

第一批罕见病目录

序号	中文名称	英文名称
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 Receptoropathy (Type B insulin resistance)
12	β -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	Cardiac 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	General Myasthenic 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, 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懈症	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	Lysine Urinary 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	Multi-Focal Motor Neurothy
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	NAGS 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	Phenylketouria
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 Dystrophies
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	视网膜色素变性症	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 Adrenoleuko Dystrophy
121	X-连锁淋巴增生症	X-linked Lymphoproliferative Disease