Review

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Epidemiology and distribution of 207 rare diseases in China: A systematic literature review

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SUMMARY Epidemiological data on rare diseases in China are currently limited. The objective of this study was to provide a comprehensive understanding of the prevalence and incidence of rare diseases by systematically analyzing the available epidemiological data. We conducted a comprehensive search of English and Chinese databases, the Incidence and Prevalence Database, the Chinese Rare Disease Guideline, and the Taiwan Health Promotion Administration from 2010 to 2023. We identified the top diseases and regions based on epidemiological data and present the maximum, minimum, and median prevalence and incidence values in tables and forest plots. 1,264 prevalence and incidence data were retrieved from 277 studies, guidelines and official websites, covering 110 rare diseases (53.1%) and 32 regions (94.1%). In terms of geographical regions, incidence or prevalence data were available for 32 regions (94.1%), excluding Tibet Hui Autonomous Region and Macao Special Administrative Region. In terms of rate, 60 and 77 out of 207 diseases (29.0% and 37.2%) had available incidence and prevalence data, respectively. Eight diseases had an incidence rate equal to or greater than that of 1,000 patients per million. The present study provides a comprehensive epidemiological analysis and valuable insights into the prevalence and incidence of rare diseases in China. Our findings underscore the pressing need for sustained drug research and medical support for individuals and families impacted by rare diseases.

Keywords rare disease, epidemiology, China

1. Introduction

Rare diseases (RDs) are a class of diseases characterized by a low incidence, low prevalence, and low total number of patients. RDs encompass a wide range of diseases that exhibit distinct features, such as variable types, inheritance patterns, difficult diagnoses, severe conditions, and low treatability (*1*). Globally, there are more than 7,000 known RDs, with more than 250 million patients affected (*2*). In China, 207 diseases are listed as RDs by the government, affecting an estimated 20 million patients (*3*). RDs place a significant economic burden on patients and their families (*4*). In recent years, the Chinese government has given increasing attention to RDs with the rapid development of the country (*5*). The establishment and release of the National Rare Diseases Registry System of China (NRDRS) and the first batch of RD catalogs are significant milestones toward improving the diagnosis, treatment, and protection of patients with RDs in China (*6-8*). Furthermore, the number of RD drugs covered by medical insurance in China continues to increase, providing patients with greater access to necessary medical care, ultimately leading to better health outcomes and quality of life (*5,6,9*).

The current epidemiological research on RDs in China is insufficient (*1,10,11*). The absence of comprehensive epidemiological data, the availability of scattered and incomplete data, and a lack of systematic retrospective epidemiological analyses are notable issues. To address these shortcomings, this study aimed to systematically summarize the incidence, prevalence and corresponding population information of RDs in China by conducting a thorough search of variable sources, including academic literature on the epidemiology of RDs in China, RD guidelines, and official RD information websites. Through this comprehensive analysis, we aimed to gain a comprehensive understanding of the epidemiology and distribution of RDs in China. Thorough research on RDs requires relevant epidemiological studies as the basis. This study not only provides the basis for basic medicine, clinical medicine, and economic and social research related to RDs but also has the potential to inform the development of a standard consensus on the definition of RDs in this field and has significant implications for

evaluating the RD drug market.

We present this article in accordance with the PRISMA reporting checklist.

2. Materials and Methods

The search process in this study adhered strictly to the Preferred Reporting Items for Systematic reviews and Meta-Analyses (PRISMA) guidelines (*12*).

2.1. Search strategy

The relevant literature published between 1 January 2010 and 31 December 2023 in both English and Chinese databases was identified. The English databases included PubMed and Web of Science. The Chinese databases included SinoMed, China National Knowledge Internet (CNKI), Wangfang database, Vip database. The search strategy utilized a general search string was: disease name [Title/Abstract/Text Word] AND (prevalence [Title/Abstract/Text Word] OR incidence [Title/Abstract/Text Word]). Additionally, we supplemented our search with data from Incidence and Prevalence Database® (IPD), as well as from Guidelines for Diagnosis and Treatment of Rare Diseases (2019 edition) and the Taiwan Health Promotion Administration. This approach was taken to avoid missing relevant epidemiological data. The full details of the search strings are provided in Supplemental Table S1 (*http://www.irdrjournal.com/action/getSupplementalData.php?ID=195*).

2.2. Eligibility criteria

Reviews, letters to the editor, case reports, case studies, original research literature, academic dissertations and conference abstracts were included. The inclusion and exclusion criteria are shown in Figure 1.

2.3. Data extraction

The data were extracted using Microsoft Excel 2019. The target data were categorized into three parts: basic epidemiological data, additional data and notes. *i*) Basic epidemiological data included the disease name, the available ICD-10 codes, the subcategory of disease, the incidence and prevalence, and the region, age, sex, and ethnicity of the patients with corresponding epidemiological data. *ii*) Additional data included the number of participants, the observation period and the year of publication. *iii*) Notes included the data remarks (whether the data were standardized), literature remarks (whether the study was a retrospective analysis, systematic review, meta-analysis or original research), and citations of the literature, literature links and data sources (literature or guidelines). The data were retrieved from the aforementioned sources by two team members. The results were subsequently summarized on a single table using the "entry tested twice" approach to ensure accuracy. Finally, a third member reviewed the table for further verification.

2.4. Statistical analysis

Forest plots were generated based on the maximum and minimum rates of the disease. However, if there were at least three data points available and all of them were precise values rather than ranges, the median value was also included in the plot. All the forest plots were generated using GraphPad Prism software, version 8.0 (San Diego, California, USA).

The search strategy summary can be seen in Table 1.

3. Results

A total of 1,264 incidence and prevalence data points

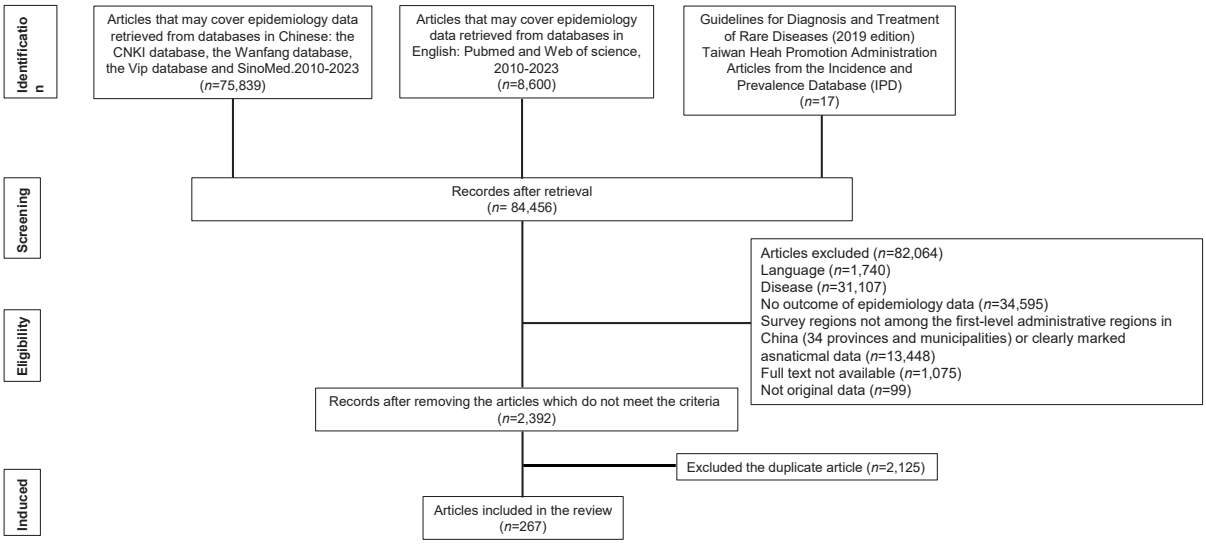

Figure 1. Study flow diagram. Abbreviations: CNKI, China National Knowledge Infrastructure.

Table 1. The search strategy summary

Items	Specification
Date of search (specified to date, month and year)	From 1 January 2010 to 31 December 2023.
Databases and other sources searched	PubMed, Web of Science, SinoMed, China National Knowledge Internet (CNKI), Wangfang database, Vip database, Incidence and Prevalence Database® (IPD), the Chinese Rare Disease Guideline (2019) and the Taiwan Health Promotion Administration.
Search terms used (including MeSH and free text search terms and filters)	Please see Supplementary Table S1.
Timeframe	Last retrieval on 11 February 2024.
Inclusion and exclusion criteria (study type, language restrictions, *etc.*)	Inclusion criteria: the literature may cover epidemiology data retrieved from databases above which published from 2010 to 2023. Exclusion criteria: the literature without outcome of epidemiology data, survey regions among the 34 regions original data.
Selection process (who conducted the selection, whether it was conducted independently, how consensus was obtained, *etc.*)	The data were extracted using Microsoft Excel 2019. The data were retrieved from the aforementioned sources by two team members. The results were subsequently summarized on a single table using the "entry tested twice" approach to ensure accuracy. Finally, a third member reviewed the table for further verification.

were extracted from 277 literature sources that met the retrieval requirements (Table 2, online data, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=195*).

Among the diseases included in the study, 110 of the 207 diseases examined (53.1%) had available incidence or prevalence data. The five diseases with the largest number of cumulative incidence and prevalence data points were Phenylketonuria (374 data points), Melanoma (220 data points), Multiple Sclerosis (123 data points), Hepatolenticular Degeneration (Wilson Disease) (43 data points) and Malignant pleural mesothelioma (30 data points).

In terms of geographical regions, incidence or prevalence data were available for 32 regions (94.1%), excluding Tibet Hui Autonomous Region and Macao Special Administrative Region. The five regions with the highest number of cumulative incidence and prevalence data points were Taiwan Province (143 data points), Zhejiang Province (83 data points), Inner Mongolia Autonomous Region (81 data points), Liaoning Province (62 data points), and Qinghai Province (58 data points). On the other hand, the five regions with the lowest number of cumulative incidence and prevalence data points were Tibet Hui Autonomous Region (zero data point), Macao Special Administrative Region (zero data point), Hubei Province (one data points), Jiangsu Province (two data points), and Heilongjiang Province (three data points).

According to the 1,264 incidence and prevalence data, 502 (39.7%) were related to the newborn population. Among the 207 diseases considered, 36 were classified as an Inherited Metabolic Disease (IMD) (*27*), and only four out of these 36 IMDs (Hereditary Hypomagnesemia, Hyperornithinaemia-Hyperammonaemia-Homocitrullinuria, *N*-acetylglutamate Synthase Deficiency, Very Long Chain Acyl-CoA Dehydrogenase Deficiency and X-linked Agammaglobulinemia) lacked any available incidence or prevalence data (Table 3).

3.1. Incidence

A total of 921 incidence data points were extracted from 277 literature sources that met the retrieval requirements.

Regarding the scope of the study, 60 out of 207 diseases (29.0%) had available incidence data. The five diseases with the highest number of reported incidence data were Phenylketonuria (372 data points), Gastrointestinal stromal tumor (35 data points), Malignant pleural mesothelioma (30 data points), Hepatolenticular Degeneration (Wilson Disease) (24 data points), and Methylmalonic Academia (nineteen data points).

Regarding geographical regions, incidence data were available for 32 out of 34 regions (94.1%). The five regions with the largest number of reported incidence data points were Taiwan Province (59 data points), Beijing (43 data points), Inner Mongolia Autonomous Region (36 data points), Shanghai (31 data points) and Xinjiang Uygur Autonomous Region (24 data points). The five regions with the lowest number of cumulative incidence data points were Tibet Hui Autonomous Region (zero data point), Macao Special Administrative Region (zero data point), Hubei Province (one data points), Jiangsu Province (two data points), and Heilongjiang Province (three data points).

Additionally, there were eight diseases with an incidence equal to or greater than 1,000 patients per million (ppm): Homocysteinemia (275,000 ppm), Retinopathy of prematurity (179,000), Retinoblastoma (2,121 ppm), Non-Syndromic Deafness (1,860 ppm), Phenylketonuria (1,480 ppm), Isovaleric Acidemia

Table 3. Lists of inherited metabolic diseases among the 121 rare diseases

Disease Name	Number of incidence data point	Number of prevalence data point
21-Hydroxylase Deficiency	3	0
Arginase Deficiency	1	0
Beta-ketothiolase Deficiency	7	0
Biotinidase Deficiency	0	1
Carnitine Deficiency	10	6
Citrullinemia	5	2
Galactosemia	4	2
Glutaric Acidemia Type I	4	2
Hereditary Fructose Intolerance	1	0
Hereditary Hypomagnesemia	0	0
Holocarboxylase Synthetase Deficiency	1	1
Homocysteinemia	2	13
Homozygous Hypercholesterolemia	0	1
Hyperornithinaemia-Hyperammonaemia-Homocitrullinuria Syndrome	0	0
Hyperphenylalaninemia	1	0
Hypophosphatasia	1	2
Isovaleric Acidemia	8	1
Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency	1	0
Lysinuric Protein Intolerance	1	0
Lysosomal Acid Lipase Deficiency	0	1
Maple Syrup Urine Disease	7	4
Medium Chain Acyl-CoA Dehydrogenase Deficiency	9	8
Methylmalonic Academia	13	7
Multiple Acyl-CoA Dehydrogenase Deficiency	2	0
N-acetylglutamate Synthase Deficiency	0	0
Neonatal Diabetes Mellitus	0	1
Ornithine Transcarbamylase Deficiency	2	0
Phenylketonuria	329	0
Porphyria	0	1
Progressive Familial Intrahepatic Cholestasis	0	1
Propionic Acidemia	0	2
Sitosterolemia	0	1
Tetrahydrobiopterin Deficiency	1	1
Tyrosinemia	0	2
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	0	0
X-linked Agammaglobulinemia	0	0

(1,207.6 ppm), Maple Syrup Urine Disease (1,107.0 ppm), and Noonan Syndrome (1,000 ppm).

The five diseases with the widest range of reported incidence values were Homocysteinemia (range: 274,996.7 ppm), Retinopathy of prematurity (range: 86,160 ppm), Retinoblastoma (range: 2,120.3 ppm), Phenylketonuria (range: 1,471 ppm) and Isovaleric Acidemia (range: 1,207.0 ppm) (Figure 2A).

3.2. Prevalence

A total of 343 prevalence data points were extracted from 277 literature sources that met the retrieval requirements.

Within the scope of the study, 77 out of 207 diseases (53.7%) had available prevalence data. The five diseases with the highest number of reported prevalence data points were Multiple Sclerosis (87 data points), Hepatolenticular Degeneration (Wilson Disease) (24 data points), Hemophilia (24 data points), Hidradenitis suppurativa (fifteen data points), Homocysteinemia (thirteen data points) and Methylmalonic Academia (eight data points).

Regarding geographical regions, prevalence data were available for nineteen out of 34 regions (55.9%). The five regions with the largest number of reported prevalence data points were Taiwan Province (69 data points), Zhejiang Province (58 data points), Liaoning Province (47 data points), Guangdong Province (47 data points), and Inner Mongolia Autonomous Region (46 data points). On the other hand, there were 15 provinces, autonomous regions and municipalities (44.1%) that lacked any reported prevalence data. These locations were Tibet Autonomous Region, Macao Special Administrative Region, Shanxi Province, Jilin Province, Heilongjiang Province, Jiangsu Province, Hubei Province, Sichuan Province, Guizhou Province, Yunnan Province, Shaanxi Province, Gansu Province, Ningxia Hui Autonomous Region, Xinjiang Uygur Autonomous Region, and Hainan Province.

Additionally, the five diseases with the highest reported prevalence rates were Homocysteinemia (743,962.5 ppm), Retinopathy of prematurity (128,000 ppm), Primary biliary cholangitis (4,150.5 ppm), Hidradenitis suppurativa (1,856 ppm) and Fragile X syndrome (800 ppm).

The five diseases with the widest range of reported

prevalence values were Homocysteinemia (range: 5,825,962.5 ppm), Primary biliary cholangitis (3,980.5 ppm), Hidradenitis suppurativa (range: 1,750 ppm), Phenylketonuria (range: 281.6 ppm) and Congenital Hyperinsulinemic Hypoglycemia (range: 229.1 ppm) (Figure 2B).

4. Discussion

4.1. Basic epidemiological information

This study examined 207 RDs and found that 97 (46.9 %) lacked data on disease incidence and prevalence. Among the 110 diseases with available incidence or prevalence

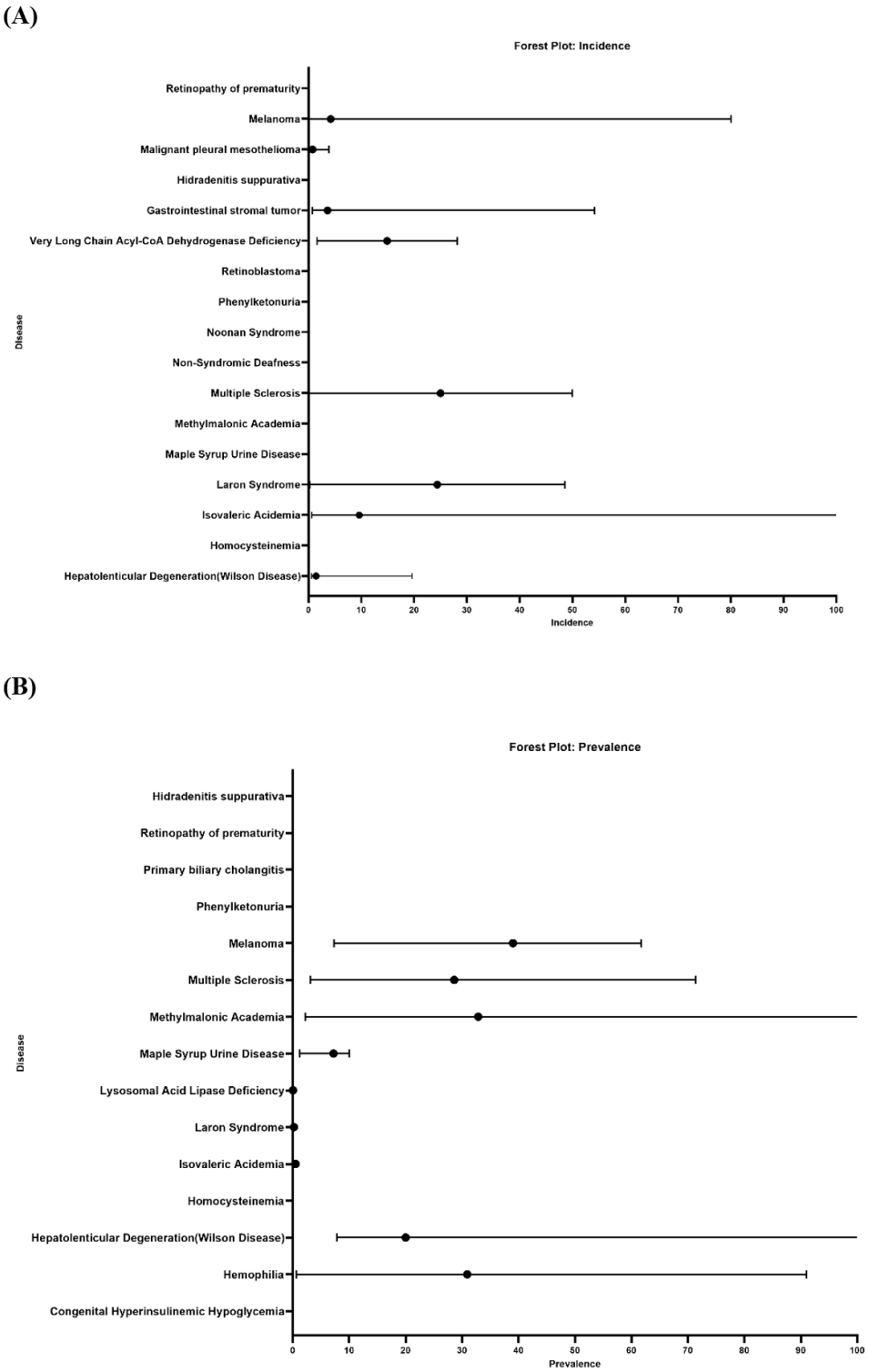

Figure 2. Incidence and prevalence rates of RDs. (A) Incidence rates of RDs; **(B)** Prevalence rates of RDs. Abbreviations: RDs, rare diseases; PPM, patients per million. *Notes:* The black points represent the medians, and the two ends of the line represent the extreme values. A line is displayed when the amount of data is no less than 3 and the data are not approximately within the range. The data were not available if there are no data for the disease.

data, 38 had only one prevalence value, and 14 had only one incidence value. These findings indicate a significant amount of missing disease and regional epidemiological data. As we can see from the results, there are currently several challenges in the epidemiological study of RDs in China. Although the Chinese government has released the two RD catalogs, there is still no universally recognized standard for defining which is RDs (*280*). This lack of standardization could be attributed to two main factors: insufficient epidemiological information on RDs and heterogeneity in the existing incidence and prevalence data (*281,282*).

4.2. The reasons for insufficient epidemiological information and heterogeneity

The reasons for the lack of epidemiological data on RDs are as follows. First, there are few patients of RDs, resulting in insufficient research for this topic. Second, RDs are difficult to diagnose (*62*). Third, the RD management system (NRDRS) in China has not yet been covered nationwide (*5*). Statistics concerning patient household registration and place of medical treatment are incomplete, increasing the regional bias caused by remote medical treatment (*5,11*).

Furthermore, substantial heterogeneity in the incidence or prevalence of the same disease was observed across different regions or populations. For instance, eight diseases (6.6%) had an incidence or prevalence rate exceeding 1,000 ppm. The disease with the highest incidence rate was Homocysteinemia (2-390,800 ppm), and this disease also had the highest prevalence rate Homocysteinemia (37.5-744,000 ppm). Moreover, heterogeneity was not limited to different diseases but was evident within the same disease. For example, Lysosomal Acid Lipase Deficiency had a prevalence of 0.04 ppm, which is lower than that of Homocysteinemia. Similarly, incidence of Laron Syndrome was 0.2 ppm, which was lower than the maximum incidence of Homocysteinemia. Heterogeneity exists in terms of epidemiological rates and data volume in different regions due to differences in prevalence and allopathic treatment (*283*). The findings show that the greater the level of social development is, the greater the proportion of patients treated, resulting in a lower rate than that reported in previous studies (*283*). These variations further complicate the establishment of a unified definition standard for RDs.

To address these challenges and enhance academic quality, this study systematically collected and summarized incidence and prevalence data, along with corresponding population information such as region, sex, age, and race. By consolidating scattered epidemiological information, researchers can more easily access the essential epidemiological details of RDs. This comprehensive analysis also aids researchers in identifying RDs with high heterogeneity in incidence or prevalence rates. Furthermore, by considering the population-specific characteristics associated with the data, this study provides valuable insights into the number of patients affected and serves as a reference for reaching a consensus on the definitions of RDs through epidemiological data.

4.3. Research and development of RD drugs

Although significant progress has been made in the development of RD drugs in China, there are still diseases for which there is no reference indication for drug treatment. Currently, based on data from the Pharnexcloud (*https://data.pharnexcloud.com/*, a website providing Chinese drug information) and our research findings, out of the 110 diseases with available incidence or prevalence data, only 43 (39.1%) have drugs with reference indications. In contrast, among the 97 diseases without any incidence or prevalence data, only fifteen (15.5%) have drugs with reference indications. This finding suggested a positive correlation between the completeness of epidemiological information for RDs and the availability of corresponding drugs (*284*).

This phenomenon can be attributed to several factors. First, the acquisition of epidemiological data serves as the initial step in the research and development of drugs. For RDs without any available drugs, the lack of systematic epidemiological data prevents pharmaceutical companies from adequately evaluating market demand and potential profitability. Consequently, limited commercial viability restricts the investment and production capabilities of pharmaceutical companies in the field of RDs (*5*).

Second, comprehensive epidemiological studies in terms of efficacy play a crucial role in identifying a larger patient population. Only through epidemiological studies on patient cohorts can the effectiveness of drugs be determined, thus facilitating the transition of new drugs from clinical trials to the market (*285*). This emphasizes the importance of regular epidemiological investigations and summaries of the development of RD drugs. Given that there are still 95 RDs in China without any drugs with reference indications, this study collected epidemiological information for 85 diseases, thereby providing assistance for the development of drugs for at least 53 RDs. From the perspective of benefiting patients with RDs, the introduction of new drugs enables the possibility of treatment.

However, the key to alleviating the economic burden on these patients lies in the inclusion of RD drugs in medical insurance coverage. In 2019, Zhejiang Province became the first region in China to establish a provincial drug security system for RDs (*286*). The inclusion criterion for RD drugs in medical insurance was epidemiological information obtained from the neonatal

disease screening information system. It is worth noting that among the 112 drugs for which RD was used as a reference indication, 57 (50.9%) are still not covered by medical insurance, indicating room for improvement in the inclusion rate of RD drugs in medical insurance schemes. By collecting epidemiological information on 207 RDs nationwide, this study provides a valuable reference for the inclusion of RD drugs in medical insurance. He *et al*. noted that if RDs identified can be included only in medical insurance, it will strongly promote patients to apply for certification and doctors to report diseases (*10*). Furthermore, it offers support for disease identification, aiding in the expansion of regional and disease-specific RD medical security systems (*287*).

4.4. Future challenges and opportunities of RDs in China

However, there are still limitations in our research. The amount of epidemiological data is currently limited, and the data cannot be critically analyzed due to the late start of RD research in China (*1,10,11*). However, the findings of this study significantly contribute to filling the gaps in epidemiological information concerning RDs in China. These gaps include 52 diseases with only one reported incidence or prevalence value within a single region, as well as 106 RDs that lack any recorded incidence or prevalence values in the initial RD catalog. Additionally, it is crucial to prioritize epidemiological research on diseases that may be included in future iterations of the RD catalog. By conducting thorough epidemiological raw data research and aggregating the data, we can not only provide valuable data references but also facilitate dynamic updates of the RD catalog (*287*). As the amount of epidemiological data continues to increase, additional high-quality information can be obtained in the future. The epidemiological data identified using this approach could be included in further studies. This approach will enhance the academic quality of our understanding and management of RDs in China.

5. Conclusion

This study aimed to enhance the academic quality of epidemiological information and drug marketing assessment for RDs in China, based on the two RD catalogs.

By providing a systematic and comprehensive data reference, this study contributes significantly to the understanding of epidemiological information and facilitates accurate assessments of the RD drug market.

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Conflict of Interest: The authors have no conflicts of interest to disclose.

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