Pacific Bridge Medical's Report On Orphan Drugs And Rare Diseases In China

On May 22, 2018, China's government issued a List of Rare Diseases. This List was jointly issued by the – 1. National Health Commission, 2. the Ministry of Science and Technology, 3. the Ministry of Industry and Information Technology, 4. the China Food and Drug Administration, and 5. the State Administration of Traditional Chinese Medicine. It is a landmark government document. This rare disease list will be taken as a reference and basis for the prevention, screening, diagnosis and rehabilitation of rare diseases, as well as the development of relevant R&D, insurance, medical assistance policies, etc.

Responsibility

The *Expert Committee for the Diagnosis, Treatment and Protection of Rare Diseases* under the National Health Commission (namely the MOH) is responsible for providing technical support and policy suggestions. The Office of the *Expert Committee* is responsible for receiving, summarizing and sorting out the new rare disease applications.

Next update of rare disease list

The list of rare diseases will be updated. However, in principle, it will be at least 2 years from May 22, 2018 until the next update.

Inclusion Criteria

The diseases to be included in the Rare Disease List shall meet all of the conditions below:

1) There is international or domestic evidence indicating a low incidence or prevalence;
2) The disease will cause great harm to patients and their families;
3) There is a definite method of diagnosis;
4) There is a financially affordable treatment or intervention means, or if there is no effective treatment or intervention means, it shall be included in the national special scientific research.

Who is eligible to make the application

Relevant state departments, provincial health administrative departments, national industry institutes/associations, and relevant NGOs registered with the Ministry of Civil Affairs may apply for the inclusion of a rare disease into the List and submit the materials to the Office of Expert Committee. Also, please see the "Other" section on the bottom of page 3.

Application Materials for the application

The application materials should include the following:

1) Basic conditions of the disease. It includes the name of the disease (in English and Chinese), incidence or prevalence, the sex, age of onset, diagnostic criteria, treatment (including drugs), prognosis (including survival), etc.
2) What is the annual cost of treatment in China; and is there insurance.
3) Management of diagnosis, treatment, medical insurance? Outline the social assistance (or insurance) for the disease in other major countries or regions;
4) Data sources and references for the above materials;
5) Applicant information, contact person and contact information, such as mailing address, telephone number and email address.
6) Other supporting information, clinical data, and documents.
7) Please keep in mind this is a very detailed application with detailed data and support from clinical papers.

Procedure to add a new rare disease

1) Before officially applying to add a rare disease, the applicant should identify the appropriate Chinese KOLs and key Chinese associations and lobby them.
2) Submit application
3) After receiving the application materials, the Office of the *Expert Committee* shall conduct a preliminary review. If the materials do not meet their requirements, the applicant will be required to submit additional supplements and documents.

   FYI, the contact information for the Office of the Experts Committee is:

   Office of the Experts Committee

   Address: No.1 Shuaifuyuan, Dongcheng District, Beijing 100005

   Tel: 86-10— 69154259

   Email: RD@NHFPC.GOV.CN

4) The Office of the Expert Committee will review and summarize the application materials and send a report to the National Health Committee and propose to hold a symposium in due course.

5) The National Health Committee will organize a larger meeting with all members of the Expert Committee according to the recommendations and application status from the Office of the Expert Committee and invite other experts in relevant fields to join in the discussion.

6) The Expert committees and other experts in the related fields shall, in accordance with the Inclusion Criteria, form a List (Exposure Draft) and submit the draft List to the National Health Commission with the signatures of the experts.

7) The National Health Committee will then seek comments from the relevant state departments, provincial health administrative departments, national industry institutes/associations, and relevant NGOs registered with the Ministry of Civil Affairs. Based on this feedback, the NHC will decide whether or not to organize another meeting. If no other meetings are planned, the rare disease will be accepted, and then be released to the public on the NHC website. In some cases, the application may be rejected.

8) In general, we suggest you proceed ASAP so that there is sufficient time for pre-meetings, etc.

The list of rare diseases

No.	Chinese Name	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	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	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、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 Subcortic al 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-Homoci trullinuria 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	Lymphangioleiomyomatosis (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-Albrigh 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 Academia
72	线粒体脑肌病	Mitochodrial 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)	n
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)	i
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	e
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	