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
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).

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

<table>
<thead>
<tr>
<th>No.</th>
<th>Diseases</th>
<th>No.</th>
<th>Diseases</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>21-Hydroxylase Deficiency</td>
<td>60</td>
<td>Langerhans Cell Histiocytosis</td>
</tr>
<tr>
<td>2</td>
<td>Albinism</td>
<td>61</td>
<td>Larson Syndrome</td>
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<tr>
<td>3</td>
<td>Alport Syndrome</td>
<td>62</td>
<td>Leber Hereditary Optic Neuropathy</td>
</tr>
<tr>
<td>4</td>
<td>Amyotrophic Lateral Sclerosis</td>
<td>63</td>
<td>Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency</td>
</tr>
<tr>
<td>5</td>
<td>Angelman Syndrome</td>
<td>64</td>
<td>Lymphangioleiomyomatosis (LAM)</td>
</tr>
<tr>
<td>6</td>
<td>Arginase Deficiency</td>
<td>65</td>
<td>Lysine Urinary Protein Intolerance</td>
</tr>
<tr>
<td>7</td>
<td>Asphyxiating Thoracic Dystrophy (Jeune Syndrome)</td>
<td>66</td>
<td>Lysosomal Lipase Deficiency</td>
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<tr>
<td>8</td>
<td>Atypical Hemolytic Uremic Syndrome</td>
<td>67</td>
<td>Maple Syrup Urine Disease</td>
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<tr>
<td>9</td>
<td>Autoimmune Encephalitis</td>
<td>68</td>
<td>Marfan Syndrome</td>
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<tr>
<td>10</td>
<td>Autoimmune Hypophysitis</td>
<td>69</td>
<td>McCune-Albright Syndrome</td>
</tr>
<tr>
<td>11</td>
<td>Autoimmune Insulin Receptopathy (Type B insulin resistance)</td>
<td>70</td>
<td>Medium Chain Acyl-CoA Dehydrogenase Deficiency</td>
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<tr>
<td>12</td>
<td>Beta-ketothiolase Deficiency</td>
<td>71</td>
<td>Methylmalonic Acidemia</td>
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<tr>
<td>13</td>
<td>Biotinidase Deficiency</td>
<td>72</td>
<td>Mitochondrial Encephalopathy</td>
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<tr>
<td>14</td>
<td>Cardiac Channelopathies</td>
<td>73</td>
<td>Mucopolysaccharidosis</td>
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<tr>
<td>15</td>
<td>Carnitine Deficiency</td>
<td>74</td>
<td>Multi-Focal Motor Neuropathy</td>
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<tr>
<td>16</td>
<td>Chorion-Marcie-Tooth Disease</td>
<td>75</td>
<td>Multiple Acyl-CoA Dehydrogenase Deficiency</td>
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<td>17</td>
<td>Citrullinemia</td>
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<td>Multiple Sclerosis</td>
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<td>Congenital Adrenal Hypoplasia</td>
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<td>Multiple System Atrophy</td>
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<td>Congenital Hyperinsulinemic Hypoglycaemia</td>
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<td>Myotonic Dystrophy</td>
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<td>NAGS Deficiency</td>
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<td>Neonatal Diabetes Mellitus</td>
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<td>Congenital Myotonia Syndrome (Non-Dystrophic Myotonia, NDM)</td>
<td>81</td>
<td>Neuromyelitis Optica</td>
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<td>Congenital Scoliosis</td>
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<td>Niemann-Pick Disease</td>
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<td>Ornithine Transcarbamylase Deficiency</td>
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<td>27</td>
<td>Fabry Disease</td>
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<td>Osteogenesis Imperfecta (Bristle Bone Disease)</td>
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<td>28</td>
<td>Familial Mediterranean Fever</td>
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<td>Parkinson Disease (Young-onset, Early-onset)</td>
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<td>Fanconi Anemia</td>
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<td>Paroxysmal Nocturnal Hemoglobinuria</td>
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<td>Galactosemia</td>
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<td>Peutz-Jeghers Syndrome</td>
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<td>Gaucher's Disease</td>
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<td>Phenylketonuria</td>
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<td>General Myathetic Gravis</td>
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<td>Gittelman Syndrome</td>
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<td>Porphyria</td>
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<td>Glutaric Acidemia Type I</td>
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<td>Prader-Willi Syndrome</td>
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<td>35</td>
<td>Glycogen Storage Disease (Type I, II)</td>
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<td>Primary Combined Immune Deficiency</td>
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<td>Hemospheria</td>
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<td>Primary Hereditary Dystonia</td>
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<td>Hepatocellular Degeneration (Wilson Disease)</td>
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<td>Primary Light Chain Amyloidosis</td>
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<td>38</td>
<td>Hereditary Angiodema (HAE)</td>
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<td>Progressive Familial Intrahepatic Cholestasis</td>
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<td>Hereditary Epidermolytic Bullosa</td>
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<td>Progressive Muscular Dystrophies</td>
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<td>Hereditary Fructose Intolerance</td>
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<td>Propionic Acidemia</td>
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<td>41</td>
<td>Hereditary Hypomagnesemia</td>
<td>100</td>
<td>Pulmonary Alveolar Proteinosis</td>
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<td>42</td>
<td>Hereditary Multi-infarct Dementia (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, CADASIL)</td>
<td>101</td>
<td>Pulmonary Cystic Fibrosis</td>
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<td>Hereditary Spastic Paraplegia</td>
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<td>Severe Myoclonic Epilepsy In Infancy (Dravet Syndrome)</td>
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<td>Sistosterolemia</td>
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<td>Hyperphenylalaninemia</td>
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<td>Spinal and Bulbar Muscular Atrophy (Kennedy Disease)</td>
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<td>Spinal Muscular Atrophy</td>
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<td>Leber Hereditary Optic Neuropathy</td>
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<td>X-linked Lymphoproliferative Disease</td>
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</tbody>
</table>

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

References


(Received May 27, 2018; Accepted May 29, 2018)