Uncovering Worldwide Real-World Data Sources for Fabry Disease

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BACKGROUND

- Fabry disease (FD) is a rare X-linked lysosomal disorder characterized by the abnormal accumulation of lipids in tissues.[1] It is a progressive, debilitating condition that can be life-threatening if not managed properly.[2,3]
- In recent years, real-world data (RWD) has become increasingly valuable for stakeholders to understand the burden of the disease and assess treatment outcomes in everyday clinical settings.[4]
- The objective of this study is to identify and map the existing RWD sources of FD, addressing the gap in comprehensive global data availability.

METHODS

- A targeted literature review was conducted using Embase and Medline, focusing on publications from last 3 years (Jan 2022 to May 2024). Clinical trials, systematic and narrative reviews, and commentaries were excluded. Abstracts identified were screened for relevance at the title/abstract and then full-text review.
- Real-world data (RWD) sources were categorized into 3 groups:
  - ✓ Studies and registries (i.e., observational studies, health surveys, interviews, and registries)
  - ✓ Clinical records (i.e., electronic medical records, case notes, administrative records, and claims)
  - ✓ Unsupervised sources (i.e., personal devices, smartphones, and social media)
- Data related to disease characteristics, patient demographics, and treatment specifics were extracted.

RESULTS

- The literature search provided 239 publications. Of these, 80 publications (33%) relating to 65 potentially unique RWD sources were included.
- RWD sources majorly comprised studies and registries and clinical records. None of the publications used any unsupervised sources (**Fig. 1**).
- FD RWD sources were primarily from EU4 countries (n=14, 22%), followed by Asia (n=11, 17%), USA and Canada (n=10, 15%), the UK (n=5, 8%), and Latin America (n=4, 6%; **Fig. 2**). Key registries identified are shown in **Fig. 3**.
- Most studies (75%) reported demographics and baseline data of patients. The number of patients reported across the data sources varied widely, ranging from 1 to 10,637 patients.
- These sources encompassed a wide range of FD aspects, including differential diagnosis, genetic screening, comorbidities, disease complications and burden, quality of life, patient-reported outcomes, and treatment effectiveness and safety (**Fig. 4**). However, the comorbidities most frequently examined in the studies were renal, cardiac, and vascular conditions.

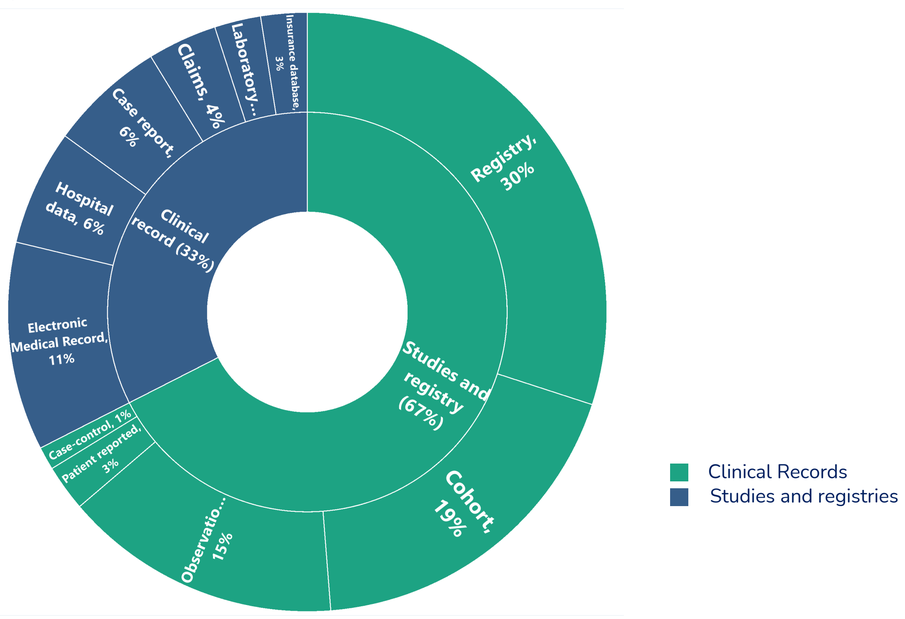

Figure 1: Distribution of RWD data sources by data types

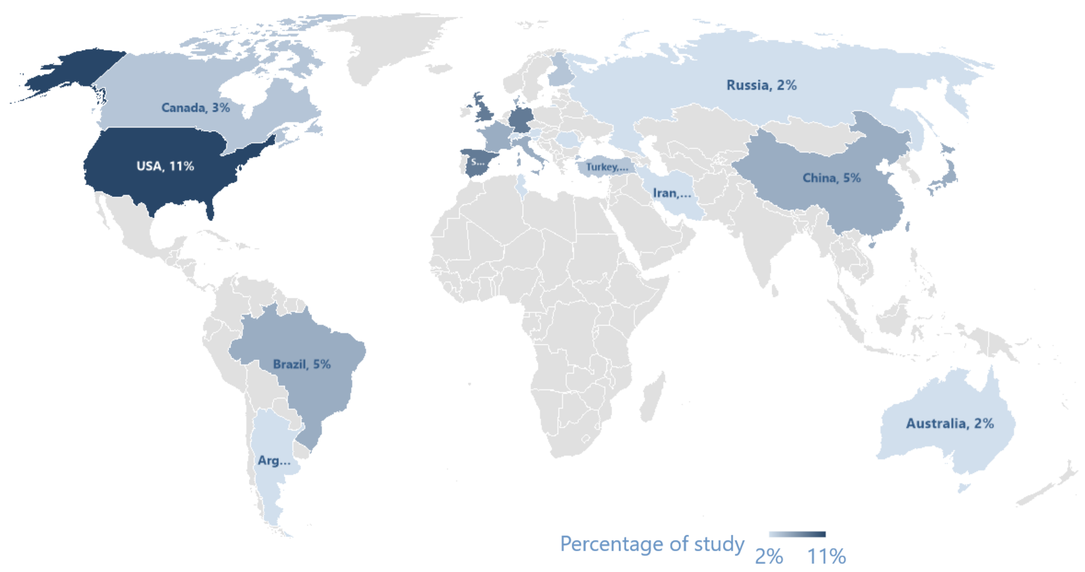

Figure 2: Global distribution of studies by percentage

This study identified Fabry Disease RWD sources that exist globally and can be leveraged to identify disease patterns, evaluating treatment effectiveness, and capturing patient experiences in the real-world setting that differ from controlled clinical trials.

By incorporating these diverse data sources into research frameworks, healthcare professionals can develop personalized and effective treatment strategies, ultimately enhancing the management of Fabry Disease and improving patients' quality of life.

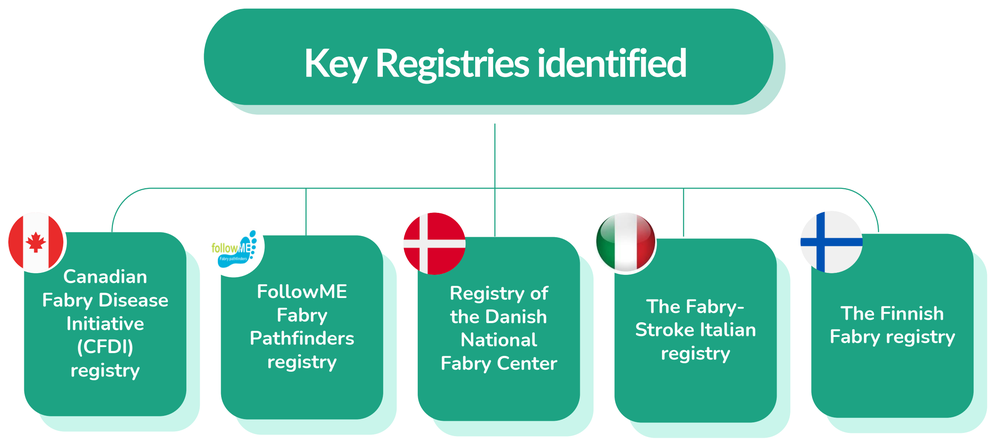

Figure 3: Key unique registries identified

Figure 4: FD themes covered across included study

DISCUSSION

- The existence of varied numerous registries is invaluable for collecting extensive information from around the world. Studies involving substantially large populations with demographic details contribute significantly to developing personalized treatment approaches. Currently, a primary areas of interest that was observed during this study is managing complications related to the kidneys, heart, and vascular systems.
- This review was conducted utilizing only the Embase and MEDLINE databases. While these sources provide a substantial volume of information, the exclusion of other relevant databases may result in an important amount of pertinent literature being overlooked. Despite these constraints, this review contributes valuable insights into the current body of work in the field, highlighting the ongoing efforts to extract RWD related to FD. Such an analysis aids in understanding the landscape of research and the progress made toward elucidating the complexities of this condition. This review lays the groundwork for future research initiatives, paving the way for innovative personalized treatment strategies tailored to the unique needs of individuals with FD.

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