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8 – 12 JULY 2023 | SYDNEY, AUSTRALIA

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Construction of national multicentre rare diseases registration system

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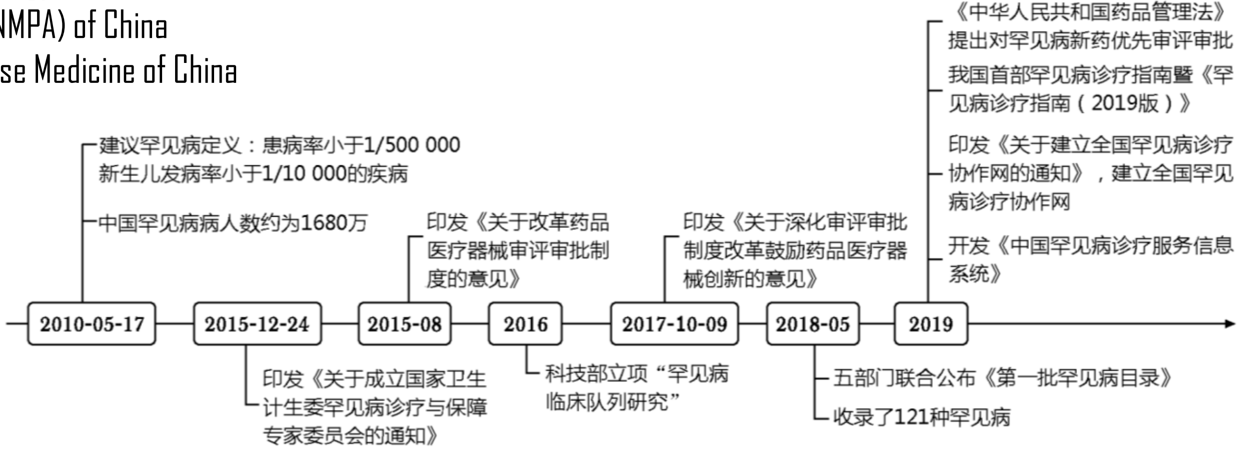
AIDH IMIA

Milestone event of rare disease development in China

- From 2010 to 2023
- National Health Commission of China
- State Council of China
- Ministry of Science and Technology of China
- Ministry of Industry and Information Technology of China
- National Medical Products Administration (NMPA) of China
- National Administration of Traditional Chinese Medicine of China
- 7 policy documents



2010-05-17
- 建议罕见病定义：患病率小于1/500 000新生儿发病率小于1/10 000的疾病
- 中国罕见病病人数约为1680万

2015-12-24
- 印发《关于成立国家卫生计生委罕见病诊疗与保障专家委员会的通知》

2015-08
- 印发《关于改革药品医疗器械审评审批制度的意见》

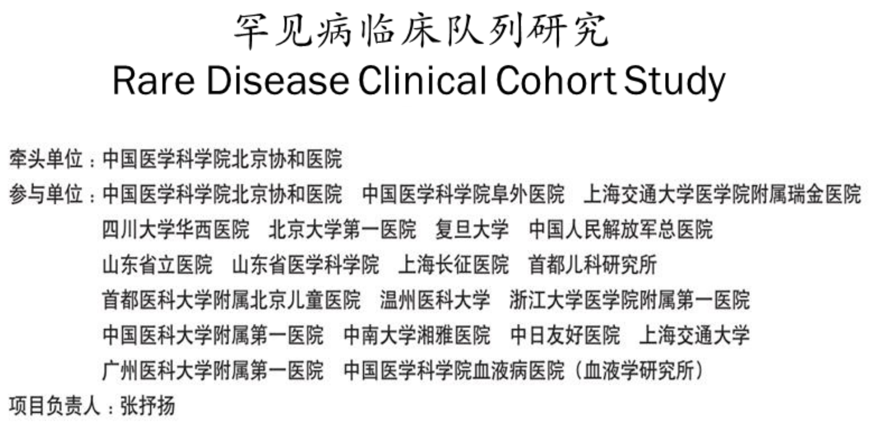
2016
- 科技部立项“罕见病临床队列研究”

2017-10-09
- 印发《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》

2018-05
- 五部门联合公布《第一批罕见病目录》
- 收录了121种罕见病

2019
- 《中华人民共和国药品管理法》提出对罕见病新药优先审评审批
- 我国首部罕见病诊疗指南暨《罕见病诊疗指南（2019版）》
- 印发《关于建立全国罕见病诊疗协作网的通知》，建立全国罕见病诊疗协作网
- 开发《中国罕见病诊疗服务信息系统》

Rare Disease Clinical Cohort Study



罕见病临床队列研究
Rare Disease Clinical Cohort Study

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项目负责人：张抒扬

"13th Five-Year Plan"
National Key Research and Development Program
'Key Special Project for Precision Medicine Research'

- In **2016**, China initiated **the first nationwide rare disease registration and enrollment study**
- Peking Union Medical College Hospital (**PUMCH**) led a consortium of **20 top-tier teaching hospitals** in China
- It was required to register and study **over 50** types of rare diseases, with at least **50,000** related cases, and establish multi-omics databases and multi-center clinical biobanks. This aimed to provide decision-making basis for precise classification, diagnosis, treatment, and prevention of rare diseases.

National Rare Diseases Registry System of China (NRDRS)

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

FIGURE 1 | The regional distribution map of registered hospitals from NRDRS (as of Mar. 24, 2021). (The National Rare Diseases Registry System of China, 2021).

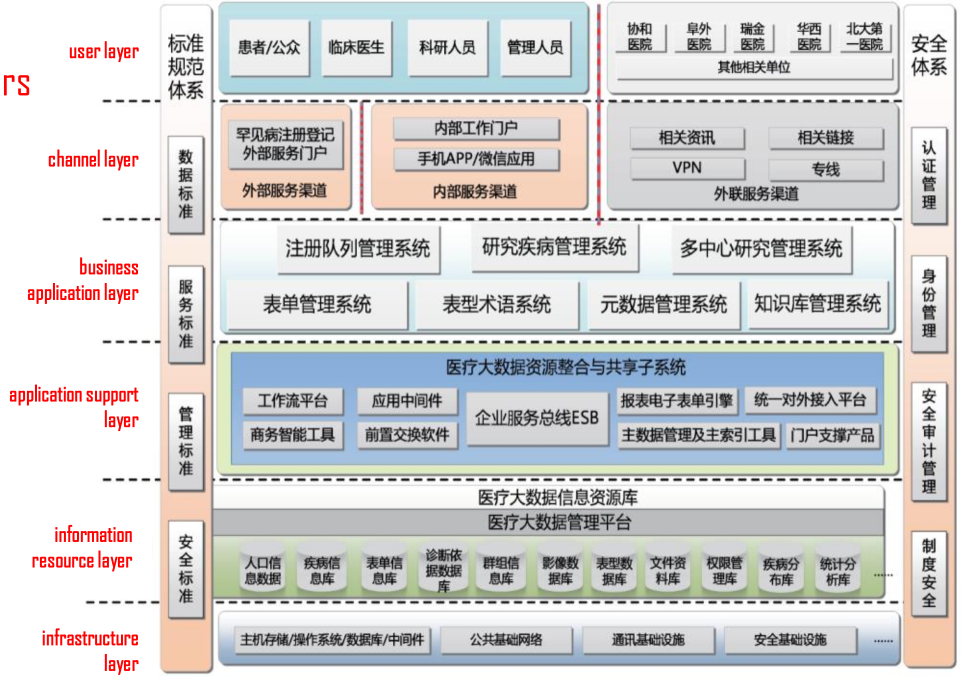
https://www.nrdrs.org.cn/app/rare/index.html

Website Accessible from 2017

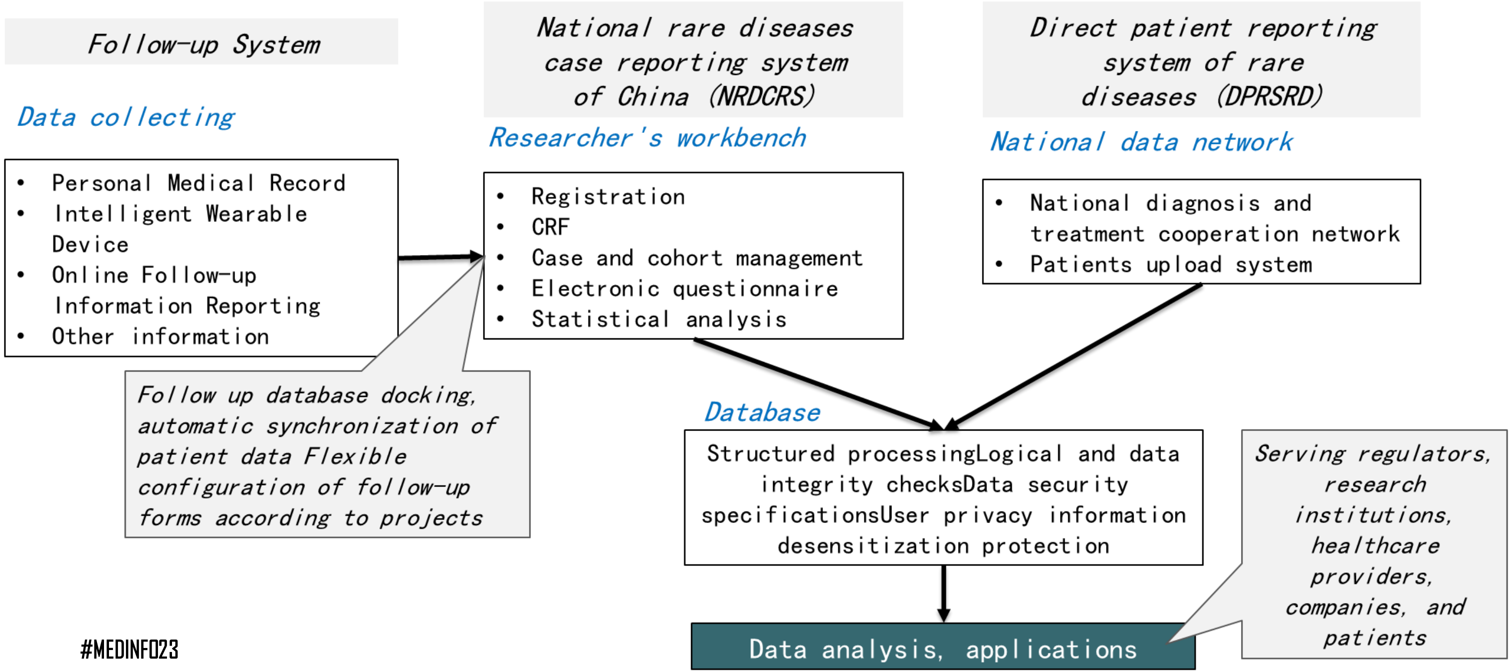
- Functions: Registry + Statistics + Quality control
- 300+ Hospitals
- 200+ Diseases
- 60,000+ Registered Cases
- 1000+ Researchers
- RMB 50m Budget
- From 2017 to 2021

NRDRS architecture

- The overall architecture of the system is composed of six layers from top to bottom.
- establishes national rare diseases clinical data and biological sample database, gathers the advantages of Chinese rare diseases resources, gathers scattered patient and expert resources on one platform , forms an efficient diagnosis and treatment network for rare diseases, and makes in-depth research on disease characteristics and mechanism



Patient-centric data collection solution, one-stop solution for data management



Follow-up System

Data collecting

- Personal Medical Record
- Intelligent Wearable Device
- Online Follow-up Information Reporting
- Other information

Follow up database docking, automatic synchronization of patient data Flexible configuration of follow-up forms according to projects

National rare diseases case reporting system of China (NRDCRS)

Researcher's workbench

- Registration
- CRF
- Case and cohort management
- Electronic questionnaire
- Statistical analysis

Direct patient reporting system of rare diseases (DPRSRD)

National data network

- National diagnosis and treatment cooperation network
- Patients upload system

Database

Structured processingLogical and data integrity checksData security specificationsUser privacy information desensitization protection

Data analysis, applications

Serving regulators, research institutions, healthcare providers, companies, and patients

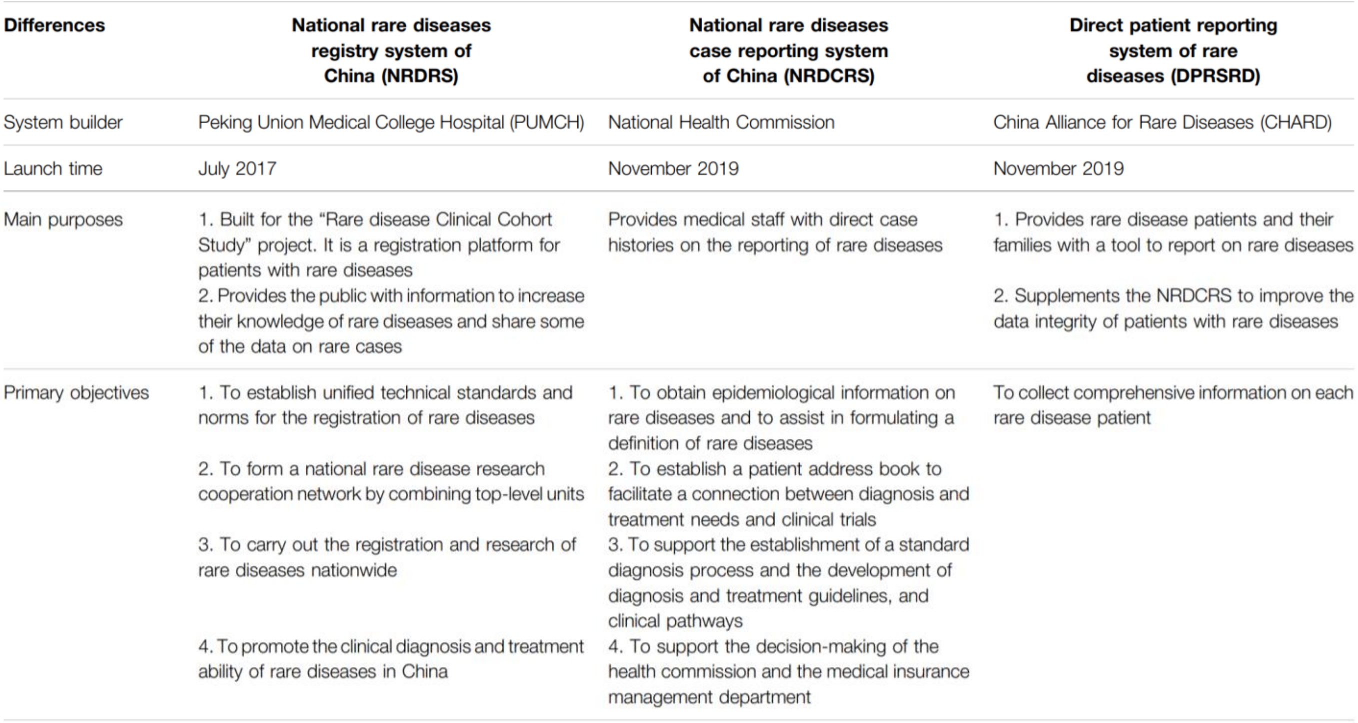
The regional distribution map of registered patients from NRDRS

The largest knowledgebase for
rare diseases in Chinese

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	Castleman Disease
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

FIGURE 2 | The regional distribution map of registered patients from NRDRS (as of Mar. 24, 2021). (The National Rare Diseases Registry System of China, 2021).

The differences between NRDRS, NRDCRS and DPRSRD



Differences	**National rare diseases registry system of China (NRDRS)**	**National rare diseases case reporting system of China (NRDCRS)**	**Direct patient reporting system of rare diseases (DPRSRD)**
System builder	Peking Union Medical College Hospital (PUMCH)	National Health Commission	China Alliance for Rare Diseases (CHARD)
Launch time	July 2017	November 2019	November 2019
Main purposes	1. Built for the "Rare disease Clinical Cohort Study" project. It is a registration platform for patients with rare diseases 2. Provides the public with information to increase their knowledge of rare diseases and share some of the data on rare cases	Provides medical staff with direct case histories on the reporting of rare diseases	1. Provides rare disease patients and their families with a tool to report on rare diseases 2. Supplements the NRDCRS to improve the data integrity of patients with rare diseases
Primary objectives	1. To establish unified technical standards and norms for the registration of rare diseases 2. To form a national rare disease research cooperation network by combining top-level units 3. To carry out the registration and research of rare diseases nationwide 4. To promote the clinical diagnosis and treatment ability of rare diseases in China	1. To obtain epidemiological information on rare diseases and to assist in formulating a definition of rare diseases 2. To establish a patient address book to facilitate a connection between diagnosis and treatment needs and clinical trials 3. To support the establishment of a standard diagnosis process and the development of diagnosis and treatment guidelines, and clinical pathways 4. To support the decision-making of the health commission and the medical insurance management department	To collect comprehensive information on each rare disease patient

The national coordinating strategy for the management of rare diseases

Outcome	**Improving the accessibility of health services for rare diseases**										
↑											
Process	Multicenter research groups	Rare diseases epidemiology		Medical professional training		Patients referral and hierarchical medical system			Expert consensus on HTA of OMP		
↑											
Structure	Informatics infrastructure			Government-led			Multi-stakeholder involvement				
	NRDRS	NRDCRS	Other rare disease information system (e.g. DPRSRD)	Scientific research projects	Policy making (e.g. NNRD)	Direct reporting of rare diseases	Government	Hospitals	Research institutions	Patients	Medical industries
	Knowledgebase construction										
	Patient privacy protection										
	Links to international terms										

Introducing leading international data technology to support the development of rare disease disciplines in China

Delivering SNOMED CT The global language of healthcare

LOINC® from Regenstrief

GTR: GENETIC TESTING REGISTRY All GTR Human Tests

orphanet

GARD Genetic and Rare Diseases Information Center

50 YEARS OMIM Human Genetics Knowledge for the World

GENEReviews

human phenotype ontology

MalaCards HUMAN DISEASE DATABASE

IRDiRC INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

NRDRS provides strong data support for rare disease research in China

- **Rapid Development of Rare Diseases empowered by IT infrastructure Improvement in China**
- **National databases**
- **www.nrdrs.org.cn**

> Front Pharmacol. 2021 Oct 15;12:719415. doi: 10.3389/fphar.2021.719415. eCollection 2021.

Innovation in Informatics to Improve Clinical Care and Drug Accessibility for Rare Diseases in China

Peng Liu [1], Mengchun Gong [2], Jie Li [3], Gareth Baynam [4] [5], Weiguo Zhu [1], Yicheng Zhu [1], Limeng Chen [1], Weihong Gu [1], Shuyang Zhang [1]

Affiliations – collapse

Affiliations

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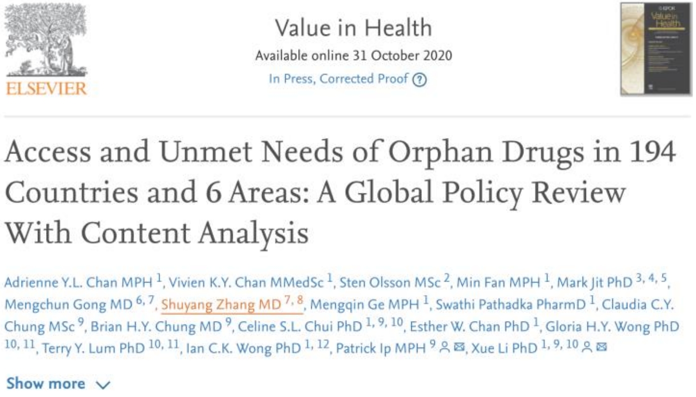
PMID: 34721018 PMCID: PMC8553959 DOI: 10.3389/fphar.2021.719415

Global Policy Evaluation



Value in Health
Available online 31 October 2020
In Press, Corrected Proof

Access and Unmet Needs of Orphan Drugs in 194 Countries and 6 Areas: A Global Policy Review With Content Analysis

Adrienne Y.L. Chan MPH [1], Vivien K.Y. Chan MMedSc [1], Sten Olsson MSc [2], Min Fan MPH [1], Mark Jit PhD [3, 4, 5], Mengchun Gong MD [6, 7], Shuyang Zhang MD [7, 8], Mengqin Ge MPH [1], Swathi Pathadka PharmD [1], Claudia C.Y. Chung MSc [9], Brian H.Y. Chung MD [9], Celine S.L. Chui PhD [1, 9, 10], Esther W. Chan PhD [1], Gloria H.Y. Wong PhD [10, 11], Terry Y. Lum PhD [10, 11], Ian C.K. Wong PhD [1, 12], Patrick Ip MPH [9], Xue Li PhD [1, 9, 10]

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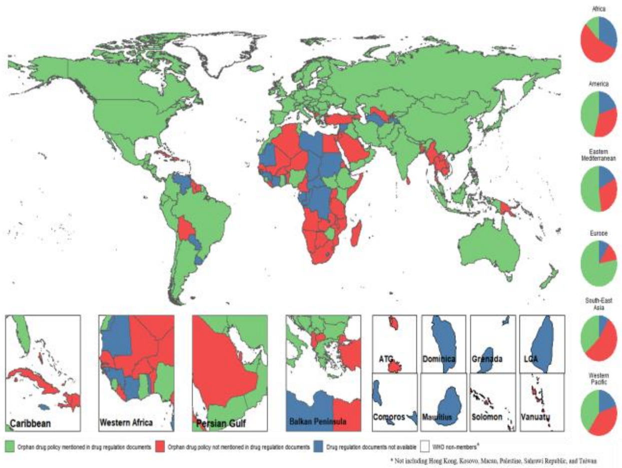

Effort from the Global Alliance to improve patient care for rare diseases patients

REVIEW ARTICLE

Front. Pediatr., 14 December 2020 | https://doi.org/10.3389/fped.2020.579924

Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations

Carla S. D'Angelo[1], Azure Hermes[2], Christopher R. McMaster[3], Elissa Prichep[4], Étienne Richer[5], Francois H. van der Westhuizen[6], Gabriela M. Repetto[7], Gong Mengchun[8], Helen Malherbe[9,10], Juergen K. V. Reichardt[11], Laura Arbour[12], Maui Hudson[13], Kelly du Plessis[10], Melissa Haendel[14], Phillip Wilcox[15], Sally Ann Lynch[16,17], Shamir Rind[18], Simon Easteal[2], Xavier Estivill[19], Yarlalu Thomas[18] and Gareth Baynam[18,20,21,22,23,24,25,26*]

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Comment | Published: 23 December 2019

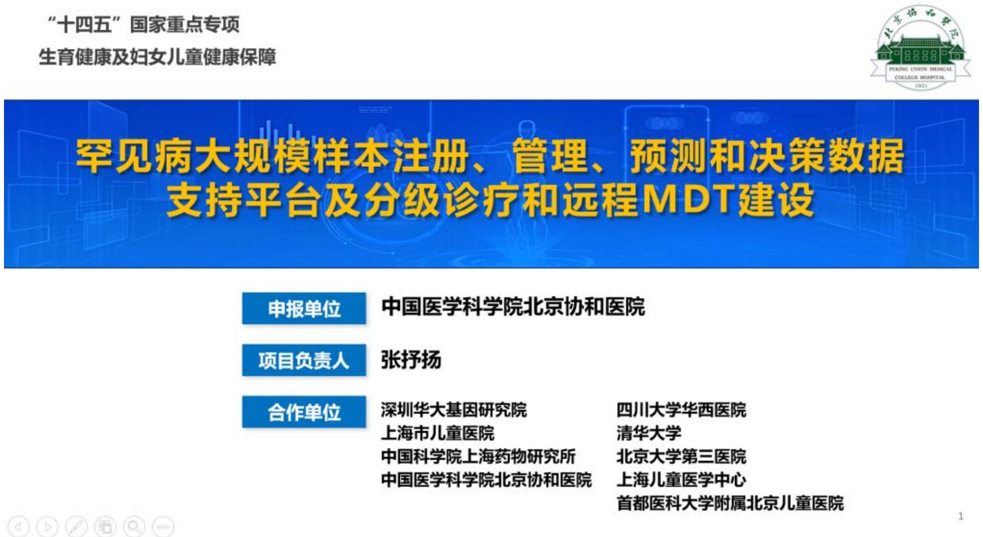
A call for global action for rare diseases in Africa

Gareth S. Baynam, Stephen Groft, Francois H. van der Westhuizen ✉, Safiyya D. Gassman, Kelly du Plessis, Emily P. Coles, Eda Selebatso, Moses Selebatso, Boikobo Gaobinelwe, Tebogo Selebatso, Dipesalema Joel, Virginia A. Llera, Barend C. Vorster, Barbara Wuebbels, Benjamin Djoudalbaye, Christopher P. Austin, Judit Kumuthini, John Forman, Petra Kaufmann, James Chipeta, Désirée Gavhed, Annika Larsson, Maja Stojiljkovic, Ann Nordgren, Emilio J. A. Roldan, Domenica Taruscio, Durhane Wong-Rieger, Kristen Nowak, Gemma A. Bilkey, Simon Easteal, Sarah Bowdin, Juergen K. V. Reichardt, Sergi Beltran, Kenjiro Kosaki, Clara D. M. van Karnebeek, Mengchun Gong, Zhang Shuyang, Ruty Mehrian-Shai, David R. Adams, Ratna D. Puri, Feng Zhang, Nicholas Pachter, Maximilian Muenke, Christoffer Nellaker, William A. Gahl, Helene Cederroth, Stephanie Broley, Maryke Schoonen, Kym M. Boycott & Manuel Posada -Show fewer authors

- International Terminology for Statistics (ICD-11/ORDO) and for Care and Research (SNOMED CT/HPO)
- Higher requirements for Data Infrastructure because of the integration of genomics data

The state continues to invest in NRDRS from 2022 to 2026

Large-scale sample registration, management, prediction and decision support platform for rare diseases and construction of hierarchical diagnosis and treatment and remote MDT



"十四五"国家重点专项
生育健康及妇女儿童健康保障

罕见病大规模样本注册、管理、预测和决策数据支持平台及分级诊疗和远程MDT建设

申报单位 中国医学科学院北京协和医院

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中国医学科学院北京协和医院
四川大学华西医院
清华大学
北京大学第三医院
上海儿童医学中心
首都医科大学附属北京儿童医院

1

"14th Five-Year Plan"
National Key Research and Development Program

- One collaborative network, N disease cohorts
- One intelligent MDT platform
- A number of core technologies to tackle, two national distribution mapping
- Two evaluation systems, three application systems

Summary of construction of national multicentre rare diseases registration system

- Comprehensive analysis of resource advantages, accurate selection of entry angle
- Deep understanding of national development needs and integrated planning to solve urgent problems
- Fully respect the authority of data, win-win cooperation to promote the development of the discipline
- Benchmarking with international technical standards and norms, forming international leading academic results

Insights from the perspective of medical informatics regarding NRDRS

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

- Data Collection and Integration
- Rare Disease Diagnosis Support
- Disease Monitoring and Epidemiological Research
- Patient Management and Resource Integration
- Facilitating Collaboration and Knowledge Sharing