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China has officially released its first national list of rare diseases

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Summary **Over the past few years, the Chinese Government has paid greater attention to rare diseases and it has incorporated rare diseases in national health strategy and planning. On May 22, 2018, the Chinese Government officially released its first list of rare diseases, which included 121 rare diseases. The list was published to facilitate greater societal awareness of rare diseases, to improve the ability of front-line medical staff to treat rare diseases, to introduce incentives for research and development of orphan drugs, and to increase the availability of medicines for rare diseases. This effort will enhance the management of rare diseases in China, raise the level of diagnosis and treatment for rare diseases, and safeguard the health-related rights and interests of patients with rare diseases. The classification of rare diseases is based on a common international standards, which will promote international cooperation in drug research and policymaking with regard to rare diseases.**

Keywords: Rare diseases, disease classification, disease management, orphan drugs

On May 22, 2018, China's *First National List of Rare Diseases* (hereinafter referred to as the Chinese Rare Diseases List, CRDL) was issued jointly by five national bodies, including the National Health Commission, Ministry of Science and Technology, Ministry of Industry and Information Technology, State Drug Administration, and State Administration of Traditional Chinese Medicine (*1*). The CRDL gives priority to rare diseases with a relatively high prevalence, that pose a heavy burden, and that are highly treatable. The CRDL includes a total of 121 rare diseases (Table 1), such as albinism, amyotrophic lateral sclerosis, Gaucher's disease, Kallmann syndrome, Marfan syndrome, Fabry disease, and hemophilia.

The CRDL represents a clear "list of rare diseases" as mentioned in the *Opinions on Further Reform of the Review and Approval System to Encourage Innovation in Drugs and Medical Devices* issued by the former State Administration of Food and Drug Administration of China in October 2017 (*2*). Publication of the CRDL will help China enhance its management of rare diseases, improve the diagnosis and treatment of rare diseases, and safeguard the health-related rights of patients with rare diseases. The CRDL will serve as a reference for relevant government agencies and ministries in the future.

Thus far, rare diseases have not been officially defined in China. The definition in popular use was based on a consensus of experts reached by the Genetics Branch of the Chinese Medical Association in May 2010. According to this definition, a rare disease is a disease with a prevalence of less than 1/500,000 or a neonatal morbidity of less than 1/10,000 (*3*). Since epidemiological data on rare diseases are lacking in China, the current list of rare diseases is based on actual conditions, and the list is mainly derived from the professional opinions of the *Expert Committee on the Diagnosis, Treatment, and Care for Rare Diseases* established by the Medical Administration Bureau of the former National Health and Family Planning Commission.

As early as 2016, the Shanghai Health and Family Planning Commission published the first local list of rare diseases in China entitled *The List of Major Rare Diseases in Shanghai (2016 Edition)*. The Shanghai list which includes 56 rare diseases, 50 of which are also included in the CRDL (*4*). On September 23

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Table 1. The first national list of rare diseases in China*

No.	Diseases	No.	Diseases
1	21-Hydroxyulase Deficiency	60	Langerhans Cell Histiocytosis
2	Albinism	61	Laron Syndrome
3	Alport Syndrome	62	Leber Hereditary Optic Neuropathy
4	Amyotrophic Lateral Sclerosis	63	Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency
5	Angelman Syndrome	64	Lymphangioleiomyomatosis (LAM)
6	Arginase Deficiency	65	Lysine Urinary Protein Intolerance
7	Asphyxiating Thoracic Dystrophy (Jeune Syndrome)	66	Lysosomal Acid Lipase Deficiency
8	Atypical Hemolytic Uremic Syndrome	67	Maple Syrup Urine Disease
9	Autoimmune Encephalitis	68	Marfan Syndrome
10	Autoimmune Hypophysitis	69	McCune-Albright Syndrome
11	Autoimmune Insulin Receptopathy (Type B insulin resistance)	70	Medium Chain Acyl-CoA Dehydrogenase Deficiency
12	Beta-ketothiolase Deficiency	71	Methylmalonic Academia
13	Biotinidase Deficiency	72	Mitochondrial Encephalomyopathy
14	Cardic Ion Channelopathies	73	Mucopolysaccharidosis
15	Carnitine Deficiency	74	Multi-Focal Motor Neurothy
16	Castleman Disease	75	Multiple Acyl-CoA Dehydrogenase Deficiency
17	Charcot-Marie-Tooth Disease	76	Multiple Sclerosis
18	Citrullinemia	77	Multiple System Atrophy
19	Congenital Adrenal Hypoplasia	78	Myotonic Dystrophy
20	Congenital Hyperinsulinemic Hypoglycemia	79	NAGS Deficiency
21	Congenital Myasthenic Syndrome	80	Neonatal Diabetes Mellitus
22	Congenital Myotonia Syndrome (Non-Dystrophic Myotonia, NDM)	81	Neuromyelitis Optica
23	Congenital Scoliosis	82	Niemann-Pick Disease
24	Coronary Artery Ectasia	83	Non-Syndromic Deafness
25	Diamond-Blackfan Anemia	84	Noonan Syndrome
26	Erdheim -Chester Disease	85	Ornithine Transcarbamylase Deficiency
27	Fabry Disease	86	Osteogenesis Imperfecta (Brittle Bone Disease)
28	Familial Mediterranean Fever	87	Parkinson Disease (Young-onset, Early-onset)
29	Fanconi Anemia	88	Paroxysmal Nocturnal Hemoglobinuia
30	Galactosemia	89	Peutz-Jeghers Syndrome
31	Gaucher's Disease	90	Phenylketonuria
32	General Myathenic Gravis	91	POEMS Syndrome
33	Gitelman Syndrome	92	Porphyria
34	Glutaric Acidemia Type I	93	Prader-Willi Syndrome
35	Glycogen Storage Disease (Type I, II)	94	Primary Combined Immune Deficiency
36	Hemophilia	95	Primary Hereditary Dystonia
37	Hepatolenticular Degeneration (Wilson Disease)	96	Primary Light Chain Amyloidosis
38	Hereditary Angioedema (HAE)	97	Progressive Familial Intrahepatic Cholestasis
39	Hereditary Epidermolysis Bullosa	98	Progressive Muscular Dystrophies
40	Hereditary Fructose Intolerance	99	Propionic Acidemia
41	Hereditary Hypomagnesemia	100	Pulmonary Alveolar Proteinosis
42	Hereditary Multi-infarct Dementia (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalqpathy, CADASIL)	101	Pulmonary Cystic Fibrosis
		102	Retinitis Pigmentosa
		103	Retinoblastoma
43	Hereditary Spastic Paraplegia	104	Severe Congenital Neutropenia
44	Holocarboxylase Synthetase Deficiency	105	Severe Myoclonic Epilepsy In Infaricy (Dravet Syndrome)
45	Homocysteinemia	106	Sickle Cell Disease
46	Homozygous Hypercholesterolemia	107	Silver-Russell Syndrome
47	Huntington Disease	108	Sitosterolemia
48	Hyperornithinaemia-Hyperammonaemia-Hhomocit rullinuria Syndrome	109	Spinal and Bulbar Muscular Atrophy (Kennedy Disease)
		110	Spinal Muscular Atrophy
49	Hyperphenylalaninemia	111	Spinocerebellar Ataxia
50	Hypophosphatasia	112	Systemic Sclerosis
51	Hypophosphatemia Rickets	113	Tetrahydrobiopterin Deficiency
52	Idiopathic Cardiomyopathy	114	Tuberous Sclerosis Complex
53	Idiopathic Hypogonadotropic Hypogonadism	115	Tyrosinemia
54	Idiopathic Pulmonary Arterial Hypertension	116	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
55	Idiopathic Pulmonary Fibrosis	117	Williams Syndrome
56	IgG4 related Disease	118	Wiskott-Aldrich Syndrome
57	Inborn Errors of Bile Acid Synthesis	119	X-linked Agammaglobulinemia
58	Isovaleric Acidemia	120	X-linked Ldrenoleuko Dystrophy
59	Kallmann Syndrome	121	X-linked Lymphoproliferative Disease

*From the *Notice on the First National List of Rare Diseases in China* jointly issued by five bodies, including the National Health Commission. (*1*).

of the same year, a social organization, the Chinese Organization for Rare Disorders (CORD), published the *Reference List of Rare Diseases in China* (*5*). The list includes 147 rare diseases, 88 of which are also included in the CRDL.

Currently, there are no specific policies on rare diseases or orphan drugs nationwide, but the area of rare diseases has received increasing attention in recent years. The central government has included rare diseases in major health planning and strategy, including the *five-year plan on public healthcare (2016-2020)* (*6*) and the *"Healthy China 2030" Planning Outline* (*7*). Local government has actively promoted care for rare diseases. In Qingdao, a city in Shandong Province, medical insurance has covered Behcet syndrome, multiple sclerosis, and myasthenia gravis since 2005 (*8*). In Shanghai, the Children's Hospitalization Fund has covered Pompeii disease, Gaucher's disease, mucopolysaccharidosis, and Fabry disease since 2011 (*9*). In Zhejiang Province, medical insurance has covered Gaucher's disease, amyotrophic lateral sclerosis, and phenylketonuria since 2016 (*10*).

Issuance of the CRDL has resolved the issue of "no official definition, no specific policy support, and no coverage by medical insurance" of rare diseases in China. The list facilitates greater societal awareness of rare diseases, it improves the ability of medical personnel to diagnose and treat rare diseases, it furnishes incentives for research and development of orphan drugs, and it increases the affordability of orphan drugs. The classification of rare diseases is based on an international consensus, which is sure to promote international cooperation in clinical trials and healthcare policymaking in the area of rare diseases.

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