

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.



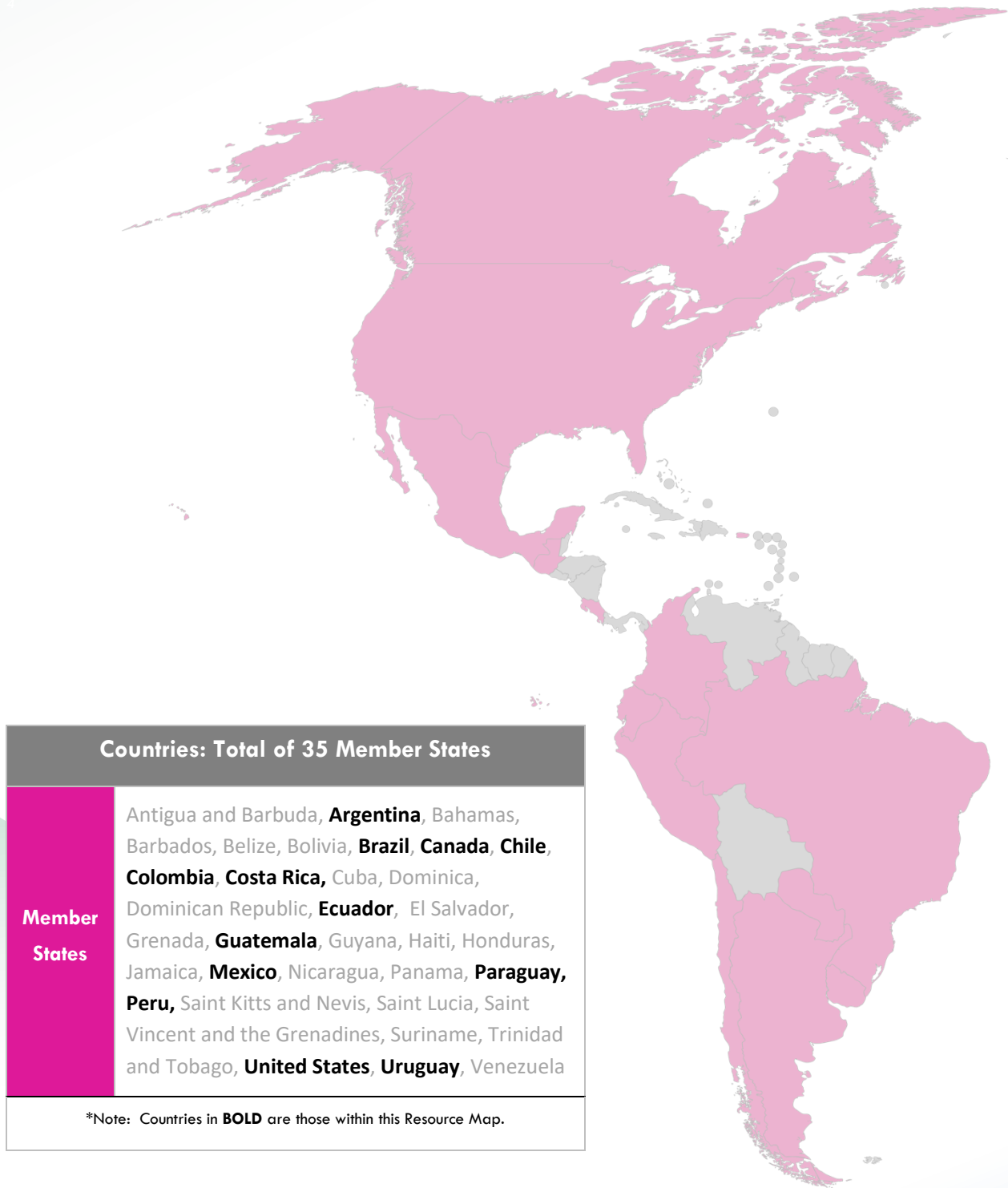
| | Population size (2020) In Millions ² | Estimated rare disease population prevalence (3.5-5.9%) in Millions ³ |
|------------------------------|--|---|
| 1. African Region | 1,036.1 | 35.8 - 60.4 |
| 2. Region of the Americas | 1,018.1 | 35.6 - 60.1 |
| 3. East Mediterranean Region | 730.8 | 25.6 - 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>

REGION OF THE AMERICAS (AMR)




⁴ Image created with mapchart.net




AMR Rare Disease Landscape Overview

As reported by patient leaders and medical experts from 20 countries in the region

Rare Disease Policy Landscape


-  Across the regions, there have been several developments in the recognition of rare diseases and the legal frameworks to support access to diagnosis, treatment, and care for persons living with a rare disease (Argentina, Brazil, Canada, Chile, Colombia, Ecuador, Guatemala, Mexico, Peru, United States). However, public healthcare system strength, lack of resources and national action plans are a challenge to implementation.

Rare Disease Health System Landscape



-  North America generally has highly competent clinical services and some national reference centers officially recognized and registered. In Central America, countries provide some coverage and treatment can be secured under national healthcare for certain rare diseases. However, most healthcare systems in this sub-region are weak and cannot respond to the full needs of the population. In South America, there are pockets of expertise and existing rare disease healthcare is organized around individual experts or genetic services, there are some designated Centers of Excellence in a few countries (Argentina, Brazil).
-  Across the region, most expertise is concentrated in capital and major cities, whereas a large portion of the population live in rural areas. This creates a barrier for rural communities to access expertise.
-  There are public and private newborn screening programs across the region but these vary highly in number of conditions covered within and across countries.

Both the U.S. and Canada carry out significant research and innovation and share an entrepreneurial approach, resulting in an environment with a high capacity for clinical trials and fosters high-quality research programs.

Rare Disease Patient Organization Landscape

-  North American countries have strong and well-organized patient communities led by national patient groups. In South America, the rare disease patient community is active in supporting both policy and patient rights and is supported by regional alliances. In Central America there are legislative and administrative barriers that prevent the formation of patient associations, however informal patient communities exist.

REGIONAL ALLIANCES (Central & South America)

-  Alianza Iberoamericana de Enfermedades Raras (ALIBER)
-  Enfermedades raras del Caribe y América Latina (ERCAL)

ARGENTINA

Rare Disease Policy Landscape

- Argentina's constitution outlines access to healthcare as a constitutional right and is widely represented in the form of laws, decrees, and human rights treaties ratified.
- Argentina Law 26.689 (2011)⁵ is a legal framework for the "Comprehensive Healthcare of People with Diseases of Low Prevalence"; it mandates national patient- and disease-specific registries, centers of excellence, interdisciplinary care, expanded newborn screening programs, and research and development.
- Resolution 2329 (2014), Decree 794 (2015), Resolution 271 (2019), Regulation 641 (2021) further create and regulate programs for rare diseases, including establishing a national advisory board and patient registries, reviewing existing care infrastructure and approving a list of rare diseases.⁵

- RD Definition: fewer than 1 in 2,000
- Low Prevalence Diseases Law, 2011

Rare Disease Health System Landscape

- The public health system is set up to provide free coverage for public care; specialized services and treatment available to citizens vary depending on provincial jurisdiction in which the patient resides. Sometimes, citizens have to travel outside of Argentina for care.

NETWORKS & CENTERS

- There are centers of reference focusing on rare diseases, pediatric conditions and transnational medicine. Others are specialized by pathology, for example hemophilia.
- There are 20 regional newborn screening programs with varying coverage of disorders.⁶

CENTERS

- Centro de Referencia en Enfermedades Raras y de Dificultoso Diagnóstico (CERyD) (B.Aires)
- Hospital El Cruce (Buenos Aires)
- Hospital Garrahan (Buenos Aires)
- Hospital Universitario Austral (Buenos Aires)
- Unidad de Metabolismo-Hospital de Niños Sor María Ludovica (Buenos Aires)
- Consultorio de Enfermedades Poco Frecuentes CEMAFE (Santa Fe)
- Hospital del Niño Jesús (Tucuman)
- Hospital Humberto Notti (Mendoza)

Rare Disease Patient Organization Landscape

NATIONAL GROUPS:

- Federación Argentina de Enfermedades Poco Frecuentes (FADEPOF)
- Allianza Argentina de Pacientes (ALAPA)

⁵ Mayrides, Mo & Castilla, Eva & Szelepsi, Silvina. (2020). A civil society view of rare disease public policy in six Latin American countries. Orphanet Journal of Rare Diseases. 15. 10.1186/s13023-020-1314-z.

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

⁶ Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. Front. Genet. 13:1053559. doi: 10.3389/fgene.2022.1053559

BRAZIL

Rare Disease Policy Landscape

- Brazil's Ordinance 199 (2014) instituted the National Policy for Comprehensive Care for People with Rare Diseases⁷ which has been important in enabling the conditions for the promotion of rare diseases in the country. The act calls for greater public awareness, improving diagnosis, increasing education and training among health professionals, expanding reference centers, and developing clinical protocols for prioritized rare diseases.

- RD Definition: fewer than 65 in 100,000
- National Policy for Rare Diseases, 2014

Rare Disease Health System Landscape

- Since 1988, the Sistema Único de Saúde (Unique Health System, SUS) provides free universal health care coverage to all people living in Brazil. Ordinance 199 established Guidelines for Comprehensive Care for People with Rare Diseases under the SUS. However, due to inadequate funding, the healthcare system suffers from shortages in many areas. About 20% of the Brazilian population opt for private health care.

NETWORKS & CENTERS

- The National Policy for Rare Diseases (2014) designated 10 Reference Centers for Rare Diseases (RCRD). The RCRD model is now being expanded into Centers for Comprehensive Care & Training in Rare Diseases (Casa dos Raros) and connected under a National Network for Rare Diseases.
- Rare Disease experts in the region are working to have one Casa dos Raros Center designated in each of the 5 regions of Brazil, forming a Regional Hub for rare diseases, to improve equitable access for all people living with a rare disease in Brazil.
- Brazil started its nationwide NBS program in 2001 with reference centers covering more than 80% of newborns; in 2021 a law passed to expand scope of the newborn screening panel⁸

CENTERS

- Hospital de Clínicas de Porto Alegre (Porto Alegre)
- Casa dos Raros (House of Rares) (Porto Alegre)
- Casa dos Raros (House of Rares) (São Paulo)

Rare Disease Patient Organization Landscape

NATIONAL GROUPS:

- Instituto Vidas Raras
- Casa Hunter
- Associação Brasileira de Enfermedades Raras (FEBER)

⁷ http://bvsmis.saude.gov.br/bvs/saudelegis/gm/2014/prt0199_30_01_2014.html; http://www.scielo.org.ar/pdf/sc/v16/en_1851-8265-sc-16-e2210.pdf

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

⁸ Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. Front. Genet. 13:1053559. doi: 10.3389/fgene.2022.1053559

CANADA

Rare Disease Policy Landscape

- Launched in 2024, Canada's Rare Disease Strategy integrates three strategic components the Canadian Rare Disease Network (CRDN), the National Strategy for Drugs for Rare Diseases and an empowered patient community.⁹
- In 2019, the Canadian government committed to set up a National Strategy for Drugs for Rare Diseases¹⁰ with \$1 billion (CAD) over two years (2022-2023) and an additional \$500 million to accelerate access to new and emerging rare disease therapies, to promote equitable access to existing therapies, and to support early diagnosis and screening.
- Canada's Ontario and Quebec provinces have endorsed and consulted on provincial Rare Disease Strategies or Plans

● RD Definition: fewer than 5 in 10,000

● National Rare Diseases Strategy, 2024

Rare Disease Health System Landscape

- Canada possesses a public universal healthcare system which is funded and administered by its ten provinces and three territories. These provinces must adhere to the common principles outlined in the Canada Health Act.
- While there are no formal agreements for cross-provincial care, funding, and support, "out-of-province" care and treatment occur regularly. Provinces have "discretionary" authority to pay for the provision of "necessary" medical services in other provinces or countries (often the USA) when not available in the home province; approved on a case-by-case basis.
- There is a history of collaboration in the rare disease care and research space in Canada as well as collaboration between expert centers (Canada) and centers of excellence (USA). Collaboration also exists in the realm of research programs, such as drug research and development programs, and hospital-based innovative research and treatment programs.

NETWORKS & CENTERS

- The newly launched CRDN (2024) is a collaboration among healthcare professions, patient groups, and research institutes with the aim of addressing the complex challenges experienced by rare disease patients and families across Canada.
- Canada has excellent children's hospitals and specialist centers in the major cities. Centers of Expertise in Canada exist for some disease areas and have been accredited or validated by external bodies, e.g., in hemophilia, cystic fibrosis, muscular dystrophy, ALS, etc. No formal national accreditation of 'Rare Disease Center of Excellence' exists.
- Formal and informal networking and collaboration exist among the provinces, namely in specialty programs, research, and treatment initiatives such as Canada's Maternal Infant, and Child Research Network which includes all the children's hospitals where most rare disease centers are located. Networks of specialists are established to deliver care, drug therapy, and develop consensus standards.

⁹ <https://www.rarediseasesinternational.org/mappingrare/>

¹⁰ <https://www.canada.ca/en/health-canada/programs/consultation-national-strategy-high-cost-drugs-rare-diseases-online-engagement/what-we-heard.html>

- A newly established pilot genomic sequencing program supports diagnosis of ultra-rare and undiagnosed conditions through a network of five centers across Canada

CENTERS

- Hemophilia Treatment Centers Network (Canada)
- Canadian Hereditary Metabolic Treatment Centers (Canada)
- Neuromuscular Disease Network for Canada (Canada)
- Maternal Infant Child Youth Research Network (MICYRN) (Canada)
- Pediatric Bone Health Clinic (Canada)
- Prairie Metabolic Network (Canada)
- BC Children's Hospital (British Columbia)
- Alberta Children's Hospital in Calgary (Alberta)
- University of Saskatchewan – Department of Pediatrics (Saskatchewan)
- Children's Hospital of Manitoba (Manitoba)
- Children's Hospital of Eastern Ontario (Ontario)
- McMaster University – Department of Pediatrics (Ontario)
- Queen's University – Department of Pediatrics (Ontario)
- The Hospital for Sick Children (Ontario)
- Montreal Children's Hospital - McGill University (Quebec)
- CHU-Ste Justine - University of Montreal (Quebec)
- IWK Health Center (Nova Scotia)
- Memorial University (Newfoundland and Labrador)



Rare Disease Patient Organization Landscape



- Canada has a mature and well-organized patient community.
- Canadian Organization for Rare Disorders (CORD) is active in supporting both policy advocacy and patient rights. It leads the Rare Drugs Strategy in Canada which promotes the identification of expert centers and their formation into national networks.

NATIONAL GROUPS
Canadian Organization for
Rare Disorders (CORD)



CHILE

Rare Disease Policy Landscape


-  Law 19.966¹¹ in 2005 introduced a Universal System with Explicit Guarantees in Health (GES) to assure timely access to quality healthcare services and financial protection for a prioritized set of programs, diseases and health conditions based on prevalence, severity, cost, and impact on quality of life; coverage of this law is particularly relevant for hemophilia and cystic fibrosis communities.
-  Law 20.850 or the “Ricarte Soto Law” (2015)¹², which was initially focused on rare diseases, defines a financial protection for high-cost health technologies. This law, which increasingly covers a growing number of diseases, indicates a notable increase national in rare disease awareness but these efforts do not cover all rare diseases.

-  RD Definition: fewer than 5 in 10,000
-  Ricarte Soto Law, 2015


Rare Disease Health System Landscape

-  The public health system is organized into public and private sectors covering about 95% of the population.¹⁰ Healthcare is delivered through Primary Care, Secondary Care, or specialists. Patients can see a specialist or seek secondary care if referred by a primary healthcare provider.
-  The public newborn screening program was approved in 1992 and was implemented stepwise in all 15 regions of the country by 1998. It currently screens for only two conditions.¹³



CENTERS

-  Clinica Las Condes (Santiago, Chile)

Rare Disease Patient Organization Landscape

-  Various associations focused on less frequent illnesses have joined together to work for the enforcement of the Ricarte Soto law and fight for illnesses not included to provide comprehensive coverage to the population. Efforts are also being made to include visceral disabilities in the Disability Act, relating to diseases whose limitations are not observed.

NATIONAL GROUPS:

-  Federación Chilena de Enfermedades Raras (FECHER)
-  Federación de Enfermdades Poco Frecuentes (FENPOF)

¹¹ Encina, G., Castillo-Laborde, C., Lecaros, J.A. *et al.* Rare diseases in Chile: challenges and recommendations in universal health coverage context. *Orphanet J Rare Dis* 14, 289 (2019). <https://doi.org/10.1186/s13023-019-1261-8>

¹² <https://www.leychile.cl/Navegar?idNorma=1078148>



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

¹³ Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. *Front. Genet.* 13:1053559. doi: 10.3389/fgene.2022.1053559

COLOMBIA

Rare Disease Policy Landscape











- Colombia has strong legislation supporting the interests of the rare disease community including laws, decrees, and resolutions.¹⁴
- Law 1392, 2010¹⁵, recognizes rare diseases as a national priority. The Act sets out measures to support the rare disease community, including developing a list of rare diseases to be updated every two years; in 2018, the list included 2190 conditions.¹⁶ The Act also calls for the reporting of individuals diagnosed with a rare disease.
- In 2011, Law 1438 narrowed the RD definition down to 1 in 5,000 individuals.¹⁷

 RD Definition: fewer than 1 in 5,000
 Orphan Disease Law, 2010

Rare Disease Health System Landscape

- Law 1392 provides for the creation of a network of centers of excellence or reference centers, but there are none yet designated as such. In 2018, Colombia published the 'Manual for Enabling Reference Centers for Diagnosis, Treatment, and Pharmacies for the Comprehensive Care of Orphan Diseases,' which outlines essential criteria to guarantee a base level of quality.
- Colombia established a national newborn screening program in 2000; by 2015 the program covered approximately 80% of newborns in the country.¹⁸

CENTERS

-  Mediacarte (Network) (Colombia)
-  National Institute of Inborn Errors of Metabolism (EIM)
-  Hospital Universitario San Jose (Bogotá)
-  Javeriana University (Bogotá)
-  Instituto Roosevelt (Bogotá) – reference center
-  Biochemistry Research Center of the Universidad de los Andes (Bogotá)
-  Fundación Valle de Lili (Cali)
-  Center for Research on Congenital Anomalies and Rare Diseases (CIACER), ICESI University (Cali)
-  Hospital San Vicente Fundación (Medellin)
-  Center for Genomic and Metabolism Medicine at the Cardiovascular Foundation (Bucaramanga)

Rare Disease Patient Organization Landscape

NATIONAL GROUPS

Federacion Colombiana De Enfermedades Raras (FECOER)

¹⁴https://www.researchgate.net/publication/339542197_A_civil_society_view_of_rare_disease_public_policy_in_six_Latin_American_countries

¹⁵<https://www.minsalud.gov.co/sites/rid/Lists/BibliotecaDigital/RIDE/DE/DIJ/ley-1392-de-2010.pdf>

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

¹⁶<https://www.minsalud.gov.co/English/Paginas/Colombia-Takes-on-the-Challenge-of-Comprehensive-Care-for-Orphan-Diseases.aspx>

¹⁷<https://icj2.wenginepowered.com/wp-content/uploads/2013/05/Colombia-Ley-1438-2011-spa.pdf>

¹⁸ Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. *Front. Genet.* 13:1053559. doi: 10.3389/fgene.2022.1053559

COSTA RICA

Rare Disease Policy Landscape

- Although there is no specific law on rare diseases, Costa Rica has healthcare legislation committing to universal health coverage (UHC), and implementation is strong
- Law of Equal Opportunities for Persons with Disabilities, 1996, covers the rights of some individuals living with a rare disease¹⁹

Rare Disease Health System Landscape

- Costa Rica's health systems is primarily focused on common diseases, although there are a couple of rare disease measures in place. Costa Rica has several institutes that deal directly or indirectly with genetic and some forms of rare diseases.

NETWORKS & CENTERS

- The National Network for Rare Diseases (Red Nacional de Enfermedades Raras) was launched in February 2020 with the support of Asociación Nacional Segunda Oportunidad de Vida (ANASOVI) and the national Pulmonary Hypertension patient group. The key objective of the Network is education and holistic care of people living with rare diseases.
- As of 2021, national newborn screening was performed for 51 conditions¹⁷

CENTERS

- Instituto de Investigaciones en Salud (INISA)
- Centro de Investigación en Biología Celular y Molecular (CIBCM)

¹⁹ http://ercal.icord.es/wp-content/uploads/2021/06/Dr-Carlos-Salazar-Vargas_Costa-Rica.pdf

ECUADOR

Rare Disease Policy Landscape

- In Ecuador, health and social care is organized under Health Law 67's five pillars: Registry, Coverage, Diagnosis, Education and Treatment.²⁰ A specific chapter covering the treatment of rare and catastrophic diseases was incorporated into Health Law 67 (O.R.625, 2012). Under the revised law, rare diseases are recognized as a national interest and the community catalogued as a double vulnerability population; this means priority in the management and care.
- Because there is no national registry, current regulation and Ministry of Health guidelines are poorly implemented, and there are budget constraints. Rare disease patients only have rights to the possibility of social assistance when they qualify for a disability card.

Rare Disease Health System Landscape

- There is a national list of covered diseases that include 106 rare conditions,²¹ however, there have been challenges in implementation and actual coverage is limited.
- There is no formal designation process or body with certification criteria to identify and recognize Rare Disease Centers of Expertise in the public health system.

²⁰ <https://aplicaciones.msp.gob.ec/salud/archivosdigitales/documentosDirecciones/dnn/archivos/REFORMA%20A%20LA%20LEY%20ORG%C3%81NICA%20DE%20SALUD%20PARA%20INCLUIR%20EL%20TRATAMIENTO%20DE%20LAS%20ENFERMEDADES%20RARAS,%20HU%C3%89RFANAS.pdf>

²¹ https://www.fomentoacademico.gob.ec/wp-content/uploads/2020/02/listado_de_enfermedades_catastro%CC%81ficas_hue%CC%81rfanas_o_raras..pdf

GUATEMALA

Rare Disease Policy Landscape

- The Law on Comprehensive Care for People with Suspected or Diagnosed Suffering from Rare Diseases, which advocates for holistic care of rare disease patients, is in the process of approval in Guatemala.²²

Rare Disease Health System Landscape

- Guatemala has a public and private health system. Many individuals are forced to use the private health system when the public health cannot respond to all the needs of the population.
- Healthcare is focused on hospitalization, the third level of health system coverage, with the lowest ratio of healthcare spending per capita (2 hospital beds per 1000) in the region.
- There is a private center that specializes in about 6-7 rare diseases.

CENTERS

- Instituto de Investigaciones Químicas, Biológicas, Biomédicas y Biofísicas UMG
- Instituto para la Investigación Científica y la Educación Acera de las Enfermedades Genéticas y Metabólicas Humanas (INVEGEM)

Rare Disease Patient Organization Landscape


- Patient groups active in the country include ANGEL is the Guatemalan National Association for lysosomal storage diseases, genetic, autosomal recessive conditions that are included in rare or infrequent diseases.

²² https://www.congreso.gob.gt/detalle_pdf/iniciativas/4559#gsc.tab=0

MEXICO

Rare Disease Policy Landscape

- Article 224 bis and bis 1 of the General Health Law (2012)²³ defines orphan medicines for rare diseases and mandates the nation's Secretary of Health to consider the necessary means to make orphan medicines available to the Mexican population.

 RD Definition:
fewer than 5 in 10,000

Rare Disease Health System Landscape

- In Mexico, the health system is diversified. Services are provided to workers through employers', workers', and state contributions; union benefits cover others; additional services cover the population without another type of health coverage.
- Mexico was the first country in Latin America to institute a newborn screening program in 1974; currently public and private institutions screen for between 4-70 conditions, at their discretion²⁴

NETWORKS & CENTERS

- There are pockets of experts across Mexico as well as Centers of Excellence and a center for RD diagnosis where rare disease cases can be referred for diagnosis, treatment, and follow-up.

CENTERS

- Institute for Social Security and Services for State Workers (ISSSTE) (Mexico City/Mexico)
- Hospital Juan I. Menchaca (Guadalajara)
- Hospital Civil Nuevo de Guadalajara (Guadalajara)
- UMAE Pediatría Centro Médico Nacional de Occidente (CMNO) (Guadalajara)
- Hospital Infantil de Mexico (Mexico City)
- Instituto Nacional de Pediatría (Mexico City)
- Centro Médico La Raza (Mexico City)
- Hospital Regional 45 (Nuevo Leon)

Rare Disease Patient Organization Landscape

- The Federación Mexicana de Enfermedades Raras (FEMEXER) is working with national stakeholders in Mexico towards the development of a rare disease national plan which will be submitted to the Mexican Ministry of Health for endorsement.

NATIONAL GROUPS

- Federación Mexicana de Enfermedades Raras (FEMEXER)
- Organización Mexicana de Enfermedades Raras (OMER)

²³ <http://www.ordenjuridico.gob.mx/Documentos/Federal/html/wo11037.html>

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

²⁴ Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. Front. Genet. 13:1053559. doi: 10.3389/fgene.2022.1053559

PARAGUAY

Rare Disease Policy Landscape

- Bill D-1122038 (2011) proposes the establishment of a free pension for people who are living with a rare or unusual diseases and that a national registry of rare diseases be created.²⁵
- Rare diseases are being made visible, in part through the National Program for the Prevention of Rare Diseases, a national registry and a newborn screening program for the identification of inborn errors of metabolism.

Rare Disease Health System Landscape

- The Ministry of Public Health and Social Welfare is responsible for the care of 70% of the population at the national level through a network of hospitals, centers, and health posts. The remaining 30% are served by the Social Welfare Institute, by Military and Police services, by private insurance, and by the Hospital de Clínicas, part of the National University of Asuncion.
- Rare diseases care is usually done at the Hospital de Clínicas and in Hospitals of the Ministry of Health; for the latter, care is concentrated in one or at most two hospitals, usually located near the capital and focuses on children.
- Paraguay has a public nationwide newborn screening program since 2004 that covers three disorders.²⁶

CENTERS




- Hospital de Clínicas



²⁵ <https://py.vlex.com/vid/proyecto-d-1122038-primer-896533426>

²⁶ Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. *Front. Genet.* 13:1053559. doi: 10.3389/fgene.2022.1053559




PERU

Rare Disease Policy Landscape





-  Law 29698 (2011)²⁷ “National Interest and Preferential Attention to the Treatment of People Suffering from Rare or Orphan Diseases” calls on the Ministry of Health (MoH) to adopt mechanisms to ensure orphan drugs are available to patients with rare diseases, develop a rare disease patient registry, build a national action plan, seek measures to improve diagnosis, and include rare diseases in medical education and training. The law considers rare diseases a priority for budgeting and resource allocation purposes.
-  The National Plan for Rare Diseases (2021-2024) aims to establish strategies to improve access to comprehensive care and improve quality of life.²⁸ by implementing data management systems, promoting education and training, and fostering research on rare diseases.
-  The country established an official rare diseases list that recognizes more than 400 diseases

 RD Definition:
fewer than 1 in 100,000
 Rare Disease Law, 2011





Rare Disease Health System Landscape

-  In Peru, the Ministry of Health (MINSa) is in charge of the national health service. MINSa directly influences the comprehensive health insurance organism Seguro Integral de Salud (SIS).
-  In the healthcare system, rare diseases do not have an allocated budget, nor do they have comprehensive social and health care, but there is some coverage. The Peruvian state covers the high cost of rare disease healthcare for those citizens affiliated to the SIS through the Intangible Solidarity Health Fund (FISSAL).
-  Healthcare for rare diseases is organized around individual experts or genetic services. There is no formal designation process or body with certification criteria to identify and recognize Rare Disease Centers of Expertise in the public health system.

Rare Disease Patient Organization Landscape

-  Umbrella organizations in Peru work for the welfare of rare disease patients
-  FEPER works for the recognition and fulfilment of the rights of all patients with rare diseases, including the enactment of the rare disease law.
-  COPEPOFRE presented a proposed budget or rare diseases to the MoH, seeking to secure funding based on scientific evidence.
-  FEPECROH promotes expanded neonatal screening and provides support to children in palliative care.

NATIONAL GROUPS

-  Esperantra
-  Peruvian Federation of Rare Diseases (FEPER)
-  Peruvian Coalition for Uncommon Diseases (COPEPOFRE)
-  Paediatric Federation of Chronic and Orphan Diseases (FEPECROH)

²⁷ <https://busquedas.elperuano.pe/dispositivo/NL/1743601-5>

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

²⁸ Andina A. Peru: Minsa approves national plan for rare or orphan diseases, (2020). <https://andina.pe/ingles/noticia-peru-minsa-approves-national-plan-for-rare-or-orphan-diseases-827084.aspx>.

UNITED STATES OF AMERICA

Rare Disease Policy Landscape

- The Orphan Drug Act of 1983 establishes incentives for the development of drugs for rare diseases. By 2019, the US had granted 838 orphan indications for 564 distinct drugs.²⁹

- RD Definition:
Fewer than 200,000 people
- Orphan Drug Act of 1983

Rare Disease Health System Landscape

- The healthcare system is mainly private, regulated both at the national and state level. Joint federal-state programs exist to support the elderly or disabled (Medicare), low income (Medicaid), and children (CHIP), along with veterans, military, and Native Americans.
- Most US citizens receive healthcare from their employer through 'sponsored plans' in the health insurance marketplace. These plans usually have a high deductible (annual contribution) and monthly premium payment along with a cost-sharing structure. Additionally, each insurance plan only covers select physicians so patients are restricted when seeking care or may incur high out-of-pocket cost.
- There are no universal guidelines for diagnostic or treatment protocols for rare disease patients unless patient organizations have developed standards for their disease areas.

NETWORKS & CENTERS




- While there are "expert care centers" across the country for specific diseases or groups of diseases, many of these centers are either self-identified (meaning the individual center promotes itself as having particular expertise) or externally accredited by the patient community.
- The National Institutes of Health (NIH) developed an Undiagnosed Disease Program (NIH-UDP) and connected the center in a national Undiagnosed Disease Network (NIH-UDN), which extends across the USA to ensure national coverage. While the NIH-UDN exists as a system/network, it only accepts the most challenging cases, meaning that most patients looking for a diagnosis will not qualify for the NIH-UDN. Patients who are already diagnosed cannot seek ongoing expertise and care through the NIH-UDN.

²⁹ <https://rarediseases.org/wp-content/uploads/2021/03/orphan-drugs-in-the-united-states-NRD-2020.pdf>

CENTERS

-  Rare Diseases Clinical Research Network (RDCRN) (USA)
-  Undiagnosed Diseases Network (USA)
-  The Mass General Center for Rare Neurological Diseases (CRND) (Boston, MA)
-  The Broad Institute of MIT and Harvard (Boston, MA)
-  Institute for Rare Diseases Research at UMass Medical School (MA)
-  Precision Medicine Network – Columbia University (New York, NY)
-  The Very Rare Cancer Consortium (VRCC) (NY)
-  The Center for Rare Disease Therapy at UPMC Children's Hospital of Pittsburgh (Pittsburgh, PA)
-  Kennedy Krieger Institute (Baltimore, MD)
-  Rare Disease Institute at Children's National (Washington DC, USA)
-  Cleveland Clinic Cancer Center (Cleveland, OH)
-  The Rare Genetic Disease Program at Cincinnati Children's (Cincinnati, OH)
-  Children's Wisconsin (Milwaukee, WI)
-  The California Center for Rare Diseases at UCLA (Los Angeles, CA)
-  Rare Genomics Institute (Los Angeles, CA)
-  The Translational Genomics Research Institute (Phoenix, AZ)
-  UF Health Center for Pediatric Neuromuscular and Rare Diseases (Gainesville, FL)

Rare Disease Patient Organization Landscape

-  The United States has a strong and well-organized patient community.
-  Among the groups leading national advocacy efforts, National Organization for Rare Disorders (NORD) supports patients' access to care. Among its activities, NORD has designated 40 medical institutions across the United States as NORD Rare Disease Centers of Excellence (NORD RD CoE's)³⁰ given their exceptional programs for patients with rare diseases. NORD plans to connect these Rare Disease Centers of Excellence under a national network and developed databases to track expertise in a given disease-state or area. Some NORD RD CoE's have connections with international groups, but each center varies in terms of collaboration with other centers, nationally and globally.
-  There are other accreditation and designation programs managed by disease-specific patient groups such as hemophilia, cystic fibrosis, and muscular dystrophy.

NATIONAL GROUPS
National Organization for Rare
Disorders (NORD)

³⁰ <https://rarediseases.org/center-of-excellence/>

URUGUAY

Rare Disease Policy Landscape

- In 2017, a bill to create a National Program for Rare Diseases was proposed but it has not yet been signed into law.³¹ Uruguay has a national program for rare diseases

Rare Disease Health System Landscape

- In Uruguay, each patient has a health care provider either through private mutual insurance companies or the public health care system. The only exception to this is police and military personnel, who have their own service. Psychological and social worker attention is provided as individuals enter into the Transplant Program waiting list. Patients who do not have stable income or resources are automatically referred to the public health system, and the Ministry of Economy and Finance covers the cost of treatment.
- Patients who do not have stable income or resources are automatically referred to the public health system, and the Ministry of Economy and Finance covers the cost of treatment.
- There are currently 28 disorders and 23 metabolic conditions that are screened conditions in Uruguay's healthcare system.
- Uruguay has specialist centers for rare disease or congenital defects.
- A national newborn screening program was implemented in Uruguay in 1994; mandatory newborn screening provides over 90% coverage of the population.³²

Rare Disease Patient Organization Landscape

NATIONAL GROUPS

Asociación Todos Unidos Enfermedades Raras Uruguay (ATUERU)

³¹ <https://parlamento.gub.uy/noticias/eventos/noticias/node/87607>

³² Giugliani R, et al. (2022), Opportunities and challenges for newborn screening and early diagnosis of rare diseases in Latin America. *Front. Genet.* 13:1053559. doi: 10.3389/fgene.2022.1053559