Selected Government Definitions of Orphan or Rare Diseases
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Table 1: Country Definitions of Orphan or Rare Diseases

Africa		
Africa: South Africa		No legislation on orphan drugs or definition of rare disease in South Africa
Asia		
Asia: China		No legislation on orphan drugs in China. They tried to come to an agreement on a definition of rare diseases but instead of a prevalence-based definition Chinese authorities published a list of rare diseases.
Asia: India	Ministry of Health and Family Welfare, Government of India formulated a National Policy for Treatment of Rare Diseases (NPTRD)	India faces the limitation of lack of epidemiological data to be able to define rare diseases in terms of prevalence or prevalence rate, which has been used by other countries. Until epidemiological data is available and the Country arrives at a definition of a rare disease based on prevalence data, the term rare diseases, for the purpose of this policy, shall construe the following groups of disorders identified and categorized by experts based on their clinical experience: Group 1: Disorders amenable to one time curative treatment: Group 2: Diseases requiring long term / lifelong treatment having relatively lower cost of treatment and benefit has been documented in literature and annual or more frequent surveillance is required Group 3: Diseases for which definitive treatment is available but challenges are to make optimal patient selection for benefit, very high cost and lifelong therapy
Asia: Indonesia		No legislation on orphan drugs or definition of rare diseases.

Region: Country	Legal Framework	Content of Law / Definition
Asia: Israel		There is no official definition of rare diseases in the legislation and regulations in Israel.
Asia: Japan	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	Article 77-2 (1) When the Minister of Health, Labour and Welfare receives an application from a person intending to market medical devices or regenerative medicine products that fall under both of the following items (including a person who manufactures products in a foreign country and exports them to Japan), they may designate pharmaceuticals, medical devices or regenerative medicine products pertaining to the application as orphan drugs, orphan medical devices or orphan regenerative medicine products by seeking the opinions of the Pharmaceutical Affairs and Food Sanitation Council: (i) the number of subjects pertaining to the usage does not reach the number specified by Order of the Ministry of Health, Labour and Welfare; (ii) with approval of marketing for the pharmaceuticals, medical devices or regenerative medicine product in application, those which will have particularly excellent value for usage. (2) When the Minister of Health, Labour and Welfare makes a designation under the preceding paragraph, they are to provide public notification thereof. Designation criteria by Ministry of Health, Labour and Welfare.[1] The Minister of Health, Labour and Welfare may designate drugs and medical devices satisfying the following criteria as orphan drugs/medical devices after receiving applications for orphan designation from the applicants. (1) Number of patients The number of patients who may use the drug or medical device should be less than 50 000 in Japan. (2) Medical needs The drugs or medical devices should be indicated for the treatment of serious diseases, including difficult-to-treat diseases. In addition, they must be drugs or medical devices for which there are high medical needs satisfying one of the following criteria.

[1] Ministry of Health, Labour and Welfare, https://www.mhlw.go.jp/english/policy/health-medical/pharmaceuticals/orphan_drug.html

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		(3) Possibility of development There should be a theoretical rationale for the use of the product for the target disease, and the development plan should be appropriate.
Asia: Pakistan		No orphan drug legislation or definition of rare diseases.
Asia: Singapore	Press Release, Singapore Ministry of Health and the SingHealth Fund have jointly established the Rare Disease Fund (RDF), July 2019[2]	Rare diseases are defined as affecting fewer than one in 2,000 people.
Asia: South Korea	Article 2 of the Rare Disease Management Act	Article 2. The meanings of terms used in this Act are as follows. 1. The term "rare disease" refers to a disease in which the prevalence population is less than 20,000 or the prevalence population is unknown due to difficulty in diagnosis, and is defined in accordance with the procedures and standards prescribed by Ordinance of the Ministry of Health and Welfare. 2. "Rare disease management" refers to all activities aimed at preventing, diagnosing, and treating rare diseases. [unofficial translation]
Oceania		
Oceania: Australia	16J of the Therapeutic Goods Regulations 1990	General criteria (3) The following criteria are specified in relation to a medicine that is not a new dosage form

[2] Ministry of Health, 2019, https://www.moh.gov.sg/news-highlights/details/rare-disease-fund-to-provide-financial-support-to-singaporeans-with-rare-diseases

Region: Country	Legal Framework	Content of Law / Definition
	Statutory Rules No. 394, 1990 made under the Therapeutic Goods Act 1989	medicine: (a) the application is for only one indication of the medicine; (b) the indication is the treatment, prevention or diagnosis of a life-threatening or seriously debilitating condition in a particular class of patients (the relevant patient class); (c) it is not medically plausible that the medicine could effectively treat, prevent or diagnose the condition in another class of patients that is not covered by the relevant patient class; (d) at least one of the following applies: (i) if the medicine is intended to treat the condition—the condition affects fewer than 5 in 10,000 individuals in Australia when the application is made; (ii) if the medicine is intended to prevent or diagnose the condition—the medicine, if it were included in the Register, would not be likely to be supplied to more than 5 in 10,000 individuals in Australia during each year that it is included in the Register; (iii) it is not likely to be financially viable for the sponsor to market the medicine in Australia unless each fee referred to in paragraph 45(12)(c) were waived in relation to the medicine; (e) none of the following has refused to approve the medicine for the treatment, prevention or diagnosis of the condition for a reason relating to the medicine's safety: (i) the Secretary; (ii) the United States Food and Drug Administration; (iii) the European Medicines Agency; (iv) Health Canada; (v) the Medicines and Healthcare products Regulatory Agency of the United Kingdom; (f) either: (i) no therapeutic goods that are intended to treat, prevent or diagnose the condition are included in the Register (except in the part of the Register for goods known as provisionally registered goods); or (ii) if one or more therapeutic goods that are intended to treat, prevent or diagnose the condition are included in the Register (except in the part of the Register for goods known as provisionally registered goods)—the medicine provides a significant benefit in relation to the efficacy or safety of the treatment, prevention or diagnosis of the condition, or a major contribution to patient care, compared to those goods.
Central American		

Region: Country	Legal Framework	Content of Law / Definition
Central America: Dominican Republic		No legislation on orphan drugs or definition of rare diseases.
Central America: Panama	Article 2 of the Law 28 of October 2014 That guarantees social protection to the population suffering from diseases rare, infrequent and orphan	Article 2. For the purposes of this Law, rare diseases shall be understood to mean little frequent and orphan those chronically debilitating or serious that threaten and life-threatening, which may be of genetic origin or of unknown causes and that require therapy specialized and permanent, with a prevalence of less than 1 in every 2,000 people. [unofficial translation]
Eurasia		
Eurasia: Russian Federation	Article 44 of the Federal law of 21.11.2011 No. 323-FZ On the basics of protecting the health of citizens in the Russian Federation	Article 44. Medical assistance to citizens suffering from rare (orphan) diseases 1. Rare (orphan) diseases are diseases that have the prevalence of no more than 10 cases of the disease per 100 thousand population.
Eurasia: Turkey		No legislation on orphan drugs or definition of rare diseases.
Europe		
Europe: European Union	Article 3 of the Regulation (EC) No 141/2000 of the European parliament and of the Council of 16 December 1999 on orphan medicinal	1. A medicinal product shall be designated as an orphan medicinal product if its sponsor can establish: (a) that it is intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting not more than five in 10 thousand persons in the Community when the application is made, or

Region: Country	Legal Framework	Content of Law / Definition
	products	that it is intended for the diagnosis, prevention or treatment of a life-threatening, seriously debilitating or serious and chronic condition in the Community and that without incentives it is unlikely that the marketing of the medicinal product in the Community would generate sufficient return to justify the necessary investment; and (b) that there exists no satisfactory method of diagnosis, prevention or treatment of the condition in question that has been authorised in the Community or, if such method exists, that the medicinal product will be of significant benefit to those affected by that condition.
Europe: Norway	Article 3 of the Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products	The Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products has been incorporated into the EEA agreement.[3] 1. A medicinal product shall be designated as an orphan medicinal product if its sponsor can establish: (a) that it is intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition affecting not more than five in 10 thousand persons in the Community when the application is made, or that it is intended for the diagnosis, prevention or treatment of a life-threatening, seriously debilitating or serious and chronic condition in the Community and that without incentives it is unlikely that the marketing of the medicinal product in the Community would generate sufficient return to justify the necessary investment; and (b) that there exists no satisfactory method of diagnosis, prevention or treatment of the condition in question that has been authorised in the Community or, if such method exists, that the medicinal product will be of significant benefit to those affected by that condition.
Europe: Switzerland	Article 4 of the 812.21 Federal Act of 15 December 2000 on	Art. 4 a Important medicinal products intended to treat rare diseases (orphan drugs) means medicinal products for human use for which it has been proven that: 1. they are indicated for the diagnosis, prevention or treatment of a life-threatening or

[3] Legal Act in Norwegian: EEA Suppl. No 12, 28.2.2002.,
https://www.efta.int/sites/default/files/documents/legal-texts/eea/other-legal-documents/solr/translated-legal-acts/norwegian/n32000R0141.pdf

Region: Country	Legal Framework	Content of Law / Definition
	Medicinal Products and Medical Devices (Therapeutic Products Act, TPA)	chronically debilitating disease affecting no more than five in ten thousand people in Switzerland when the application was submitted, or 2. they or their active substances are granted the status of Important medicinal products intended to treat rare diseases by another country with an equivalent system of medicinal product control within the meaning of Article 13
North America		
North America: Canada		No orphan drug legislation or definition of rare diseases in law
North America: Mexico	Article 224 Bis of the General Law of Health (2012)	Article 224 Bis: Orphan drugs: To drugs that are intended for the prevention, diagnosis or treatment of rare diseases, which have a prevalence of no more than 5 people per 10,000 inhabitants. [unofficial translation]
North America: United States	21 USC §360bb (a) 2: Designation of drugs for rare diseases or conditions	§360bb. (a) [...] (2) For purposes of paragraph (1), the term "rare disease or condition" means any disease or condition which (A) affects less than 200,000 persons in the United States, or (B) affects more than 200,000 in the United States and for which there is no reasonable expectation that the cost of developing and making available in the United States a drug for such disease or condition will be recovered from sales in the United States of such drug. Determinations under the preceding sentence with respect to any drug shall be made on the basis of the facts and circumstances as of the date the request for designation of the drug under this subsection is made.
South America		
South America:	Article 2 of the Law No. 26,689 Promote	Article 2 - For the purposes of this law, diseases of low prevalence are considered to be those whose prevalence in the population is equal to or less than one in two thousand (1 in 2000)

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Argentina	Comprehensive Healthcare for People with Diseases of Low Prevalence	people, referred to the national epidemiological situation. [unofficial translation]
South America: Brazil	Article. 3 I of the Ordinance No. 205 of December 28, 2017	Article. 3 For the purposes of this Resolution, the following definitions are adopted: I - rare disease: one that affects up to sixty-five people in every hundred thousand individuals, as defined by the National Policy for Comprehensive Care for People with Rare diseases, based on official national data or, when none exist, data published in technical-scientific documentation [unofficial translation]
South America: Chile		No specified definition of rare diseases in legislation. Proposed bill, initiated on a motion by the Honorable Senators, Quintana, Walker, Don Patricio, Chahuán, Rossi and Uriarte, on rare diseases. Within this proposed bill, the infrequent, minority, rare or orphan disease is defined as one with a danger of death or disability greater than 2/3 that has a prevalence of less than 5 cases per 10,000 inhabitants.[4]
South America: Colombia	Article 2 of the Law 1392 of 2010 2/9	Article 2. Denomination of Orphan Diseases. Orphan diseases are those chronically debilitating, serious, life threatening and with a prevalence of less than 1 in every 2,000 people, including rare, ultra-orphan and neglected diseases. [unofficial translation]
South America: Peru	Article 2.3 of the SUPREME DECRET NO. 004-2019-SA Regulation of Law N ° 29698, Law Declaring the Treatment of People Suffering Rare and Orphan Diseases of	Article 2.3 Rare Diseases (RD) - Are those diseases, with danger of death or chronic disability, that have a frequency, less than 1 per 100,000 inhabitants, in some cases they present many difficulties to be diagnosed and follow-up, they have a origin unknown in most cases, involve multiple social problems and have little epidemiological data. They can include congenital malformations and diseases of genetic origin. [unofficial translation]

[4] Boletín Nº 7.643-11

Region: Country	Legal Framework	Content of Law / Definition
	National Interest and Preferring Care	

Table 2: Patients per 100,000

Region: Country	Patient ratio	Fixed number of patients	Patients per 100,000
Africa: South Africa			
Asia			
Asia: China			
Asia: India			
Asia: Indonesia			
Asia: Israel			
Asia: Japan		50,000 patients	
Asia: Pakistan			
Asia: Singapore	1 in 2,000		50 in 100,000
Asia: South Korea		20,000 patients	39 in 100,000
Oceania			
Oceania: Australia	5 in 10,000		50 in 100,000
Central American			
Central America: Dominican Republic			
Central America: Panama	1 in 2,000		50 in 100,000

Region: Country	Patient ratio	Fixed number of patients	Patients per 100,000
Eurasia			
Asia: Russian Federation	10 in 100,000		10 in 100,000
Asia: Turkey			
Europe			
Europe: European Union	5 in 10,000		50 in 100,000
Europe: Norway	5 in 10,000		50 in 100,000
Europe: Switzerland	5 in 10,000		50 in 100,000
North America			
North America: Canada			
North America: Mexico	5 in 10,000		50 in 100,000
North America: United States		200,000 patients	61 in 100,000
South America			
South America: Argentina	1 in 2,000		50 in 100,000
South America: Brazil	65 in 100,000		65 in 100,000

Region: Country	Patient ratio	Fixed number of patients	Patients per 100,000
South America: Chile	5 in 10,000		50 in 100,000
South America: Colombia	1 in 2,000		50 in 100,000
South America: Peru	1 in 100,000		1 in 100,000

Annex: List of countries

- Africa
  - South Africa
- Asia
  - China
  - India
  - Indonesia
  - Israel
  - Japan
  - Pakistan
  - Singapore
  - South Korea
- Oceania
  - Australia
- Central America
  - Dominican Republic
  - Panama
- Europe
  - European Union
  - Norway
  - Switzerland
- Eurasia
  - Russian Federation
  - Turkey
- North America
  - Canada
  - Mexico
  - United States
- South America
  - Argentina
  - Brazil
  - Chile
  - Colombia
  - Peru