rare Disease Health System Landscape

- Expertise in rare diseases is largely based in the capital and main cities (not in rural areas).
- The Nation Health Security Office (NHSO) has appointed seven rare disease centers through the Thai Universal Coverage Scheme since October 2019. The centers conduct telephone consultations and special/fast-track referrals for 24 inborn metabolic disorders with possible urgent/emergent conditions, from hospitals all over the country.
- “Genomics Thailand” (GeTH) is a national-level operational research initiative focusing on genome sequencing of rare disease patients and other disease areas (cancer, noncommunicable diseases, infectious diseases, pharmacogenomics). The network for rare diseases under the GeTH project involves the seven rare disease centers appointed by NHSO and two other centers under the “Thai Rare & Undiagnosed Disease Network (T-RUN)”. The project runs between 2020 – 2024 and aims to sequence 50,000 Thai.

NETWORKS & CENTERS

- Thailand has established international collaborations, e.g., with US clinical teams.
- Thailand is also a part of international federations for the coordination and support of specific diseases (World Federation of Hemophilia and Asian Hemophilia Network).
- Collaboration among professionals through Asia Pacific Society of Human Genetics (APSHG).

- Chulalongkorn Hospital
- Thammasat Hospital
- Siriraj Hospital
- Ramathibodi Hospital
- Phramongkutklao Hospital
- Queen Sirikit National Institute of Child Health
- Khon Kean’s Srinagarind Hospital

Rare Disease Patient Organization Landscape

- Recognition of the patients’ voice is strong in Thailand.
- National alliances and disease-specific organizations, such as the Thai Rare Disease Foundation and the Thai Hemophilia Patient Club collaborate with experts and the government to develop services.
- There is also good collaboration across the country, with regional and international partners, e.g., working with World Federation of Hemophilia and Asian Hemophilia Network has led to the development of treatment and prevention guidelines.
### Countries: Total of 27 Member States

<table>
<thead>
<tr>
<th>Member States and Economic Areas</th>
</tr>
</thead>
<tbody>
<tr>
<td>Australia, Brunei Darussalam, Cambodia, China, Cook Islands, Fiji, Hong Kong, Japan, Kiribati, Lao People’s Democratic Republic, Malaysia, Marshall Islands, Micronesia (Federated States of), Mongolia, Nauru, New Zealand, Niue, Palau, Papua New Guinea, Philippines, Republic of Korea, Samoa, Singapore, Solomon Islands, Taiwan, Tonga, Tuvalu, Vanuatu, Vietnam.</td>
</tr>
</tbody>
</table>

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

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11 Image created with mapchart.net
WPR Rare Disease Landscape Overview
As reported by patient leaders and medical experts from eight countries in the region

**Rare Disease Policy Landscape**
Rare diseases are gaining increased recognition as a public health priority in the region (Australia, mainland China, Hong Kong, Japan, Malaysia, Philippines, Singapore, Taiwan). Australia and Taiwan, for example, have a national rare disease plans and China has committed to tackle rare diseases and coordinate national action across different governmental departments.

A legal definition for rare and intractable diseases exists in some countries (Taiwan, Japan) but this is not the case across the region (mainland China, Hong Kong). Rare diseases are included under the national (and federal) systems in Australia, Japan, Malaysia, and New Zealand.

**Rare Disease Health System Landscape**
The healthcare systems in the Western Pacific region are mainly funded through public-based insurance (Singapore, Philippines, New Zealand, Australia, Malaysia, China, Japan). Nonetheless, the level of entitlements and availability of treatments, and healthcare access times vary between and within countries.

Countries in this region have a large number of expert centers specializing in rare diseases as is the case in mainland China, Taiwan, Japan, Australia, New Zealand, and Singapore. However, China and Japan are the only countries to formally connect and designate these centers nationally.

Undiagnosed Disease Programs, which aid diagnostics in patients with rare diseases through next-generation sequencing, are established in the region (Singapore, Australia, Hong Kong).

Most countries have a national process and authority to evaluate and approve medicines (Japan, Malaysia, China, Hong Kong and Taiwan) providing guidelines for the national health system and health services, including access to some orphan medicines. However, access to orphan medicines is limited by the available budget in some countries (Malaysia). Also, evaluation processes can delay the approval of new medicines on the approved list.

The main hospitals are based in larger cities (for example, in China, Australia, New Zealand, Japan), which raises unique challenges in the provision to care to rural communities, particularly in geographically vast countries and island communities (Australia, Philippines).

**Rare Disease Patient Organization Landscape**
The rare disease population is generally well represented with a strong community of rare disease regional alliances, national groups, and disease-specific organizations.

These groups build multi-stakeholder collaboration including with governments bodies (China, Japan, Australia, etc.).
AUSTRALIA

Rare Disease Policy Landscape

Australia has a National Strategic Action Plan for Rare Diseases 2020 that focuses on awareness and education, care and support, and research and data. The Australia Therapeutic Goods Regulations (1997) provides guidance for orphan drug programs and incentives to market these medicines.

The national Medicare system subsidizes healthcare according to guidelines established by the Medical Services Advisory Committee (MSAC), an independent non-statutory committee established by the Minister for Health. Some healthcare services (including genomic and genetic testing) are subsidized. Applications can be submitted to add services to the Schedule.

A Medicare Pharmaceutical Benefits Scheme (PBS) is also in place to subsidize medications and treatments and enables free medicines for 139 ‘lifesaving and disease prevention’ medicines. Applications can be made to have new medicines added to this scheme; however, it can be a long process to secure approval, and it is reportedly lengthier and more complicated for rare diseases.

Health and social care are governed separately creating devolved systems with separate funding and service provision and in some cases limited collaboration and coordination. The healthcare system is multi-jurisdictional and is run by each state, but also overseen at a federal level, resulting in variation between States’ and Federal health portfolios.

The Australian Government has invested into rare disease research through the Medical Research Future Fund’s Genomics Health Futures Mission, and in the establishment of Australian Genomics.

Rare Disease Health System Landscape

There is a high level of public trust in the healthcare system and Australians are generally supportive of data sharing. Australia recently introduced the ‘My Health’ record (MHR) system, an online summary of health information where individuals have access to their own health record and consenting to data sharing. Nevertheless, there is no single national electronic medical record system, although some jurisdictions have a state-wide system.

All States have a newborn screening program supported by the Newborn Bloodspot Screening National Policy Framework, which includes coordinated national decision-making processes for adding additional tests to the program.

While there is disparity and inconsistency between states in the access to rare diseases screening and diagnostic testing, an even more significant disparity is seen between metropolitan and rural areas. A small number of rare diseases have more established standards of care and their own clinics are primarily located in metropolitan cities, but most do not. A national research project is investigating the value of case management in navigating health systems and services for PLWRD.

NETWORKS & CENTERS

Australia has established mature collaborations spanning every State and Territory and across the Asia Pacific Region and internationally (International Rare Diseases Research Consortium, IRDiRC; Undiagnosed Diseases Network International, UDNI; Asia Pacific Economic Council, APEC).

Australian Genomics is an Australian government initiative to support genomic research and its translation into clinical practice through broad engagement and a collaborative national approach. It is leading large research projects including rare disease flagships, pediatric acute care, and reproductive carrier screening.

Different states have different levels of maturity in organizing undiagnosed disease services, which aid diagnostics in patients with rare diseases through next-generation sequencing. Western Australia has a well-

RD Definition: Less than 5 in 10,000

National Strategic Action Plan for Rare Diseases 2020


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established Undiagnosed Disease Program (UDP); Victoria and New South Wales also have UDPs (RD Now and Gene2Care, respectively), and it is anticipated that other states will follow, with discussions underway to establish a national UDP network.

The National Center for Indigenous Genomics (NCIG), Canberra, and the Poche Center at the University of Sydney focus on Indigenous genomics and healthcare, respectively.

There are many Centers of Expertise across Australia, for example, disease-specific clinics for Noonan syndrome and other RASopathies (Angelman syndrome, cystic fibrosis, neurofibromatosis, etc).

**CENTERS**
- Rare Care, Center of Expertise for Rare and Undiagnosed Diseases (Perth)
- Interstitial Lung Disease of Childhood (ChILDRANZ) (virtual National Network) in collaboration with Europe, USA, Malaysia
- The Children’s Hospital at Westmead
- The Sydney Children’s Hospital, Metabolic disorders
- Genetic Health Queensland (Brisbane)
- Victorian Clinical Genetics Services (Victoria)
- The Pediatric and Reproductive Genetics Unit (Adelaide)
- Westmead Hospital Eye Clinic
- The Sydney Children’s Hospital, Connective Tissue disorders
- Connective Tissue Disorders (Sydney)
- Undiagnosed Diseases Program WA, Department of Health WA (Perth)

**Rare Disease Patient Organization Landscape**

The patient community in Australia was instrumental in the development and implementation of the *National Strategic Action Plan for Rare Diseases 2020*. Patient support groups and families, including the Genetic Support Network of Victoria fill the gaps in healthcare and support well-being.

**NATIONAL GROUPS**
- Rare Voices Australia
- Genetic Alliance Australia
CHINA

**Rare Disease Policy Landscape**

China has committed to tackling rare diseases, resulting in considerable improvements to the coordination of national action, including formal recognition of rare diseases, approval and reimbursement of medicines, selection of centers and development of national network.

Health China 2020 put rare diseases on the regional and central government agendas, resulting in orchestrated action across governmental departments. Two multi-stakeholder committees, made up of experts from across a wide spectrum of rare disease specialties, have been mandated to coordinate action to tackle rare diseases by the central government.

The first rare disease expert committee provided consultation to the government, which led to China’s ‘Rare Diseases Catalog’ (1st edition), announced in 2018, by five ministries. While there is no legal definition of rare diseases, the catalogue defined 121 “rare diseases” in China. The government has started to develop the 2nd and 3rd editions of the Catalogue.

There are huge differences on medical service capabilities between hospitals from different regions and provinces in China due to significant imbalances in social economic development. The National Rare Disease Collaboration Network was established to overcome this challenge.

**Rare Disease Health System Landscape**

There is no universal healthcare coverage, resulting in variation of services between regions. It is estimated that half of the three million patients are unable to secure access to a diagnosis.

In China, healthcare is organized under 34 provincial-level administrative divisions, classified as 23 provinces, four municipalities, five autonomous regions, and two Special Administrative Regions. Healthcare costs are reimbursed either at a national or provincial, or city level.

There are 63 treatments for 35 rare diseases available in China, of which 38 drugs for 21 rare diseases are reimbursed by the government, and a further nine rare diseases and ten drugs funded at a regional government level. More than 90 rare disease drugs are listed in the National Reimbursement Drugs List (NRDL).

China set up a patient registry, a nationally reimbursed drug list, and streamlined treatment approval process, leading to medicine approval time being reduced from 26 to six months.

**NETWORKS & CENTERS**

In mainland China the government established the ‘Chinese RD Diagnosis and Treatment Collaborative Network’, by selecting and connecting expert centers in the 23 provinces. The Network has a clear mandate and is organized to collaborate on i. enabling virtual case consultation and bi-directional referral system; ii. standardizing care through training, diagnosis, treatment guidelines, and clinical pathway; iii. supporting access to treatment by drug assessment, approval, and delivery; iv. performing clinical research; v. offering case management and support to patients to access specialist.

The collaboration network comprises 324 hospitals throughout China, with Peking Union Medical College Hospital (PUMCH) as the national-level center of excellence collaborating with 32 hospitals at the provincial level and 291 local hospitals at the municipality level. The selection of the hospitals in the network was based on their previous expertise and treatment of rare diseases as well as endorsed by their local health commissioners.

Hong Kong and Taiwan have existing collaboration within China and with the China Collaborative Rare Disease Network.
Rare Disease Patient Organization Landscape

China Alliance for Rare Diseases (CHARD) is an NGO under the auspices of National Health Commission. It’s a multi-stakeholder organization serving as a platform for medical institutions, universities, scientific research institutes, to develop rare disease treatment policies that support patients’ access to treatments in China.

The (Beijing) Illness Challenge Foundation is a patient organization localized in Beijing, China.

NATIONAL GROUPS
China Alliance for Rare Diseases (CHARD), Chinese Organization for Rare Disorders (CORD), Illness Challenge Foundation
HONG KONG

**Rare Disease Policy Landscape**
While Hong Kong reportedly has a high level of government support for rare diseases, there is no formal legal definition of rare diseases nor rare disease policies or rare disease expert centers. Rare disease policy in China does not currently apply to Hong Kong.

**Rare Disease Health System Landscape**
Hong Kong faces challenges in terms of infrastructure and expertise needed to offer comprehensive RD care. Rare disease patients are cared for through public hospitals; these are not expert centers, but they do cover some rare diseases.
Clinical diagnosis and assessments, multidisciplinary care and rehabilitation services, and subsidized drugs are available by the Hospital Authority, a statutory board that manages all public hospitals.
The region receives assistance with genetic testing and related support from Taiwan.
In recent years, to address the rising demand for rare disease treatments, Hong Kong established an expert panel to evaluate and introduce new orphan drugs to the public health care system. The drug expenses are subsidized by a government-led charity fund under a co-payment arrangement, and under a restructured health care delivery model for some specific rare disease groups. Despite these improvements, the variety of orphan products available in Hong Kong is still very limited, and the existing service model is inadequate to keep pace with the rapidly evolving rare disease science and treatment paradigm.

**NETWORKS & CENTERS**
Hong Kong is strengthening their regional collaborations, with rare disease experts in Hong Kong actively participating in the Asia Pacific Human Genetics Society to connect with other experts in the region and bring back knowledge of rare disease care to Hong Kong.
The New Hong Kong Children’s Hospital has begun collaborating on research and healthcare initiatives under the Undiagnosed Disease Network International, networking and expanding their knowledge base under the network.

**CENTERS**
- Princess Margaret Hospital (Lysosomal Storage Disease)
- Hong Kong Children Hospital

**Rare Disease Patient Organization Landscape**
The rare disease population is well represented through national groups in Hong Kong. Patient organizations are advocating the Hong Kong government to follow China’s rare disease development.

**NATIONAL GROUP**
- Rare Disease Hong Kong
**Rare Disease Policy Landscape**

In Japan, there is a universal healthcare system. All Japanese citizens bear part of medical care costs and have equal access to medical care, at the same cost, across Japan. Japan has a high level of government support for rare diseases, visible through its national efforts.\(^{13}\)

The term “NANBYO” (intractable disease) has been used in Japan for several decades, and rare diseases are encompassed in this term. Based on “the Outline of Measures against Intractable Diseases,” issued in 1972, a total of 56 disease groups had been identified by 2013. Under the NANBYO Law of 2014, health coverage scope was expanded to include 338 disease groups.

Drugs and medical devices can be designated as orphan drugs or medical devices (based on the Article 77-2 of the Act on Securing Quality, Efficacy and Safety of Pharmaceuticals, Medical Devices, Regenerative and Cellular Therapy Products, Gene Therapy Products, and Cosmetics) if they are intended for use in less than 50,000 patients in Japan and there is a high medical need.\(^{14}\)

**Rare Disease Health System Landscape**

There is a system for specific medical expenses based on the NANBYO Law, and patients with a designated NANBYO are subject to a maximum co-payment.

**NETWORKS & CENTERS**

In recent years, NANBYO and child welfare laws have shaped the development of expert centers into a national medical support network designed to support the treatment of NANBYO that are difficult to treat. The Network is organized based on prefectures. There are 47 "prefectures", and each prefecture has a base hospital for diagnosis of NANBYO, with a total of 64 hospitals.

The Network includes the National Research Center for Advanced and Specialized Medical Care, academic research groups and NANBYO societies, IRUD (see below), the NANBYO Information Center, and base hospitals for diagnosis of NANBYO in each prefecture.

The Initiative on Rare and Undiagnosed Diseases (IRUD)\(^{15}\) is a research network that coordinates people with undiagnosed diseases and enables the referral system between primary healthcare and medical specialists and clinical geneticists. IRUD has a Coordinating Center (1), Clinical Centers (39) and is supported by an IRUD Analysis Consortium (5) and Data Network (1).

**Rare Disease Patient Organization Landscape**

**NATIONAL GROUPS:** Advocacy Service for Rare and Intractable Diseases, Japan Patient Association.

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\(^{13}\) Six years' accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures - PubMed (nih.gov)

\(^{14}\) https://www.mhlw.go.jp/english/policy/health-medical/pharmaceuticals/orphan_drug.html#

\(^{15}\) Survey on patients with undiagnosed diseases in Japan: potential patient numbers benefiting from Japan’s initiative on rare and undiagnosed diseases (IRUD) - PubMed (nih.gov)

RD Definition: [https://www.keionline.org/wp-content/uploads/KEI-Briefing-Note-2020-4-Defining-Rare-Diseases.pdf](https://www.keionline.org/wp-content/uploads/KEI-Briefing-Note-2020-4-Defining-Rare-Diseases.pdf)
MALAYSIA

**Rare Disease Policy Landscape**

In May 2019, the Ministry of Health (MoH) formed the National Rare Disease Committee to support three Working Committees – i. Orphan Drug, ii. Clinical Management and Support, and iii. Advocacy and Health Education. Due to Government changes and the COVID-19 pandemic, the work is currently on hold; however, these committees will define rare diseases and management guidelines as well as promote advocacy issues in the country. A National Framework for Rare Diseases and plans to ensure the implementation of the Strategic Plan for Medical Programmes 2021-2025 was also recently announced.

Other ministries involved in rare disease care include the Ministry for Women and Family and Community Development, which issues disability registration cards to persons with disabilities, which may include those with rare disease based on their disability, not on their medical condition. Holders of the card receive various benefits, including free treatment and hospitalization, public transport, and utilities.

**Rare Disease Health System Landscape**

Due to public hospitals’ free, non-payment scheme, most rare disease patients access care and treatment through the public system, including access to some orphan drugs. However, the budget allocated for rare diseases treatments is insufficient due to the increased number of patients and it is limited to access through university teaching hospitals. Patients may receive treatment at private hospitals, but public insurance does not cover genetic or rare diseases.

When a referral is required for an assessment by geneticists or specialists, cases are referred to the main public hospital in Kuala Lumpur and some Ministry of Education (MoE) hospitals. Those needing further/extended genetic testing and supportive equipment may need to purchase it themselves or source funding from NGOs. There are limited number of genetic specialists and availability of hospitals with genetic clinics, but improvements in access for patients from rural areas and East Malaysia are currently underway. There are also plans to expand the newborn screening programs using heel-prick dried blood spots and to seek recognition for genetic counsellor’s post in public service.

There are clinical practice guidelines for selected rare disease conditions and management guidelines in pediatric and adult protocol books.

Social Worker officers are available at all public hospitals to assist patients and families from low-income groups. The Medical Social Worker officers based at the Medical Social Work Unit, are available at most MoH and MoE hospitals. At the recommendation of attending specialists/geneticists, they will support patients in seeking funding to meet/subsidize the cost of treatment, if required. This funding is obtained from the national Medical Support Fund (Tabung Bantuan Perubatan) but only applies to families that are poor and only provides 'one-off' support and not continued medication.

**NETWORKS & CENTERS**

National genetic services and referral pathways are provided by university hospitals under the National MoE and by public hospitals under the MoH.

Genetics services are networked across Malaysia and across the region through the Asia Pacific Human Genetics Society.
Malaysia has long-term collaborations with centers in Japan and genetic labs in the Republic of Korea, sending many patients from Malaysia to the Korean Genetic Lab.

**CENTERS**
- Department of Genetics, Penang Hospital
- Genetic Dept, Hospital Pulau Pinang
- Department of Genetics, Kuala Lumpur Hospital
- Medical Genetics Unit, University Malaya Medical Center
- Department of Pediatrics, Universiti Kebangsaan Malaysia (UKM) Medical Center in collaboration with Hong Kong, Singapore, UK, USA, Japan, Korea, Australia
- School of Medical Sciences, University Sains Malaysia
- Hospital USM

**Rare Disease Patient Organization Landscape**
Malaysia has various patient organizations operating in the rare disease advocacy space including Malaysian Rare Disorders Society, Malaysia Metabolic Society, Malaysia Lysosomal Diseases Association (MLDA)

**NATIONAL GROUP**
Malaysian Rare Disorders Society
NEW ZEALAND

Rare Disease Policy Landscape
There is no national plan or specific policies in place for rare diseases. The first national Rare Disorders Strategy is being developed.

Rare Disease Health System Landscape
New Zealand’s health and disability system is mainly funded by general taxation. The New Zealand Ministry of Health allocates more than three-quarters of the public funds it manages to District health boards. In turn, these boards use this budget to plan, purchase, and provide health services within their regions, including public hospitals and the majority of public health services. The remaining public fund is used for national services, such as disability support services and public health services. There are currently 20 District health boards responsible for providing or funding the provision of health services in their district.

Following a Health & Disability System Review, some changes to this structure are anticipated to increase collaboration and service cohesion. In the review, many recommendations supported a new entity, called Health NZ, to take control of the health system, including contracts, funding, and service leadership once held by the Ministry of Health. There would also be a provisionally named Māori Health Authority for the care of the Māori community.

Some notable experts and centers are specializing in rare diseases in the country.
The Pharmaceutical Management Agency (Pharmac) decides on behalf of Health New Zealand (Te Whatu Ora) which medicines and pharmaceutical products will be subsidized. The Pharmac Rare Disorders Advisory Committee evaluates funding applications and makes recommendations to the Pharmacology and Therapeutics Advisory Committee (PTAC) and Pharmac.

CENTERS
- Genetic Health Service NZ (North, Central, South Hubs)
- Auckland Network for filaminopathies, periventricular nodular heterotopia, biliary atresia
- ‘Starship center’ in Auckland is known for pediatrics but is not connected with other hospitals

Rare Disease Patient Organization Landscape
NATIONAL GROUP: Rare Disorders NZ

https://systemreview.health.govt.nz/
PHILIPPINES

Rare Disease Policy Landscape
Overall, in the Philippines, there is a high level of government support for rare diseases with the Rare Disease Act of the Philippines approved September 2015. The Act covers patients’ care, registry, research and development, information sharing and screenings. The Act categorizes persons living with a rare disease as “persons with disability” allowing patients to benefit from benefits provided by the Act 9442 including discounts on healthcare and medicines.

Rare Disease Health System Landscape
In the Philippines, the healthcare system is a dual public and private sector organized across seven regions. The Philippines has managed to connect a newborn screening network with frontline services based in over 7000 islands; this service is covered by national insurance since 2019.
The national newborn screening program is set up in the 7000+ newborn screening facilities in the country through the medical centers (hospitals and birthing centers). Newborn screening samples are sent to seven regional laboratories called Newborn Screening Centers (NSC).
The NSC has a laboratory and a short-term follow-up component. Confirmed patients are sent to a specialist within the networks of 14 regional specialist clinics, which are linked to molecular labs. The specialist centers are connected to a global network of experts.

NETWORKS & CENTERS
Newborn screening clinics in all the islands are connected to the main genetic services in the University Hospital in Manila. The newborn screening network is a mature network connecting all hospitals across the country.

CENTERS
- Institute of Human Genetics (IHG), National Institutes of Health, University of the Philippines Manila in collaboration with Taiwan and Australia for diagnostics
- Center for Human Genetic Services - satellite of IHG, NIH (Visayas)
- Center for Human Genetic Services - Satellite of the IHG, NIH (Cebu)
- Center for Human Genetic Services - satellite of IHG, NIH (Mindanao)
- Center for Human Genetic Services, a satellite of the IHG, NIH - In Progress (Davao)

Rare Disease Patient Organization Landscape
DEBRA Singapore has been helping EB (Epidermolysis bullosa) patients and their families and healthcare providers in Philippines to set up DEBRA support groups and increase access to diagnosis, treatment, innovation, and care.

RD Definition:
Less than 1 in 20,000
Rare Disease Act 2015

RD Definition:
Less than 1 in 20,000
Rare Disease Act 2015

NATIONAL GROUP
Philippine Society for Orphan Disorders

REPUBLIC OF KOREA

Rare Disease Policy Landscape
Following the implementation of the Rare Disease Management Act (RDMA) of 2015, the ‘First National Plan for Rare Disease Management (2017-2021)’ was established to define a legal standard and manage rare diseases.
Article 14 of RDMA calls for registration of rare diseases nationally.
National Health Insurance Scheme, the Medical Expense Support Project, and Support for Catastrophic Health Expenditure are programs supporting persons living with a rare disease.

Rare Disease Health System Landscape
The Korea Disease Control and Prevention Agency designated 17 institutions as rare disease specialist institutions in each region. Rare disease specialized institutions are medical institutions that treat patients with rare diseases, research rare disease management, and register rare diseases in national statistics projects. The National Bio Big Data Project on Rare Diseases supports families with diagnosis. Patients are recruited from 17 rare disease specialist institutions, and their genetic samples are fed into a centralized sequencing platform and bioinformatics pipeline.

NETWORKS & CENTERS
As a part of the Korean Undiagnosed Diseases Program (KUDP), the designated regional rare disease centers in each province establish a nationwide rare disease network.
South Korea collaborates with centers in Japan and Malaysia; a large number of patients from Malaysia are referred to the Korean Genetic Lab.

CENTERS
- Seoul National University Hospital as the central support center
- Inha University Hospital
- Ajou University Hospital
- Chungnam National University Hospital
- Chungbuk National University Hospital
- Chilgok Kyungpook National University Medical Center
- Inje University Busan Paik Hospital
- Pusan National University Yangsan Hospital
- Chonnam National University Hwasun Hospital
- Chonbuk National University Hospital
- Cheju Halla General Hospital

RD Definition: Fewer than 20,000

Rare Disease Policy Landscape

The Singapore government established a national multi-stakeholder charity Rare Disease Fund. Initially, the Fund covered only four diseases, but it is now open to considering other diseases.

Rare Disease Health System Landscape

In Singapore, public-based national health insurance is based on a person’s income level and investment into individual personal healthcare plans. However, people can still incur high out of pocket payments, because patients can only use their personal healthcare insurance on what the government’s healthcare policy designate, which does not include rare disease care.

Networks & Centers

The country has two large children’s hospitals that include specialists and experts in rare diseases: National University Hospital System (NUHS), KK Women and Children’s Hospital (KKH). These centers already collaborate with each other and run an Undiagnosed Disease Program.

The Undiagnosed Disease Program was set up in 2014 to further aid diagnostics in patients with rare diseases through next-generation sequencing. SingHealth Duke-NUS Genomic Medicine Center is a genome clinic, set up in 2019 across five hospitals in Singapore in partnership with Duke Medical School. The Center provides an undiagnosed disease program to adult patients.

The Cancer Genetics Service at National Cancer Center Singapore was formally established in 2014. While this service primarily sees families with hereditary cancer syndromes, it also provides genetic testing for other hereditary conditions and rare diseases due to limited general genetics services in Singapore. Some genetic services utilize funds raised through philanthropy donations to cover genetic testing costs for patients who have financial needs.

With the help of patient alliance DEBRA International, immunofluorescence mapping and genetic testing are made available for EB (Epidermolysis bullosa) patients by linking doctors and other healthcare providers from other countries to research laboratories in Singapore. Likewise, doctors and nurses from KKH, NUH, and the National Skin Center provide training to doctors, nurses, and other healthcare providers from other countries in diagnosing and management of EB patients.

Centers

- KK Women’s and Children’s Hospital in collaboration with Australia, Malaysia, UK, USA
- National University Health System
- SingHealth Duke-NUS Genomic Medicine Center in collaboration with Australia, Malaysia, UK, USA
- National Cancer Center Singapore

Rare Disease Patient Organization Landscape

National Group: Rare Disorders Society (Singapore)
Rare Disease Policy Landscape

Taiwan has a legal definition for a rare disease, which is a prevalence of one in 10,000, as well as being classified as preventable and challenging to diagnose and treat.

The Rare Disease and Orphan Drug Act (2000) provides the legal basis for patients’ rights to access a defined group of treatments and services, including genetic testing subsidies, psychological and nutrition consultation fees, and subsidies for medical devices and some drugs.

Taiwan has an Orphan Drugs Committee that designates orphan drugs and a Medical Committee that reviews the list of rare diseases to be nationally subsidized. There are currently 223 diseases with 17,742 patients registered, and 73 different orphan drugs for 44 diseases.

After the implementation of the second generation of health insurance in 2013, the orphan drug budget was crowded out, contributing to only six new drugs for rare diseases approved and reimbursed in the past five years.

PLWRD also have rights under the Disabilities Rights Protection Act, specifically access to special education, employment, social welfare funds, transportation support, and tax reduction.

Rare Disease Health System Landscape

The National Health Insurance (NHI) covers medical service expenses for rare disease patients and reimbursement of approved orphan drugs.

Health Promotion Administration (HPA) set up a restrictive subsidized program for expenses that could not be reimbursed by NHI, such as research funds, genetic testing fees, medical device supplements, special formula, and other medical expenses.

A newborn screening program started in 1985, covering five diseases, which expanded to cover 21 diseases in 2019.

Networks & Centers

Taiwan has 14 Rare Disease Care Centers, each center with its own genetic counselling center. These centers collaborate amongst themselves.

Centers in Taiwan provide genetic testing for Hong Kong, Singapore, Malaysia, and Indonesia, for LSD.

Rare Disease Patient Organization Landscape

Taiwan has a strong patient advocacy group and good international collaboration with other patient groups. Through this networking, they have provided their experiences to share knowledge and build the capacity of other groups in the region.