China takes a major step toward policies to support patient access to treatments for rare diseases

Policy update

On May 22, 2018 the National Health and Sanitation Commission, the Ministry of Science and Technology, the Ministry of Industry and Information Technology, the State Drug Administration, and the State Administration of Traditional Chinese Medicine jointly issued the "First Rare Diseases" list, involving a total of 121 diseases.

With an estimated 16 million people in China having been diagnosed with a rare disease and a longstanding absence of a national agenda and policies to support innovation in the industry, government action is welcomed and signals the beginning of a series of new regulations and policies that are expected to significantly improve patients' access to care.

The announcement is generating significant interest amongst international drug manufacturers that have an orphan drug portfolio.

Rare Diseases on the Chinese National Registry:

1. 21-Hydroxyulase Deficiency
2. Albinism
3. Alport Syndrome
4. Amyotrophic Lateral Sclerosis
5. Angelman Syndrome
6. Arginase Deficiency
7. Asphyxiating Thoracic Dystrophy (Jeune Syndrome)
8. Atypical Hemolytic Uremic Syndrome
9. Autoimmune Encephalitis
10. Autoimmune Hypophysitis
11. Autoimmune Insulin Receptopathy (Type B insulin resistance
12. Beta-Ketothiolase Deficiency
13. Biotinidase Deficiency
14. Cardiac Ion Channelopathies
15. Carnitine Deficiency
16. Castleman Disease
17. Charcot-Marie-Tooth Disease
18. Citrullinemia
19. Congenital Adrenal Hypoplasia
20. Congenital Hyperinsulinemic Hypoglycemia
21. Congenital Myasthenic Syndrome
22. Congenital Myotonia Syndrome (Non-Dystrophic Myotonia, NDM)
23. Congenital Scoliosis
24. Coronary Artery Ectasia
25. Diamond-Blackfan Anemia
26. Erdheim-Chester Disease
27. Fabry Disease
28. Familial Mediterranean Fever
29. Fanconi Anemia
30. Galactosemia
31. Gaucher's Disease
32. General Myasthenia Gravis
33. Gitelman Syndrome
34. Glutaric Acidemia type I
35. Glycogen Storage Disease (Type I、II)
36. Hemophilia
37. Hepatolenticular Degeneration(Wilson Disease)
38. Hereditary Angioedema(HAE)
39. Hereditary Epidermolysis Bullosa
40. Hereditary Fructose Intolerance
41. Hereditary Hypomagnesemia
42. Hereditary Multi-infarct Dementia (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, CADASIL)
43. Hereditary Spastic Paraplegia
44. Holocarboxylase Synthetase Deficiency
45. Homocysteinemia
46. Homozygous Hypercholesterolemia
47. Huntington Disease
48. Hyperornithinaemia-Hyperammonaemia-Hhomocitrullinuria Syndrome
49. Hyperphenylalaninemia
50. Hypophosphatasia
51. Hypophosphatemic Rickets
52. Idiopathic Cardiomyopathy
53. Idiopathic Hypogonadotropic Hypogonadism
54. Idiopathic Pulmonary Arterial Hypertension
55. Idiopathic Pulmonary Fibrosis
56. IgG4 related Disease
57. Inborn Errors of Bile Acid Synthesis
58. Isovaleric Acidemia
59. Kallmann Syndrome
60. Langerhans Cell Histiocytosis
61. Laron Syndrome
62. Leber Hereditary Optic Neuropathy
63. Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency
64. Lymphangioleiomyomatosis (LAM)
65. Lysine Urinary Protein Intolerance
66. Lysosomal Acid Lipase Deficiency
67. Maple Syrup Urine Disease
68. Marfan Syndrome
69. McCune-Albright Syndrome
70. Medium Chain Acyl-CoA Dehydrogenase Deficiency
71. Methylmalonic Academia
72. Mitochondrial Encephalomyopathy
73. Mucopolysaccharidosis
74. Multi-Focal Motor Neurothy
75. Multiple Acyl-CoA Dehydrogenase Deficiency
76. Multiple Sclerosis
77. Multiple System Atrophy
78. Myotonic Dystrophy
79. NAGS Deficiency
80. Neonatal Diabetes Mellitus
81. Neuromyelitis Optica
82. Niemann-Pick Disease
83. Non-Syndromic Deafness
84. Noonan Syndrome
85. Ornithine Transcarbamylase Deficiency
86. Osteogenesis Imperfecta (Brittle Bone Disease)
87. Parkinson Disease (Young-onset, Early-onset)
88. Paroxysmal Nocturnal Hemoglobinuia
89. Peutz-Jeghers Syndrome
90. Phenylketonuria
91. POEMS Syndrome
92. Porphyria
93. Prader-Willi Syndrome
94. Primary Combined Immune Deficiency
95. Primary Hereditary Dystonia
96. Primary Light Chain Amyloidosis
97. Progressive Familial Intrahepatic Cholestasis
98. Progressive Muscular Dystrophies
99. Propionic Acidemia
100. Pulmonary Alveolar Proteinosis
101. Pulmonary Cystic Fibrosis
102. Retinitis Pigmentosa
103. Retinoblastoma
104. Severe Congenital Neutropenia
105. Severe Myoclonic Epilepsy In Infancy (Dravet Syndrome)
106. Sickle Cell Disease
107. Silver-Russell Syndrome
108. Sitosterolemia
109. Spinal and Bulbar Muscular Atrophy (Kennedy Disease)
110. Spinal Muscular Atrophy
111. Spinocerebellar Ataxia
112. Systemic Sclerosis
113. Tetrahydrobiopterin Deficiency
114. Tuberous Sclerosis Complex
115. Tyrosinemia
116. Very Long Chain Acyl-CoA Dehydrogenase Deficiency
117. Williams Syndrome
118. Wiskott-Aldrich Syndrome
119. X-linked Agammaglobulinemia
120. X-linked adrenoleukodystrophy
121. X-linked Lymphoproliferative Disease

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