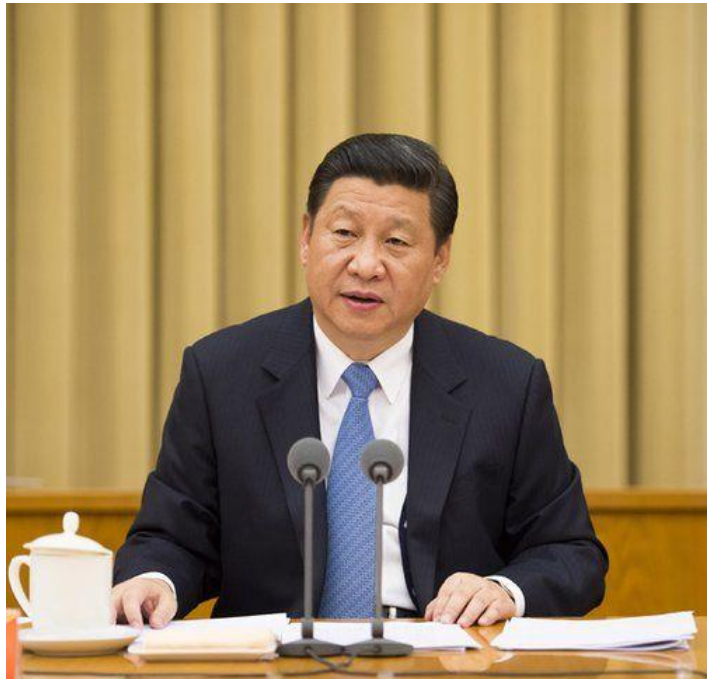
HEALTH FOR ALL

China's Initiative to Combat the Challenges of Rare Diseases

SHUYANG ZHANG MD

Peking Union Medical College Hospital

Healthy China 2030 Program
— Health for All



‘Health is a prerequisite for people's all-round development and a precondition for economic and social development.’

— President Xi Jinping, National Health Conference, 2016

Policies at National Level for Rare Diseases Care in China

1. Policy Making and the Consulting Organization
2. Clinical Care System governed by China NHC
3. National Rare Diseases Registry System supported by PUMCH
4. Specialty Communities, Workforce Training and Medical Education
5. Prioritized Approval Process by China FDA

Policies at National Level for Rare Diseases Care in China

6. Tax reduction for Orphan Drugs by Central Government
7. Research Funding in Rare Diseases
8. Social Care and Charities
9. Broad Involvement of Multiple Stakeholders
10. Collaboration with International Communities

Policy Making and the Consulting Organization

2016.1 Establishment of rare diseases experts committee

(Diagnosis/Treatment/Medical security) (Member of National Health committee)

Primary task:

Catalogue of Rare Diseases in China

医政医管局

主站首页 | 首页 | 最新信息 | 政策文件 | 工作动态 | 关于我们 | 图片集锦 | 专题专栏

通告公告

关于公布第一批罕见病目录的通知

发布时间：2018-06-08

国卫医发〔2018〕10号

各省、自治区、直辖市及新疆生产建设兵团卫生计生委、科技厅（委、局）、工业和信息化主管部门、食品药品监督管理局、中医药管理局：

为贯彻落实中共中央办公厅、国务院办公厅《关于深化审评审批制度改革鼓励药品医疗器械创新的意见》，加强我国罕见病管理，提高罕见病诊疗水平，维护罕见病患者健康权益，国家卫生健康委员会等5部门联合制定了《第一批罕见病目录》。现印发你们，供各部门在工作中参考使用。

国家卫生健康委员会　　科学技术部
工业和信息化部　国家药品监督管理局
国家中医药管理局
2018年5月11日

China's First List of Rare Diseases (2018.5)

- National Health Committee
- Ministry of Science and Technology
- Ministry of Industry and Information Technology
- State Food and Drug Administration
- State Administration of Traditional Chinese Medicine

Five ministries jointly issued

the first version of RD catalogue

121 diseases included

The second edition will be updated soon

国家卫生健康委员会
科 学 技 术 部
工业和信息化部文件
国家药品监督管理局
国家中医药管理局

国卫医发〔2018〕10号

关于公布第一批罕见病目录的通知

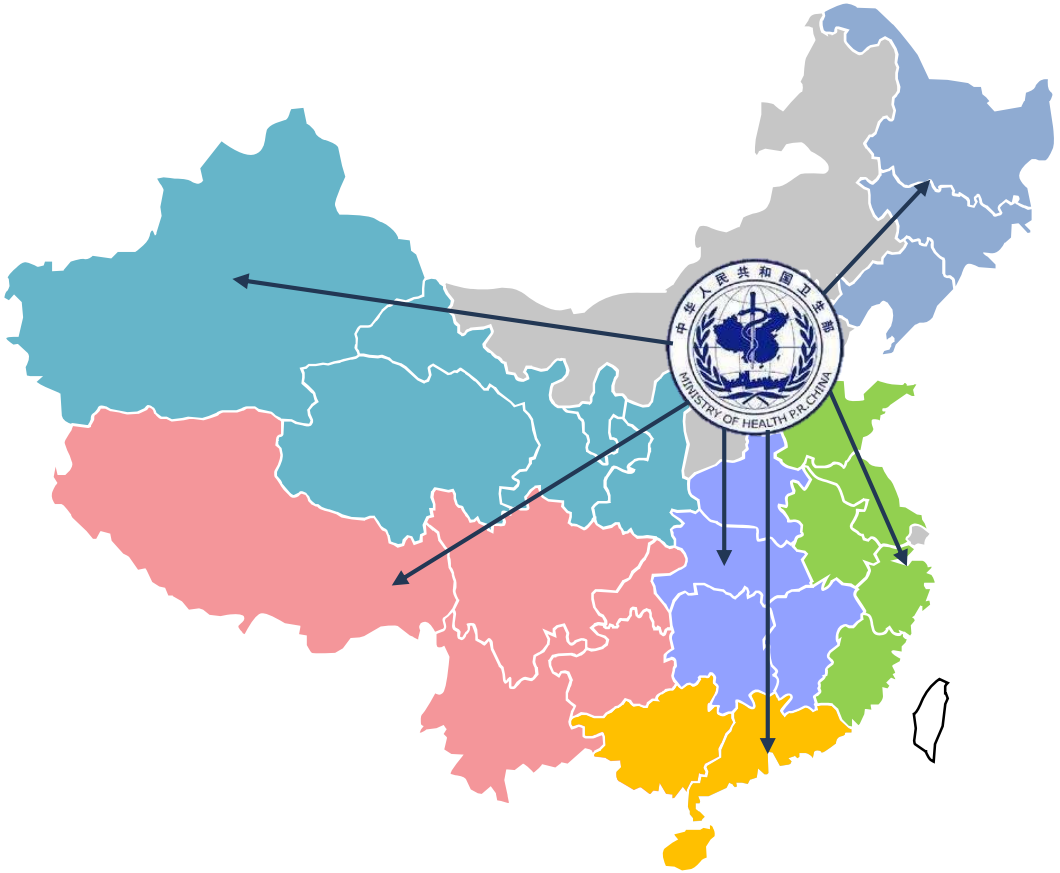
各省、自治区、直辖市及新疆生产建设兵团卫生计生委、科技厅（委、局）、工业和信息化主管部门、食品药品监督管理局、中医药管理局：

为贯彻落实中共中央办公厅、国务院办公厅《关于深化审评审批制度改革鼓励药品医疗器械创新的意见》，加强我国罕见病管理，提高罕见病诊疗水平，维护罕见病患者健康权益，国家卫生健康委员会等5部门联合制定了《第一批罕见病目录》。现印发你

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 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 Hperinsulinemic 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-Dystro 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 Myathenic 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 Angiocdema (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 (Cere Autosomal Dominant Arteriopathy with Subcor 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-Hhom rullinuria 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 Dehydrogena Deficiency
64	淋巴管肌瘤病	Lymphangioleiomyomatosis (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	McCuno-Albrigh 综合征	McCuno-Albright Syndrome
70	中链酰基辅酶 A 脱氢酶缺乏症	Medium Chain Acyl-CoA Dehydrogena 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	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 Hemoglobinuia
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

A Nation-wide Medical Care System



324 hospitals designated as the Clinical Center of Rare Diseases

National (1) PUMCH
Provincial (32), Municipal (291)

Directed by the National Committee of Health

(equivalent to China Ministry of Health)

Duties of Medical Care System

1. Screening and Primary Diagnosis
2. Dual-Direction Referral of Undiagnosed Patients
3. Long-term Clinical Management
4. Consulting for Child-birth
5. Report and Registry
6. Priority for Orphan Drug Distribution
7. Clinical Trials of Orphan Drugs

National Rare Diseases Registry System

NRDRS National Rare Diseases Registry System of China

www.nrdrs.org.cn Initiated from Dec, 2016

Supported by

①National Health Committee, China

②Ministry of Science and Technology, China

③Peking Union Medical College Hospital

④Total budget: 40,000,000 RMB

National Rare Diseases Registry System

Statistics

1. Over 30,000 Cases (Clinical + Bio samples)
2. Over 180 Disease Entities
3. > 150 Experts
4. > 50 Research Institutes

Long-term Goal

1. Phenotypic-Genomic Integrated Data Platform
2. Research Infrastructure for China and the World
3. Accurate Statistics of RD in China

Associations and Workforce Training

The Rare Diseases Research Center of Chinese Academy of Medical Sciences (CAMS)

The Rare Diseases Society of the Chinese Association of Research Hospitals

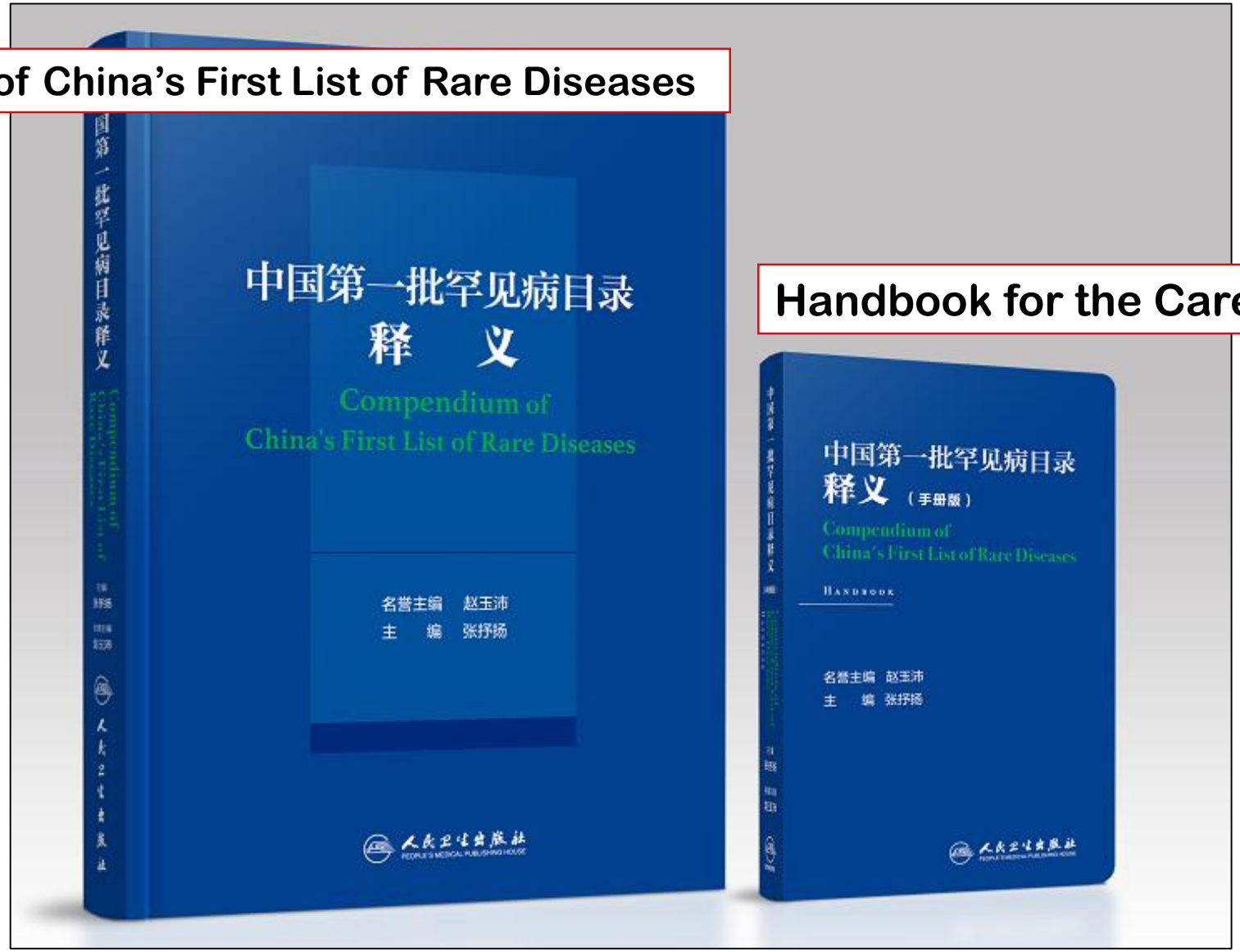
Provincial Rare Disease association

- ① Beijing
- ② Shanghai
- ③ Shandong
- ④ others

Nation-wide Training of the Clinical Care and Research of Rare Diseases

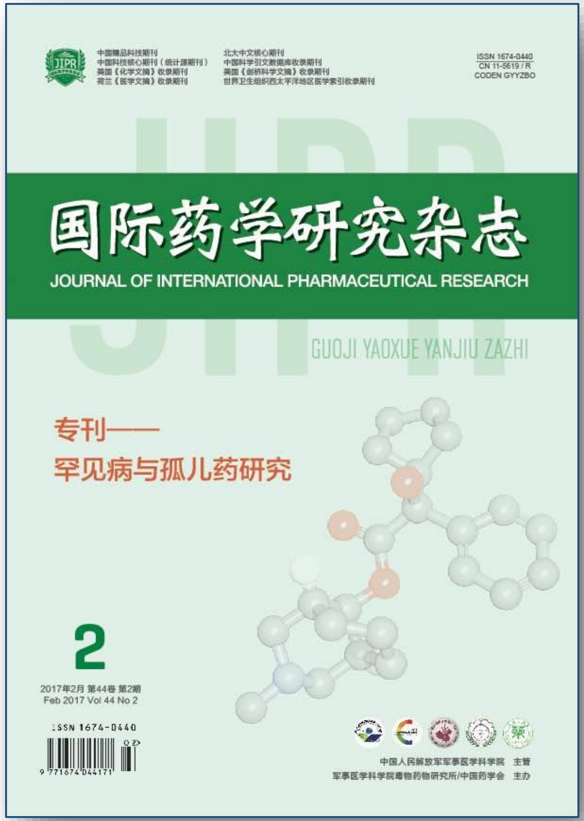
Medical Education — Publication

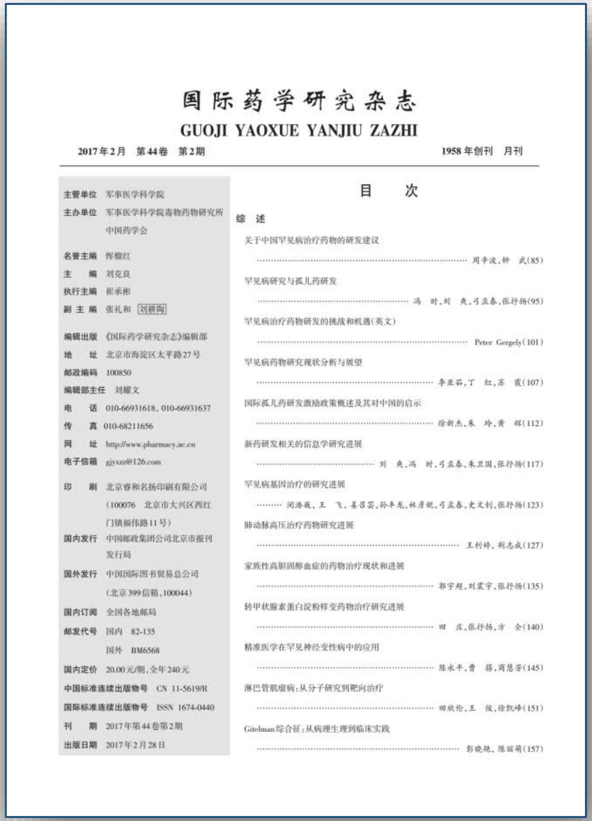
Compendium of China's First List of Rare Diseases



Handbook for the Care of Rare Diseases

Publishing Special Issues on Rare Diseases and Orphan Drugs





国际药学研究杂志

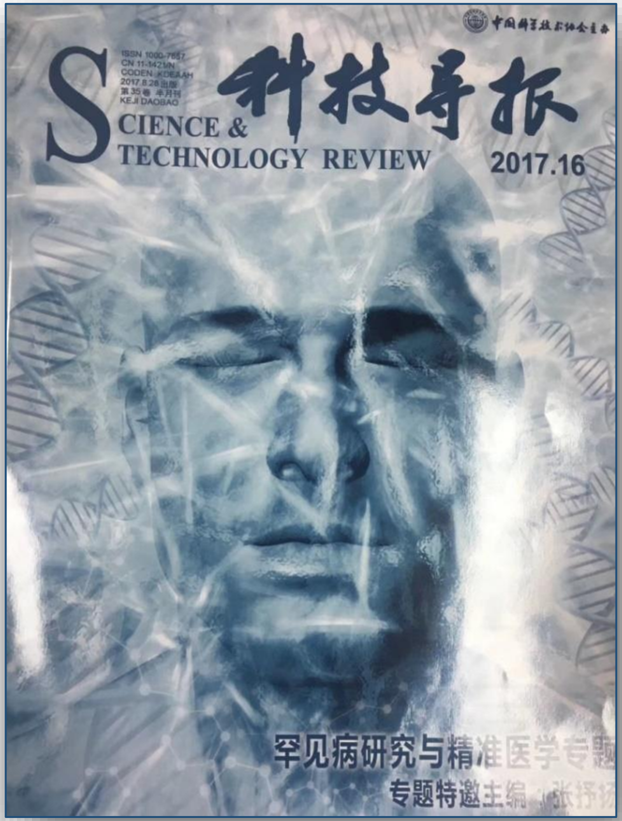
GUOJI YAOXUE YANJIU ZAZHI

2017年2月 第44卷 第2期 1958年创刊 月刊

主管单位 军事医学科学院
主办单位 军事医学科学院毒物药物研究所 中国药学会
名誉主编 恽榴红
主 编 刘克良
执行主编 崔承彬
副 主 编 张礼和 刘耕陶
编辑出版 《国际药学研究杂志》编辑部
地 址 北京市海淀区太平路27号
邮政编码 100850
编辑部主任 刘耀文
电 话 010-66931618、010-66931637
传 真 010-68211656
网 址 http://www.pharmacy.ac.cn
电子信箱 gjyxzz@126.com
印 刷 北京睿和名扬印刷有限公司（100076 北京市大兴区西红门镇福伟路11号）
国内发行 中国邮政集团公司北京市报刊发行局
国外发行 中国国际图书贸易总公司（北京399信箱，100044）
国内订阅 全国各地邮局
邮发代号 国内 82-135 国外 BM6568
国内定价 20.00元/期，全年240元
中国标准连续出版物号 CN 11-5619/R
国际标准连续出版物号 ISSN 1674-0440
刊 期 2017年第44卷第2期
出版日期 2017年2月28日

目 次

综 述



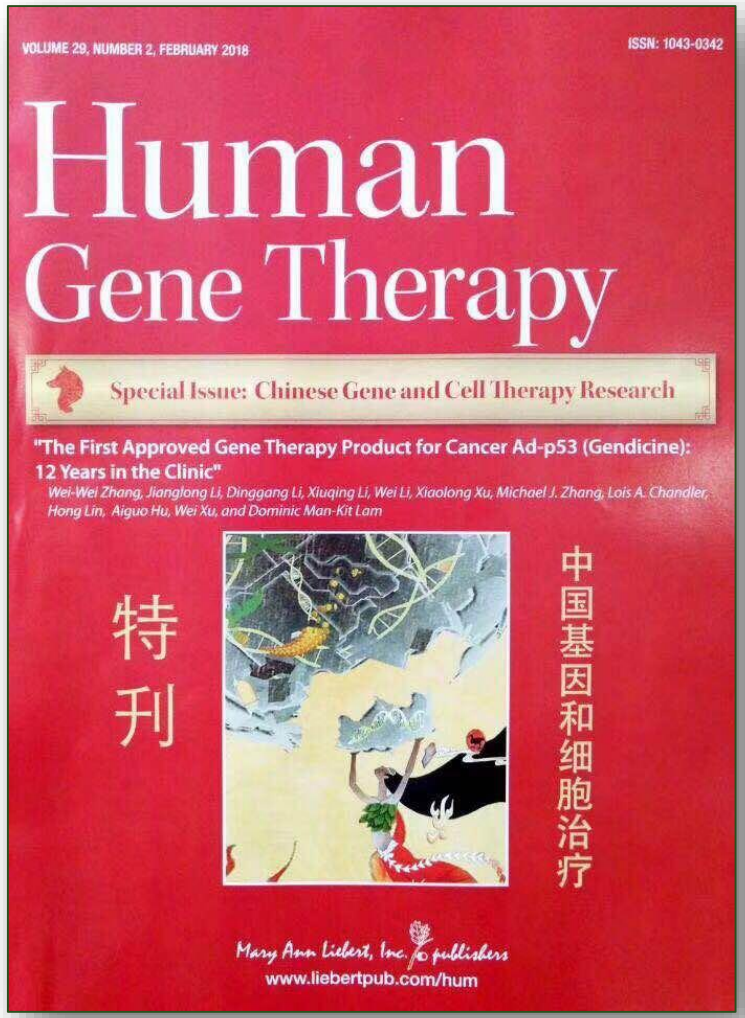
专刊寄语

罕见病患病率低，但病种繁多。据估计全球有超过3亿罕见病患者，其中在中国估计超过3000万。目前95%的罕见病没有治疗药物，因此罕见病实际给社会带来了重大疾病负担。推进罕见病研究和孤儿药研发，将增进罕见病人群健康福祉，对于经济、社会、伦理均具有重大意义，是健康中国建设必不可少的一环。

张抒扬
北京协和医院副院长
罕见病队列研究首席专家

罕见病药物研发已经成为当今生物医药产业极为关注的领域。据估计，目前全球孤儿药市场增速是常见疾病药物的2倍。据 Evaluate Pharma 分析，到2020年，全球孤儿药市场价值可达1760亿美元，接近处方药销售总额的19%。欧美等发达国家均将罕见病从国家战略层面部署，以抢占生物医药科技制高点。2014年，美国食品药品监督管理局(FDA)批准了19个罕见病治疗新药，占当年批准新药总数的38%，罕见病药物的市场展现巨大商业价值。而长期以来我国罕见病研究科研投入缺乏，罕见病药物严重依赖进口。随着近些年国家和社会对于罕见病群体的关注日益增加，预计未来孤儿药研发也将成为我国生物医药产业发展中的亮点。

Publishing Special Issues on Rare Diseases and Orphan Drugs



National Rare Diseases Registry System of China and Related Cohort Studies: Vision and Roadmap

Shi Feng,[1,†] Shuang Liu,[1,†] Chong Zhu,[2,3] Mengchun Gong,[2,4] Yicheng Zhu,[2,5] and Shuyang Zhang[2,5]

[1]Peking Union Medical College, Beijing, China; [2]National Rare Diseases Registry System of China, Beijing, China; [3]Digital China Health Technologies Co., Ltd., Beijing, China; [4]Rare Diseases Research Center, Chinese Academy of Medical Sciences, Beijing, China; [5]Peking Union Medical College Hospital, Beijing, China.
[†]These authors contributed equally to this work.

Rare diseases are major challenges in healthcare and medical research and are the basis of national development strategies in many countries. However, inadequate definition of rare diseases and lags in orphan drug development in China hinder rare disease research. In response, the first National Rare Diseases Registry System of China (NRDRS) was established, and various cohort studies have been launched since 2016. More than 20 top academic institutions in China are currently participating in this joint effort to carry out nationwide registration of rare diseases. The primary objectives are to establish standardization for the registration platform, build biobanks of genomic data, and create partnerships for data sharing and research collaboration. Innovative informatics technologies have been implemented to develop the NRDRS, including employment of ontological and knowledge bases to render standardization and support standard of care. Development of informatics analysis tools will facilitate accurate and more efficient diagnoses for rare diseases. Long-term research collaboration is encouraged to create additional national rare disease networks for research translation and to benefit patients with rare diseases. The NRDRS of China and related cohort studies are anticipated to enlighten rare disease research significantly in China.

Keywords: rare diseases, patient registry, cohort study, medical informatics, genomics

INTRODUCTION

RARE DISEASES REPRESENT a large group of health-related entities without a universally recognized definition. The definition of rare diseases as a whole varies by country and according to specific socioeconomic circumstances. For example, the United States defines rare diseases as disorders affecting <200,000 individuals, while the European definition is diseases with a prevalence of <5/10,000 people.[1] Despite the low prevalence of a given rare disease, the overall population affected by rare diseases in general is strikingly large due to the great variety that exists. Thus, rare diseases substantially contribute to a high disease burden for patients, families, and communities around the world.[2–4] Considering the vast population of China, disease burden can add up to an astronomical figure. It is estimated that >10,000,000 patients are affected with chromosomal disease syndromes, while monogenic diseases affect >1,000,000 patients.[5] Despite the heavy disease burden, rare diseases receive insufficient attention, resulting in significant lags in many aspects related to their research, understanding, and treatment.[6,7] Hence, there is an urgent need for a national coordinated and collaborative rare disease research program.[6]

Advancements in genomic testing have helped to shed light on diagnosis and management of rare diseases. As the majority of rare diseases are genetic in origin,[8] molecular genetic diagnostic techniques can play a critical role in this area. In addition to traditional genetic testing methods, high-throughput next-generation sequencing technology is being widely adopted and functions as a major tool in rare disease diagnosis and research.[9] Transcriptomics and metabolomics data may be applied to address the diagnostic demands that are unmet by traditional enzymatic detection

*Correspondence: Dr. Shuyang Zhang, 1 Shuaifuyuan, Dongcheng, Beijing, 100730, China. E-mail: shuyangzhang103@163.com

DOI: 10.1089/hum.2017.215

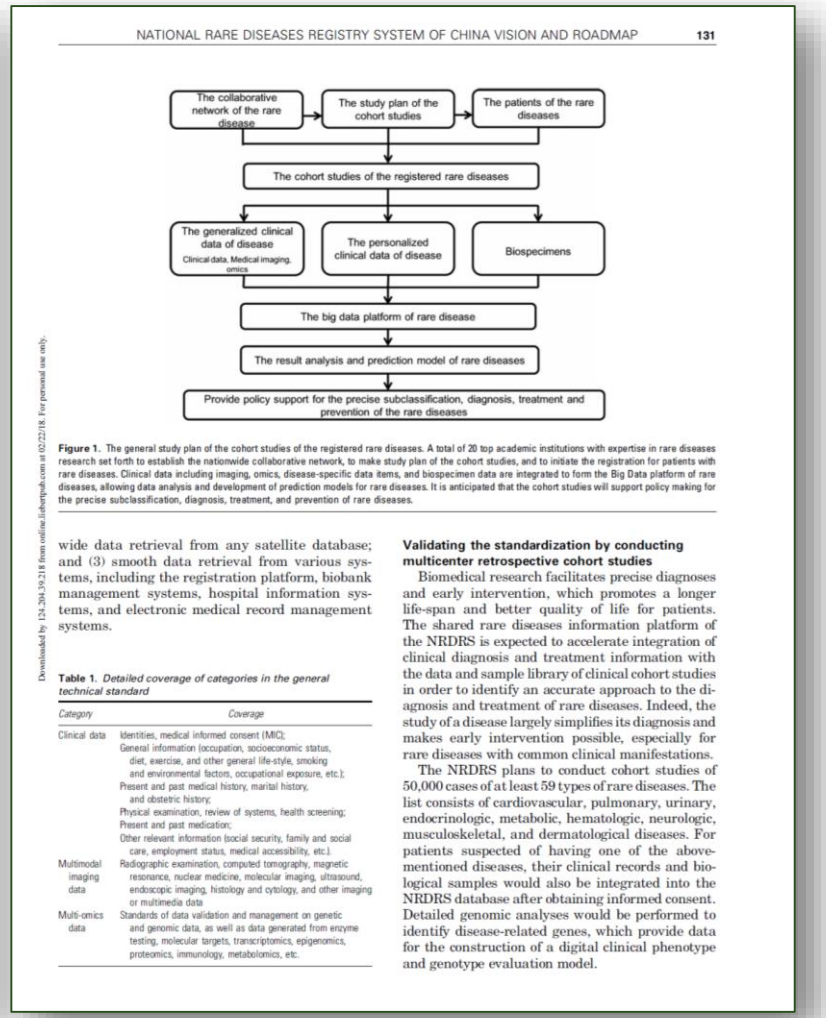


Figure 1. The general study plan of the cohort studies of the registered rare diseases. A total of 20 top academic institutions with expertise in rare diseases research set forth to establish the nationwide collaborative network, to make study plan of the cohort studies, and to initiate the registration for patients with rare diseases. Clinical data including imaging, omics, disease-specific data items, and biospecimen data are integrated to form the Big Data platform of rare diseases, allowing data analysis and development of prediction models for rare diseases. It is anticipated that the cohort studies will support policy making for the precise subclassification, diagnosis, treatment, and prevention of rare diseases.

wide data retrieval from any satellite database; and (3) smooth data retrieval from various systems, including the registration platform, biobank management systems, hospital information systems, and electronic medical record management systems.

Table 1. *Detailed coverage of categories in the general technical standard*

Category	Coverage
Clinical data	Identities, medical informed consent (MIC); General information (occupation, socioeconomic status, diet, exercise, and other general life-style, smoking and environmental factors, occupational exposure, etc.); Present and past medical history, marital history, and obstetric history; Physical examination, review of systems, health screening; Present and past medication; Other relevant information (social security, family and social care, employment status, medical accessibility, etc.).
Multimodal imaging data	Radiographic examination, computed tomography, magnetic resonance, nuclear medicine, molecular imaging, ultrasound, endoscopic imaging, histology and cytology, and other imaging or multimedia data
Multi-omics data	Standards of data validation and management on genetic and genomic data, as well as data generated from enzyme testing, molecular targets, transcriptomics, epigenomics, proteomics, immunology, metabolomics, etc.

Validating the standardization by conducting multicenter retrospective cohort studies

Biomedical research facilitates precise diagnoses and early intervention, which promotes a longer life-span and better quality of life for patients. The shared rare diseases information platform of the NRDRS is expected to accelerate integration of clinical diagnosis and treatment information with the data and sample library of clinical cohort studies in order to identify an accurate approach to the diagnosis and treatment of rare diseases. Indeed, the study of a disease largely simplifies its diagnosis and makes early intervention possible, especially for rare diseases with common clinical manifestations.

The NRDRS plans to conduct cohort studies of 50,000 cases of at least 59 types of rare diseases. The list consists of cardiovascular, pulmonary, urinary, endocrinologic, metabolic, hematologic, neurologic, musculoskeletal, and dermatological diseases. For patients suspected of having one of the above-mentioned diseases, their clinical records and biological samples would also be integrated into the NRDRS database after obtaining informed consent. Detailed genomic analyses would be performed to identify disease-related genes, which provide data for the construction of a digital clinical phenotype and genotype evaluation model.

Other Medical Education programs

- **Translation of Gene Reviews into Chinese by NRDRS**

  (7 million characters in total)

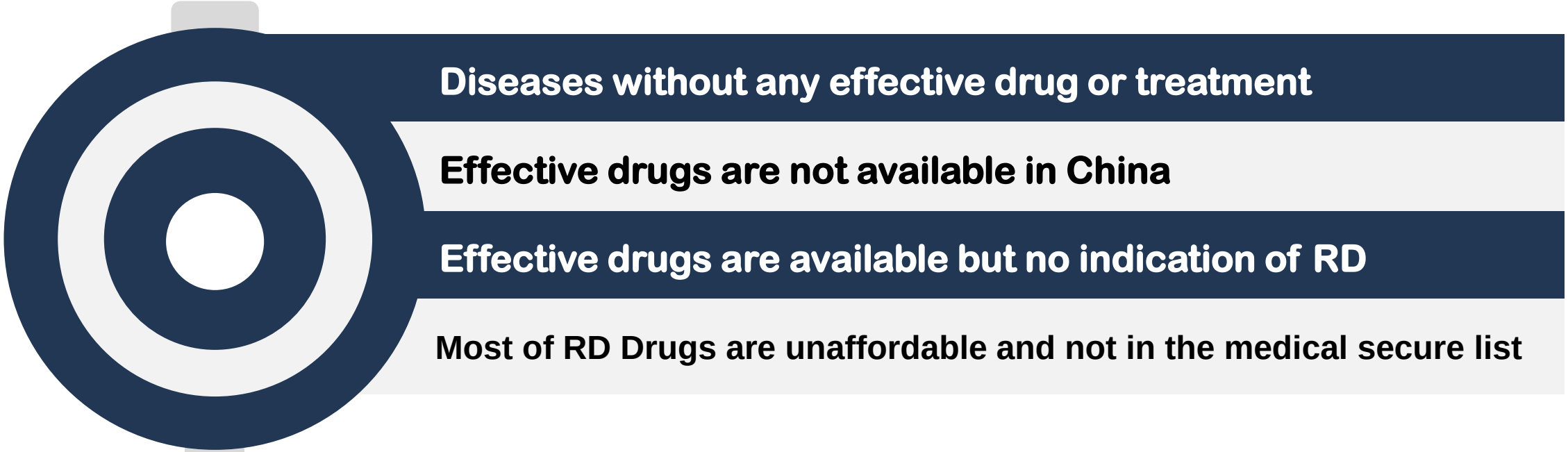
- **Initiation of the Textbook of Rare Diseases for graduate students**

- **Collaboration with different specialty societies for Continued Medical Education**

中华医学会 CHINESE MEDICAL ASSOCIATION 1915

- Cardiology
- Nephrology
- Neurology

Rare Disease Medication Dilemma



NEED TO IMPROVE:

- Orphan Drug Research and Development
- Approaches to Drug Import
- Clinical Trials
- Medical Insurance Policy

Prioritized Drug Approval Process

1. **Fast-track for drug approval for the orphan drugs approved in the US, EU and Japan**
2. **China has formed an approval process for orphan drugs in urgent need**
3. **Criteria**
   - ① With indication for the entities enlisted in the First List of Rare Diseases
   - ② Drugs for severe and life-threatening conditions that has no effective therapeutic methods
   - ③ Drugs for severe and life-threatening conditions and those with significantly increased clinical benefit.
4. **Approval Duration: 3-6 months**
5. **Post-marketing Research requested for efficacy and safety evaluation**
6. **The System of Health Technology Assessment (HTA/HEOR) for Rare Diseases and Orphan Drugs**

Tax Reduction for Orphan Drugs

1. Announced on Feb 11th by Prime Minister Li Keqiang

2. Value-added tax 16% → 3% (**80% Reduction**)

3. 21 Orphan Drugs

4. March 1st 2019

5. Perspectives

   ① Reduction of the financial burden of patients

   ② Incentive for orphan drug pharmaceutical companies

   ③ Promotion of the social awareness of rare diseases and orphan drugs

Research Funding

1. **National Key Science and Technology Research Program**
   - ① Rare Diseases Registry and Cohort Studies > 40m RMB
   - ② Diagnostics Technologies Innovation > 20m RMB
   - ③ Other related Diseases > 50m RMB
2. **National Natural Science Foundation, Special Fund for Rare Diseases**
3. **Provincial Level Research Funding for Rare Diseases**

   ①Beijing ②Shanghai ③Guangdong ④Zhejiang ⑤Shandong
4. **Novel Drug Discovery and Development Program**

Social Care and Charities

1. Regional Social Insurance Coverage
   ① Qingdao
   ② Shanghai
   ③ Beijing
   ④ others
2. Disability Care and Support
3. Rehabilitation and Enforcement of Employment
4. Prohibition and Prevention of Discrimination
5. Financial Support Program by Various Charities

Broad Involvement of Multiple Stakeholders

Broad Involvement of Multiple Stakeholders

2018.10
China Alliance For Rare Diseases

Academic organization
Research Institute

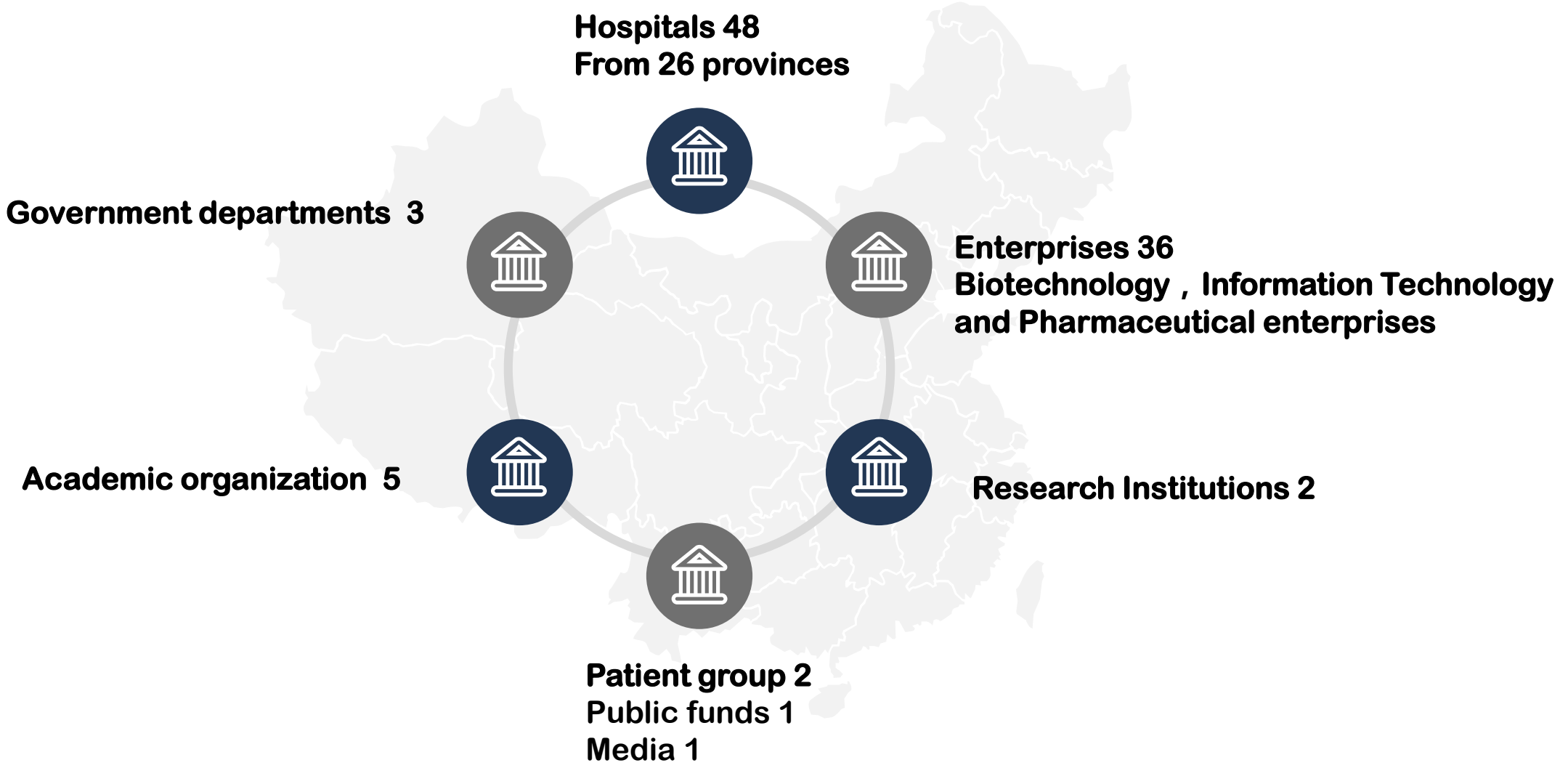
Pharmaceutical enterprises

Hospital

Patient group
Public funds
Media publicity

Government

Members of China Alliance For Rare Diseases



Duties of China Alliance For Rare Diseases

Research on policy

Academic communication

Shared data platform

Standards for medical care

Rapid Product translation

Professional training

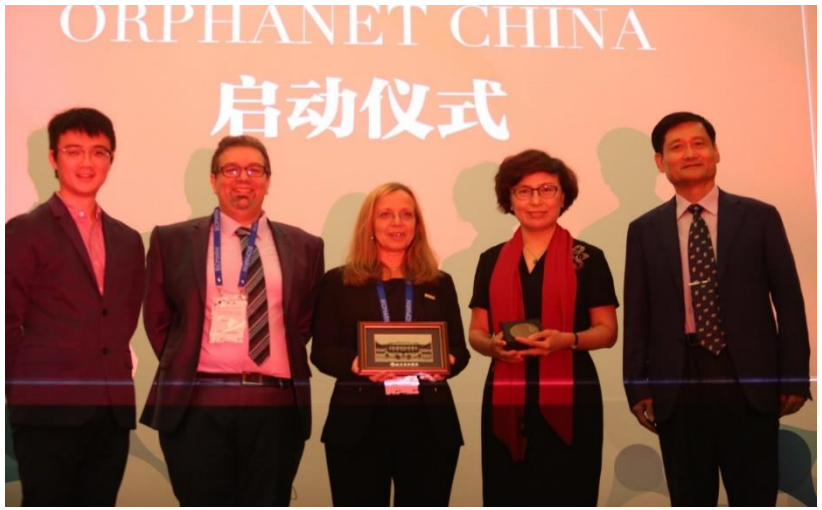
International cooperation

Social public service

Innovation of orphan drugs

Collaboration with International Communities

- ICORD
- Orphanet
- IRDiRC
- SNOMED International
- AMIA
- Broad Institute
- Yale University
- Weitzmann Institute of Science



ICORD

International Conference on Rare Diseases & Orphan Drugs

ICORD 2017 in China

12TH ANNUAL ICORD MEETING, 7-10 SEPTEMBER 2017, BEIJING, CHINA

Welcome to NCCORD

JUNE 2019，in Beijing

National Conference of China on Rare Diseases and Orphan Drugs

PEKING UNION MEDICAL COLLEGE HOSPITAL 1921

NRDRS 中国国家罕见病注册系统 NATIONAL RARE DISEASES REGISTRY SYSTEM OF CHINA

China Alliance for Rare Diseases 2018

RARE DISEASES IN CHINA:

TOWARDS A BETTER AND FAIRER FUTURE

Shuyang PUMCH

Shuyang Zhang, MD

- Professor, Chief Physician of Cardiovascular Medicine, Peking Union Medical College Hospital
- Vice President, Peking Union Medical College Hospital
- Vice-President, Peking Union Medical College & Chinese Academy of Medical Sciences
- Director, Rare Diseases Branch of Chinese Research Hospitals Association
- Director, the Office of the Expert Committee on Diagnosis, Treatment and Medical Security of Rare Diseases of National Health Committee
- Project Leader, The national rare disease clinical cohort
- Project Leader, National Rare Diseases Registry System of China
- shuyangzhang103@163.com