

Online Head: New Chinese databases are a boost for rare-disease science

Chinese databases are a boost for rare-disease science

Rare diseases are a worldwide challenge: there are around 7,000 types and it is estimated that 10% of the global population has one. China is home to the second-largest number of people diagnosed with rare diseases, behind the United States – approximately 20 million, from as many as 56 ethnic groups. Yet data on rare diseases in China are scant: whereas the first official document on such diseases in the United States dates back to 1983, it wasn't until 2018 that China released its first national list of rare diseases.

China is now expediting a data revolution, constructing databases to add to those that currently focus on Western populations. The National Rare Diseases Registry System of China (NRDRS), launched in 2020, integrates research and patient information.

The Rare Disease Data Center, established online in 2022, focuses on using genetic big data for the development of artificial-intelligence tools for bioinformatics. And at the start of this year, the Sixth People's Hospital in Shanghai, China, affiliated with Shanghai Jiao Tong University, initiated a collaboration with University College London to establish the first Chinese–British joint proteomics database for people with Ewing's sarcoma – a type of bone cancer – for which I am a co-investigator.

These initiatives demonstrate China's determination to advance rare-disease science, and I would encourage more scientists and clinicians to join in the construction of these databases.

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[Behind United States with 25-30million RD patients, sourced from National Organization for Rare Disorders (NORD): <https://rarediseases.org/understanding-rare-disease/rare-disease-facts-and-statistics/>]

whereas the first official document on RDs in the United States dates back to 1983, [National Institute of Health Public Law 97-414 97th Congress. 1983 Available online: <https://www.govinfo.gov/content/pkg/STATUTE-96/pdf/STATUTE-96-Pg2049.pdf>] it wasn't until 2018 that China released its first national list of rare diseases. [http://www.gov.cn/zhengce/zhengceku/2018-12/31/content_5435167.htm]