



# Catalog of Rare Diseases in China

Collected by ChemLinked BaiPharm Team

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## Introduction

On Sept. 20, 2023, China's National Health Commission (NHC) and five other authorities collaborated to release the Second Catalog of Rare Diseases, which comprises 86 rare diseases across various medical specialties including hematology, dermatology, rheumatology & immunology, pediatrics, neurology, and endocrinology.

This new catalog supplements the initial announcement made in 2018, expanding the number of recognized rare diseases in China to 207.

In line with China's emphasis on and support for the development of orphan drugs, medications designated for rare diseases in the catalog have a high likelihood of qualifying for [priority review](#) to expedite the process of obtaining marketing authorization. With priority review status, the technical review timeline for rare disease drugs will be reduced from the standard 200 workdays to 70 workdays, facilitating faster access to these treatments.

**Related:** [FAQ on Rare Disease Drugs \(Orphan Drugs\) in China](#)

**Table 1: China's First Catalog of Rare Diseases****(Issued 08/06/2023)**

No.	Rare disease (Chinese name)	Rare disease (English name)
1	21-羟化酶缺乏症	21-Hydroxylase Deficiency
2	白化病	Albinism
3	Alport 综合征	Alport Syndrome
4	肌萎缩侧索硬化	Amyotrophic Lateral Sclerosis
5	Angelman 氏症候群 (天使综合征)	Angelman Syndrome
6	精氨酸酶缺乏症	Arginase Deficiency
7	热纳综合征 (窒息性胸腔失养症)	Asphyxiating Thoracic Dystrophy (Jeune Syndrome)
8	非典型溶血性尿毒症	Atypical Hemolytic Uremic Syndrome
9	自身免疫性脑炎	Autoimmune Encephalitis
10	自身免疫性垂体炎	Autoimmune Hypophysitis
11	自身免疫性胰岛素受体病	Autoimmune Insulin Receptopathy (Type B insulin resistance)
12	$\beta$ -酮硫解酶缺乏症	Beta-ketothiolase Deficiency
13	生物素酶缺乏症	Biotinidase Deficiency
14	心脏离子通道病	Cardic Ion Channelopathies
15	原发性肉碱缺乏症	Carnitine Deficiency

16	Castleman 病	Castleman Disease
17	腓骨肌萎缩症	Charcot-Marie-Tooth Disease
18	瓜氨酸血症	Citrullinemia
19	先天性肾上腺发育不良	Congenital Adrenal Hypoplasia
20	先天性高胰岛素性低血糖血症	Congenital Hyperinsulinemic Hypoglycemia
21	先天性肌无力综合征	Congenital Myasthenic Syndrome
22	先天性肌强直（非营养不良性肌强直综合征）	Congenital Myotonia Syndrome (Non-Dystrophic Myotonia, NDM)
23	先天性脊柱侧弯	Congenital Scoliosis
24	冠状动脉扩张病	Coronary Artery Ectasia
25	先天性纯红细胞再生障碍性贫血	Diamond-Blackfan Anemia
26	Erdheim-Chester 病	Erdheim-Chester Disease
27	法布雷病	Fabry Disease
28	家族性地中海热	Familial Mediterranean Fever
29	范可尼贫血	Fanconi Anemia
30	半乳糖血症	Galactosemia
31	戈谢病	Gaucher's Disease
32	全身型重症肌无力	Generalized Myasthenia Gravis
33	Gitelman 综合征	Gitelman Syndrome
34	戊二酸血症 I 型	Glutaric Acidemia Type I

35	糖原累积病 (I 型、II 型)	Glycogen Storage Disease (Type I, Type II)
36	血友病	Hemophilia
37	肝豆状核变性	Hepatolenticular Degeneration (Wilson Disease)
38	遗传性血管性水肿	Hereditary Angioedema (HAE)
39	遗传性大疱性表皮松解症	Hereditary Epidermolysis Bullosa
40	遗传性果糖不耐受症	Hereditary Fructose Intolerance
41	遗传性低镁血症	Hereditary Hypomagnesemia
42	遗传性多发脑梗死性痴呆	Hereditary Multi-infarct Dementia (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, CADASIL)
43	遗传性痉挛性截瘫	Hereditary Spastic Paraplegia
44	全羧化酶合成酶缺乏症	Holocarboxylase Synthetase Deficiency
45	同型半胱氨酸血症	Homocysteinemia
46	纯合子家族性高胆固醇血症	Homozygous Hypercholesterolemia
47	亨廷顿舞蹈病	Huntington Disease
48	HHH 综合征	Hyperornithinaemia-Hyperammonaemia-Homocitrullinuria Syndrome
49	高苯丙氨酸血症	Hyperphenylalaninemia
50	低碱性磷酸酶血症	Hypophosphatasia
51	低磷性佝偻病	Hypophosphatemic Rickets

52	特发性心肌病	Idiopathic Cardiomyopathy
53	特发性低促性腺激素性性腺功能减退症	Idiopathic Hypogonadotropic Hypogonadism
54	特发性肺动脉高压	Idiopathic Pulmonary Arterial Hypertension
55	特发性肺纤维化	Idiopathic Pulmonary Fibrosis
56	IgG4 相关性疾病	IgG4 related Disease
57	先天性胆汁酸合成障碍	Inborn Errors of Bile Acid Synthesis
58	异戊酸血症	Isovaleric Acidemia
59	卡尔曼综合征	Kallmann Syndrome
60	朗格汉斯组织细胞增生症	Langerhans Cell Histiocytosis
61	莱伦氏综合征	Laron Syndrome
62	Leber 遗传性视神经病变	Leber Hereditary Optic Neuropathy
63	长链 3-羟酰基辅酶 A 脱氢酶缺乏症	Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency
64	淋巴管肌瘤病	Lymphangiomyomatosis (LAM)
65	赖氨酸尿蛋白不耐受症	Lysinuric Protein Intolerance
66	溶酶体酸性脂肪酶缺乏症	Lysosomal Acid Lipase Deficiency
67	枫糖尿症	Maple Syrup Urine Disease
68	马凡综合征	Marfan Syndrome
69	McCune-Albright 综合征	McCune-Albright Syndrome
70	中链酰基辅酶 A 脱氢酶缺乏症	Medium Chain Acyl-CoA Dehydrogenase Deficiency

71	甲基丙二酸血症	Methylmalonic Acidemia
72	线粒体脑肌病	Mitochondrial Encephalomyopathy
73	黏多糖贮积症	Mucopolysaccharidosis
74	多灶性运动神经病	Multifocal Motor Neuropathy
75	多种酰基辅酶 A 脱氢酶缺乏症	Multiple Acyl-CoA Dehydrogenase Deficiency
76	多发性硬化	Multiple Sclerosis
77	多系统萎缩	Multiple System Atrophy
78	肌强直性营养不良	Myotonic Dystrophy
79	N-乙酰谷氨酸合成酶缺乏症	N-acetylglutamate Synthase Deficiency
80	新生儿糖尿病	Neonatal Diabetes Mellitus
81	视神经脊髓炎	Neuromyelitis Optica
82	尼曼匹克病	Niemann-Pick Disease
83	非综合征性耳聋	Non-Syndromic Deafness
84	Noonan 综合征	Noonan Syndrome
85	鸟氨酸氨甲酰基转移酶缺乏症	Ornithine Transcarbamylase Deficiency
86	成骨不全症 (脆骨病)	Osteogenesis Imperfecta (Brittle Bone Disease)
87	帕金森病 (青年型、早发型)	Parkinson Disease (Young-onset, Early-onset)
88	阵发性睡眠性血红蛋白尿	Paroxysmal Nocturnal Hemoglobinuria
89	黑斑息肉综合征	Peutz-Jeghers Syndrome

90	苯丙酮尿症	Phenylketonuria
91	POEMS 综合征	POEMS Syndrome
92	卟啉病	Porphyria
93	Prader-Willi 综合征	Prader-Willi Syndrome
94	原发性联合免疫缺陷	Primary Combined Immune Deficiency
95	原发性遗传性肌张力不全	Primary Hereditary Dystonia
96	原发性轻链型淀粉样变	Primary Light Chain Amyloidosis
97	进行性家族性肝内胆汁淤积症	Progressive Familial Intrahepatic Cholestasis
98	进行性肌营养不良	Progressive Muscular Dystrophy
99	丙酸血症	Propionic Acidemia
100	肺泡蛋白沉积症	Pulmonary Alveolar Proteinosis
101	肺囊性纤维化	Pulmonary Cystic Fibrosis
102	视网膜色素变性	Retinitis Pigmentosa
103	视网膜母细胞瘤	Retinoblastoma
104	重症先天性粒细胞缺乏症	Severe Congenital Neutropenia
105	婴儿严重肌阵挛性癫痫(Dravet 综合征)	Severe Myoclonic Epilepsy in Infancy (Dravet Syndrome)
106	镰刀型细胞贫血病	Sickle Cell Disease
107	Silver-Russell 综合征	Silver-Russell Syndrome
108	谷固醇血症	Sitosterolemia

109	脊髓延髓肌萎缩症 (肯尼迪病)	Spinal and Bulbar Muscular Atrophy (Kennedy Disease)
110	脊髓性肌萎缩症	Spinal Muscular Atrophy
111	脊髓小脑性共济失调	Spinocerebellar Ataxia
112	系统性硬化症	Systemic Sclerosis
113	四氢生物蝶呤缺乏症	Tetrahydrobiopterin Deficiency
114	结节性硬化症	Tuberous Sclerosis Complex
115	原发性酪氨酸血症	Tyrosinemia
116	极长链酰基辅酶 A 脱氢酶缺乏症	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
117	威廉姆斯综合征	Williams Syndrome
118	湿疹血小板减少伴免疫缺陷综合征	Wiskott-Aldrich Syndrome
119	X-连锁无丙种球蛋白血症	X-linked Agammaglobulinemia
120	X-连锁肾上腺脑白质营养不良	X-linked Adrenoleukodystrophy
121	X-连锁淋巴增生症	X-linked Lymphoproliferative Disease

**Table 2: China's Second Catalog of Rare Diseases****(Issued 20/09/2023)**

No.	Rare disease (Chinese name)	Rare disease (English name)
1	软骨发育不全	Achondroplasia
2	获得性血友病	Acquired hemophilia
3	肢端肥大症	Acromegaly
4	成人斯蒂尔病	Adult-onset Still disease
5	Alagille 综合征	Alagille syndrome
6	$\alpha$ -1-抗胰蛋白酶缺乏症	Alpha-1-antitrypsin deficiency
7	ANCA 相关性血管炎	ANCA-associated vasculitis
8	Bardet-Biedl 综合征	Bardet-Biedl syndrome
9	白塞病/贝赫切特综合征	Behçet's disease
10	蓝色橡皮疱样痣	Blue rubber bleb nevus
11	CDKL5 缺乏症	CDKL5-deficiency disorder
12	无脉络膜症	Choroideremia
13	慢性炎性脱髓鞘性多发性神经根神经病	Chronic inflammatory demyelinating polyneuropathy
14	肾透明细胞肉瘤	Clear cell sarcoma of kidney
15	冷凝集素病	Cold agglutinin disease
16	先天性胆道闭锁	Congenital biliary atresia
17	先天性凝血因子 VII 缺乏症	Congenital factor VII deficiency
18	冷吡啉（冷炎素）相关周期性综合征/NLRP3 相关自身炎症性疾病	Cryopyrin associated periodic syndrome/ NLRP3-associated systemic autoinflammatory disease

19	皮肤神经内分泌癌（梅克尔细胞癌）	Cutaneous neuroendocrine carcinoma (Merkel cell carcinoma)
20	皮肤 T 细胞淋巴瘤	Cutaneous T-cell lymphomas
21	胱氨酸贮积症	Cystinosis
22	隆突性皮肤纤维肉瘤	Dermatofibrosarcoma protuberans
23	嗜酸性粒细胞性胃肠炎	Eosinophilic gastroenteritis
24	上皮样肉瘤	Epithelioid sarcoma
25	面肩肱型肌营养不良症	Facioscapulohumeral muscular dystrophy
26	家族性噬血细胞淋巴组织细胞增生症	Familial hemophagocytic lymphohistiocytosis
27	家族性腺瘤性息肉病	Familial adenomatous polyposis
28	进行性骨化性纤维发育不良	Fibrodysplasia ossificans progressiva
29	脆性 X 综合征	Fragile X syndrome
30	神经节苷脂贮积症	Gangliosidosis
31	胃肠胰神经内分泌肿瘤	Gastroenteropancreatic neuroendocrine neoplasm
32	胃肠间质瘤	Gastrointestinal stromal tumor
33	泛发性脓疱型银屑病	Generalized pustular psoriasis
34	遗传性甲状旁腺功能减退症	Genetic hypoparathyroidism
35	巨细胞动脉炎	Giant cell arteritis
36	骨巨细胞瘤	Giant cell tumor of bone
37	血小板无力症	Glanzmann thrombasthenia
38	胶质母细胞瘤	Glioblastoma
39	高林综合征	Gorlin syndrome
40	化脓性汗腺炎	Hidradenitis suppurativa
41	早老症	Hutchinson-Gilford progeria syndrome

42	炎性肌纤维母细胞瘤	Inflammatory myofibroblastic tumor
43	Leber 先天性黑矇	Leber congenital amaurosis
44	Lennox-Gastaut 综合征	Lennox-Gastaut syndrome
45	角膜缘干细胞缺乏症	Limbal stem cell deficiency
46	恶性高热	Malignant hyperthermia
47	恶性胸膜间皮瘤	Malignant pleural mesothelioma
48	黑色素瘤	Melanoma
49	异染性脑白质营养不良	Metachromatic leukodystrophy
50	单基因非综合征性肥胖	Monogenic non-syndromic obesity
51	多发性内分泌腺瘤病	Multiple endocrine neoplasia
52	发作性睡病	Narcolepsy
53	神经母细胞瘤	Neuroblastoma
54	神经纤维瘤病	Neurofibromatosis
55	神经元蜡样脂褐质沉积症	Neuronal ceroid lipofuscinosis
56	神经营养性角膜炎	Neurotrophic keratitis
57	骨肉瘤	Osteosarcoma
58	天疱疮	Pemphigus
59	新生儿持续肺动脉高压	Persistent pulmonary hypertension of the newborn
60	嗜铬细胞瘤	Pheochromocytoma
61	PIK3CA 相关过度生长综合征	PIK3CA related overgrowth syndrome
62	真性红细胞增多症	Polycythaemia vera
63	原发性胆汁性胆管炎	Primary biliary cholangitis
64	原发性生长激素缺乏症	Primary growth hormone deficiency
65	原发性胰岛素样生长因子-1 缺乏症	Primary IGF1 deficiency
66	原发性免疫缺陷	Primary immunodeficiency

67	原发性骨髓纤维化	Primary myelofibrosis
68	原发性硬化性胆管炎	Primary sclerosing cholangitis
69	进行性纤维化性间质性肺疾病	Progressive fibrosing interstitial lung disease
70	复发性心包炎	Recurrent pericarditis
71	早产儿视网膜病	Retinopathy of prematurity
72	Rett 综合征	Rett syndrome
73	短肠综合征	Short bowel syndrome
74	全身型幼年特发性关节炎	Systemic juvenile idiopathic arthritis
75	系统性肥大细胞增多症	Systemic mastocytosis
76	大动脉炎/多发性大动脉炎	Takayasu arteritis
77	腱鞘巨细胞瘤/色素沉着绒毛结节性滑膜炎	Tenosynovial giant cell tumor/Pigmented villonodular synovitis
78	地中海贫血（重型）	Thalassemia major
79	血栓性血小板减少性紫癜	Thrombotic thrombocytopenic purpura
80	转甲状腺素蛋白淀粉样变性	Transthyretin amyloidosis
81	肿瘤坏死因子受体相关周期性综合征	Tumor necrosis factor receptor associated periodic syndrome
82	肿瘤相关骨软化症	Tumor-induced osteomalacia
83	Von Hippel-Lindau 综合征	Von Hippel-Lindau syndrome
84	血管性血友病Ⅲ型	Von Willebrand disease type 3
85	华氏巨球蛋白血症/淋巴浆细胞淋巴瘤	Waldenström macroglobulinemia/ Lymphoplasmacytic lymphoma
86	West 综合征/婴儿痉挛综合征	West syndrome/Infantile spasms syndrome

## References

1. [The First Catalog of Rare Diseases in China](#)
2. [The Second Catalog of Rare Diseases in China](#)

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### Drug Application

Preparing and submitting drug applications, including clinical trial applications (CTA), new drug applications (NDAs), abbreviated new drug applications (ANDAs), and DMF Filing



### Local Agency Support

Serving as overseas drug marketing authorization holder (MAH)'s local agency in China, and fulfilling obligations throughout the entire lifecycle of the drug product



### Pharmacopoeia Translation

Translating Chinese Pharmacopoeia standards, covering chemical drugs, biological products, active ingredients, excipients, packaging materials, testing methods, and guidelines



### Pharmacovigilance

Offering pharmacovigilance services, including making safety & risk management plans, collecting, entering and evaluating data, following up safety cases, and preparing safety reports



### Consulting and Training

Interpreting regulatory requirements for NDA, ANDA, DMF filing, post-market change management, and GMP compliance



### Cross-border E-commerce

Establishing online stores to sell OTC drugs, conducting digital marketing campaigns, and running businesses as a local agency

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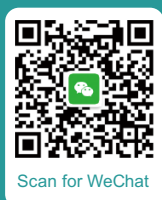
14th Floor, Building 3, Haichuang Technology Center,  
1288 West Wen Yi Road, Hangzhou, China

+86 (0)571 8710 3829

[info@baipharm.com](mailto:info@baipharm.com)

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14th Floor, Building 3, Haichuang Technology Center,

1288 West Wen Yi Road, Hangzhou, China

+86 (0)571 8710 3822

[contact@chemlinked.com](mailto:contact@chemlinked.com)