

Rare kidney disease registries: A scoping review on characteristics and lessons learnt

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Abstract

Background: Due to their low prevalence and wide distribution, rare kidney diseases pose challenges in diagnosis and management. Registries play a key role in organising data to address these issues. **Objectives:** This study aims to compare the characteristics of existing rare kidney disease registries to gain insights to create new registries or develop available ones. **Method:** This scoping review was conducted in 2024. The searches were performed in four electronic databases: PubMed, Web of Science, Scopus and ProQuest, as well as the Orphanet website. Various components of disease registries were extracted and compared narratively. **Results:** Our review of 1,534 papers revealed 29 rare kidney disease registries, and an additional 8 registries were retrieved from the Orphanet website, established between 1989 and 2023 in various countries. The objectives of these registries were categorised into clinical (15 registries), research (29 registries), and epidemiological ones (11 registries), among these, 15 and 20 registries were international and national, respectively. Data collected included demographic, medical, and family history, paraclinical, therapeutic, genetic testing, comorbidities and treatment outcomes. Various strategies were employed for data quality assurance, including the use of data quality indicators, audits and adherence to established guidelines. Some registries used both prospective and retrospective data collection methods. **Conclusion:** Establishing standardised registries for rare kidney diseases is essential to ensure data consistency and comparability. Due to legal and ethical challenges, and regional differences in disease prevalence, each country or institution may need to create its own registry. Adhering to common standards for data collection, collaboration and establishment of integrated frameworks facilitate comprehensive and comparable data analysis. **Implication for health information management practice:** This study suggests that rare kidney disease registries should adopt patient-centred designs with clearly defined objectives and standardised data elements to enable meaningful data collection and analysis. Registries must implement strong data quality assurance mechanisms and foster inter-institutional collaboration to support clinical decision-making, and drive targeted research.

Keywords (MeSH)

kidney diseases; rare diseases; surveillance; public health surveillance; data quality; data collection; registries; health information management

Supplementary keywords

disease registry; patient registry

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Introduction

Rare kidney diseases encompass more than 150 distinct conditions with an estimated prevalence of 60–80 cases per 100,000 individuals in the United States and Europe (Devuyst et al., 2014). While each rare kidney disease presents its unique challenges, they pose significant hurdles in diagnosis and treatment, including the limited number of affected individuals, unknown disease etiologies, lack of biomarkers for monitoring disease progression, and the necessity for complex care strategies (Aymé et al., 2017). Rare kidney diseases play a significant role in the global burden of kidney diseases. Over 25% of adults and more than 50% of children with kidney failure are affected by these rare conditions. Nevertheless, high-quality published data on the incidence of these diseases and the specific patient groups they impact remains limited. Consequently, patients often encounter delays in diagnosis and have difficulty accessing reliable information regarding their condition after diagnosis (Wong et al., 2023).

Patient registries are organised systems that systematically gather consistent data aimed at evaluating specific outcomes for populations affected by particular diseases or conditions. These registries are valuable tools for outcome assessment, particularly when randomised controlled trials present challenges. Registries are instrumental in tackling a significant obstacle in rare disease research, namely the limited sample sizes. Given the low prevalence of rare diseases, patient data are frequently sparse. However, the development of large databases facilitates collaboration and data sharing among various centres and countries, thereby enhancing research opportunities (Hageman et al., 2023).

Patient registries for rare diseases are considered essential tools for obtaining a sufficient sample size for clinical research, guiding healthcare planning, and promoting the development and assessment of diagnostic and therapeutic treatments. These registries play a crucial role in evidence-based personalised medicine for rare diseases, as they serve various purposes, including enhancing case definitions, revising disease classifications, evaluating treatment indications and risk stratification, as well as assessing the safety, effectiveness, feasibility, limitations and advantages of diagnostic and therapeutic approaches in real-world settings (Kölker et al., 2022). There has been significant growth in rare disease registries (RDRs) in recent years. According to Orphanet, Europe boasts over 812 RDRs (Andrunyk et al., 2018). These registries serve a variety of purposes, from managing patient clinical data to addressing epidemiological and research goals (Kodra et al., 2018). Implementing RDRs significantly contributes to understanding these conditions and providing information for clinical trials. RDRs enhance the quality and efficiency of clinical studies by offering larger sample sizes. In addition, RDRs are a vital source of data for monitoring safety and efficacy of drugs in post-marketing surveillance (Lazem et al., 2021). Although RDRs play a main role in clinical care and directing research forward, they exhibit considerable variation in structure, operation, and quality approaches. These discrepancies can influence their effectiveness as a resource for informing decisions

about the approval and funding of new treatments for rare diseases (Tarride et al., 2024).

A systematic review by Hageman et al. (2023) highlighted the challenges associated with RDRs with a focus on issues related to their design, quality management and maintenance. One of the primary difficulties in establishing these registries was their inherent complexity. Additionally, the rarity of these diseases makes it especially challenging to recruit a sufficient number of participants to collect meaningful data (Hageman et al., 2023). Data quality also represents a significant concern because only a limited number of registries implement comprehensive quality management programmes. Therefore, inconsistencies in data quality and reliability are common issues (Hageman et al., 2023).

Although international registries can be valuable, legal, ethical, and systemic differences across countries often make them difficult to establish and maintain (Roundtable on Environmental Health Sciences Research, and Medicine et al., 2016). As a result, many countries have developed unique registries for rare kidney diseases. In the absence of a global standard for rare kidney disease registries, having a practical guide can help ensure that independently developed registries remain comparable and interoperable. Therefore, this study aimed to compare the characteristics of available rare kidney disease registries to gain insights to create new registries or develop available registries. Findings from this study could provide valuable insights for the design and implementation of rare kidney disease registries and might help establish a standard in this important area.

Method

This scoping review was conducted based on the Preferred Reporting Items for Systematic reviews and Meta-Analyses (PRISMA) guideline. A scoping review approach was selected because it enables the synthesis of results from various study types and designs, offering a comprehensive overview of a subject. We reported the study based on PRISMA Extension for Scoping Reviews (PRISMA-ScR) checklist, which provides detailed guidance for reporting scoping reviews (Supplemental Figure S1; Tricco et al., 2018).

Protocol and registration

The protocol for this scoping review was registered at our university (Iran University of Medical Sciences); however, it was not registered in any international platform.

Eligibility criteria

Any quantitative or qualitative studies and registry reports related to the establishment and various dimensions of rare kidney disease registries were included in this review. However, studies and reports that were related to registries but only provided statistical data without detailed information on the operational aspects of the registries were excluded. Studies published until 2024 were considered eligible for inclusion. Reports published in any language were also eligible. Additionally, biobanks and registries without sufficient publicly accessible details were excluded.

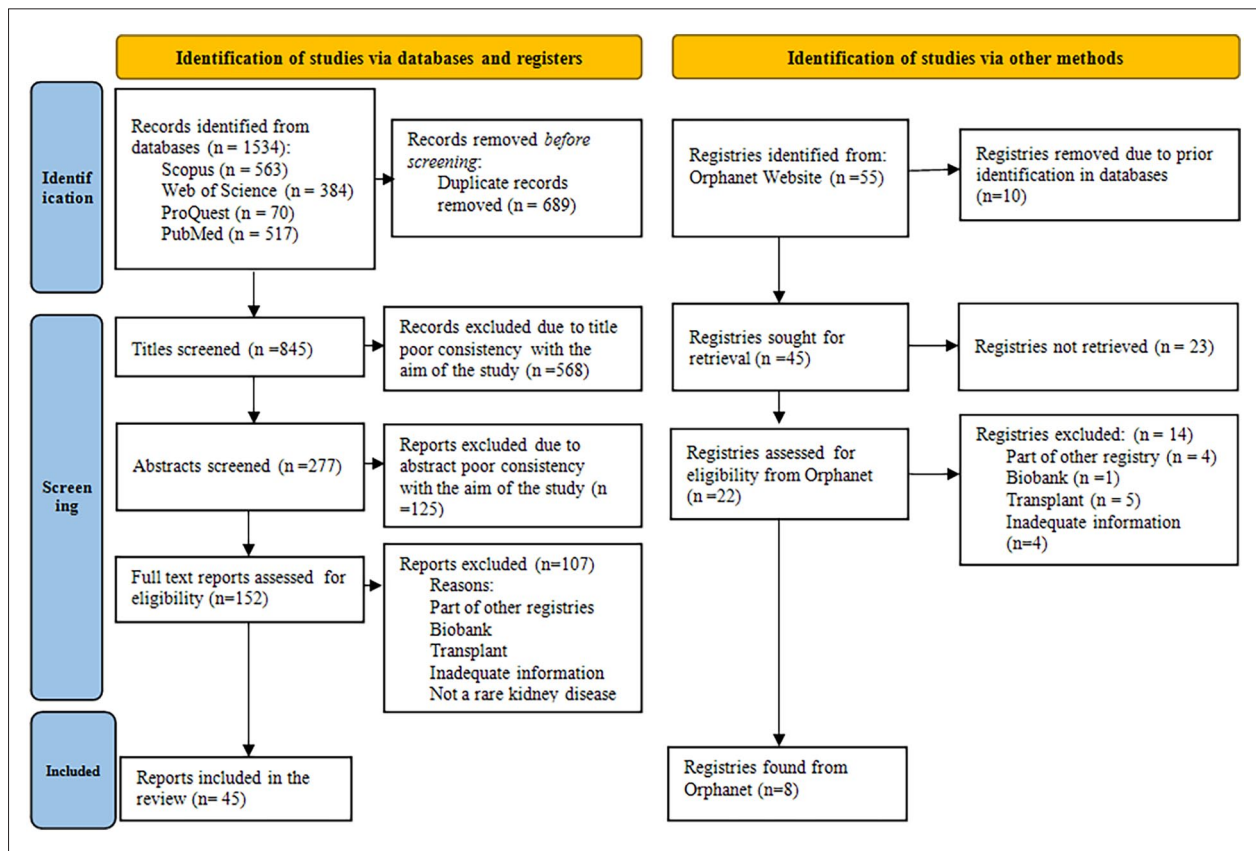


Figure 1. The process of registry identification.

Information sources and search strategy

Several electronic databases, including Web of Science, PubMed, Scopus and ProQuest, were searched by one of the researchers (ZM). The search strategy in the electronic databases utilised two groups of keywords related to rare kidney diseases and registries (the complete search strategy is available in Supplemental Box S1). The databases were searched from inception until 2024. The list of rare kidney disease registries available on the European Orphanet website was also examined. On this website, ZM and AS searched the names of all rare kidney diseases and reviewed any available corresponding registries. Furthermore, ZM and AS screened the reference lists of selected studies to ensure that all pertinent registries were included.

Selection of sources of evidence

The retrieved articles and reports were imported into EndNote, where duplicates were eliminated. Two researchers (ZM and AS) separately screened all articles and reports based on their titles, abstracts, and full texts, resolving any disagreements through discussion. Both researchers also participated in the second screening of the full texts.

Data items, data charting and synthesis

We developed a data extraction form in an Excel spreadsheet. The extracted data included the registry name, establishment year, country, geographical coverage, disease

name, objectives, steering committee members, data set, biological samples, patient-reported outcomes, data quality assessment strategy, inclusion and exclusion criteria, registrars, data collection methods and data sources. This data extraction form was created based on previous studies (Hageman et al., 2023; Lazem et al., 2021). Two independent reviewers (ZM and AS) manually extracted data from the included studies. Information on the identified registries was extracted from both included studies and relevant registry websites. Any discrepancies between the reviewers were resolved through discussion, and if necessary, a third researcher (NH) was consulted to reach a consensus. The extracted data were subsequently tabulated, summarised and synthesised narratively.

Results

A total of 1534 articles were retrieved from four databases. Then, 689 duplicates were removed. The titles were assessed, resulting in the exclusion of 568 unrelated documents. The remaining 277 documents were reviewed at the abstract level, leading to the assessment of 152 full-text articles. From these, 45 documents were finally analysed. These documents described 29 distinct registries. In parallel, 55 registries were identified from the Orphanet website, 22 of which were eligible for full assessment, with 14 subsequently excluded. We searched documents for the final 8 registries from Orphanet and identified 14 documents for these registries. Finally, 59 documents were analysed for 37 registries (Figure 1). It is worth mentioning that non-English

Table 1. List of included registries.

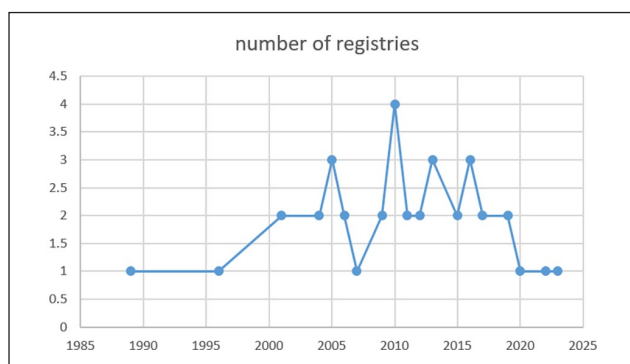
Registry codes	Registry name (references)	Objectives
1	Fabry Disease Registry (Eng et al., 2007; Germain et al., 2013, 2018; Hopkin et al., 2008, 2016; Martins et al., 2013)	Clinical Research-based Epidemiological-based
2	FOS (Beck et al., 2022; Hernberg-Ståhl, 2006)	Clinical Research-based
3	United Kingdom National Registry of Rare Kidney Diseases (radar; Kerecuk et al., 2018; Wong et al., 2023)	Clinical Research-based Epidemiological-based
4	ERKReg (Bassanese et al., 2021)	Clinical Research-based
5	Aregpkd ARPKD registry (Ebner et al., 2015)	Clinical Research-based
6	Primary hyperoxaluria registry (Lieske et al., 2005)	Clinical Research-based
7	Brazilian ahus registry (BraHUS; Nga et al., 2021; Vaisbich et al., 2022)	Clinical Research-based Epidemiological-based
8	Spanish registry of thrombotic thrombocytopenic purpura (REPTT; Mingot-Castellano et al., 2023)	Clinical Research-based
9	The MATUCIP registry in Spain (Barrezueta et al., 2023)	Clinical Research-based
10	The hereditary TTP registry (van Dorland et al., 2019)	Clinical Research-based
11	The UK TTP registry (Scully et al., 2008; Taylor et al., 2024)	Clinical Research-based Epidemiological-based
12	Korean TTP registry (Oh et al., 2012)	Clinical Research-based
13	The French national registry for thrombotic microangiopathy (Joly et al., 2016; Mariotte et al., 2016)	Research-based
14	The global ahus registry (Lazem et al., 2021; Licht et al., 2015; Woodward et al., 2016)	Clinical Research-based
15	Turkish paediatric atypical hemolytic uremic syndrome registry (Besbas et al., 2017; Lazem et al., 2021)	Clinical Research-based
16	The Australian TTP/TMA registry (Blombery et al., 2016; Lazem et al., 2021)	Clinical Research-based Epidemiological-based
17	Milan TTP registry (Lazem et al., 2021; Mancini et al., 2019)	Clinical Research-based
18	Oklahoma TTP-HUS registry (George et al., 2008, 2013; Page et al., 2017; Vesely et al., 2003)	Clinical
19	Italian Registry of hemolytic-uremic syndrome (Italian Registry HUS, 2017; Italian Registry HUS, 2020; Lazem et al., 2021)	Clinical Epidemiological-based
20	International Registry and Biorepository for TMA (International Registry Biorepository TMA, 2013a, 2013b; Lazem et al., 2021)	Clinical Research-based Epidemiological-based
21	German STEC-HUS registry (German EHEC-HUS Registry, 2011; Kielstein et al., 2012)	Clinical Epidemiological-based
22	Iran HUS registry (Lazem et al., 2022)	Clinical Research-based Epidemiological-based
23	TRNA (Metjian et al., 2015, 2016)	Clinical Research-based Epidemiological-based
24	Registry for C3 Glomerulopathy and Immune complex-mediated MPGN (Registry C3Glomerulopathy Immune complex mediated MPGN, 2023)	Clinical Research-based
25	Renaltube (Mejia et al., 2013)	Clinical
26	CCIR (CCIR Registry, 2014, 2023)	Clinical Research-based Epidemiological-based

(Continued)

Table 1. (Continued)

Registry codes	Registry name (references)	Objectives
27	Oxaleurope registry (Martin-Higueras et al., 2021; Metry et al., 2023)	Clinical Research-based
28	Podonet registry (Agnes et al., 2015; Trautmann et al., 2018)	Clinical Research-based
29	Nephronophthisis registry for patients in Germany, Austria and Switzerland (Nephronophthisis Registry, 2024)	Clinical Research-based
30	NEOCYST patient registry for childhood cystic kidney disease (König et al., 2018)	Clinical Research-based
31	ADPKD registry: Spanish registry for autosomal dominant polycystic kidney diseases (Hoover et al., 2022)	Clinical Research-based
32	Polish Registry of Inherited Tubulopathies (PoITube; Zaniew et al., 2014)	Clinical
33	The German ADPKD Tolvaptan Treatment Registry (AD(H)PKD; Arjune et al., 2024)	Clinical
34	French National Registry of C3 Glomerulopathy (C3G; Chabannes et al., 2023)	Clinical Research-based
35	The IFDCR (Limongelli et al., 2024)	Clinical
36	The national registry of clinical personal records for primary Membranoproliferative Glomerulonephritis (Nakagawa et al., 2023)	Clinical
37	Fabry-Stroke Italian Registry (Romani et al., 2022)	Clinical Research-based

FOS: Fabry Outcome Survey; TRNA: TMA Registry of North America; CCIR: Cure Cystinosis International Registry; IFDCR: Italian Fabry Disease Cardiovascular Registry; ERKReg: European Rare Kidney Disease Registry.

**Figure 2.** The number of registries established each year.

documents were translated and analysed; however, no non-English sources were finally included.

Characteristics of the selected registries

Table 1 lists the included registries and their objectives. Figure 2 shows the number of registries established each year and demonstrates fluctuation in the number of registries established per year, over time. Table 2 compares the characteristics of the selected registries established between 1989 and 2023. Among the registries, 15 (40.5%) have international coverage, 20 (54%) are national, and 2 (5.4%) are regional. Remarkably, 35 of these registries were established after 2000 (Figure 2). The United States and Germany had the highest number of registries, each hosting seven. Spain and Italy each had four registries, while the United Kingdom and Spain each had two. Most registries focused on a specific disease, with only two covering all rare kidney diseases (registry codes: 3 and 4; Bassanese et al., 2021;

Kerecuk et al., 2018; Wong et al., 2023). The most frequently represented condition was atypical hemolytic uremic syndrome (HUS), which featured the largest number of registries, totalling eight. Additional details regarding the registries can be found in Supplemental Tables S1–S3).

As shown in Table 2, the steering committee comprised healthcare professionals, patient representatives, and a variety of experts. Most registries did not disclose steering committee members, with only 10 registries including them (registry codes: 1,2,4,6,7,14,15,16,22,37), of which eight featured healthcare professionals on their steering committees. Moreover, five registries included patient representatives as members of the steering committee (registry codes: 1,2,4,14,16). The objectives of the registries varied and can be classified into three primary categories: clinical (all registries, except registry 13), research (29 registries; all registries except registries 18,19,21,25,32,33,35,36), and epidemiological (11 registries including: 1,3,7,11,16,19,20,21,22,23, 26; Table 1). The research objectives of 29 registries focused on evaluating patients, understanding diseases, uncovering unknown factors, and identifying treatments and outcomes.

In most of the compared registries (27 registries), the physicians themselves acted as registrars. In two cases, the registrars were nurses (registry codes: 2,3). Furthermore, two of the reviewed registries permitted patients to input their data (registry codes: 26,31). Additionally, six other registries had registrars from other professional backgrounds (registry codes: 4,6,8,14,17,37). The data resources of 26 registries included health care centres, research centres (registry codes: 3,6,7,16,20,29) and diagnostic centres (Registry codes: 1,2,3,7,13,29).

The inclusion criteria for all registries were the confirmed diagnosis of a rare kidney disease. Three registries also included a relevant family history as part of their

Table 2. Comparison of characteristics of registries.

Reg. codes	Reg. 1	Reg. 2	Reg. 3	Reg. 4	Reg. 5	Reg. 6	Reg. 7	Reg. 8	Reg. 9	Reg. 10
Registries characteristics										
Starting year	2001	2001	2010	2019	2013	2004	2017	2004	2017	2006
Country	US	Sweden	England	European Reference Network	Europe	US	Brazil	Spain	Spain	Austria, Czech Republic, Germany
Disease	Fabry disease	Fabry disease	Rare kidney diseases	Includes 335 rare kidney diseases	ARPKD	Primary hyperoxaluria	aHUS	TTP	TMA	Hereditary TTPOR Upshaw-Schulman Syndrome
Steering committee members	* * * Healthcare professionals Patient representative Others (researchers, coordinators, epidemiologists, web designers, etc.)	* * *		* * *		* *	*			
Geographical coverage										
Purpose										
Inclusion criteria										
Exclusion criteria										
Registrars										
Data sources										

(Continued)

Table 2. (Continued)

Reg. codes	Reg. 11	Reg. 12	Reg. 13	Reg. 14	Reg. 15	Reg. 16	Reg. 17	Reg. 18	Reg. 19	Reg. 20
Registries characteristics										
Starting year	2009	2005	2005	2012	2013	2011	1996	1989	2005	2007
Country	England	South Korea	France	US	Turkey	Australia	Italy	US	Italy	US
Disease	TTP	TTP	TMA	aHUS	aHUS	TTP TMA	US TTP	HUS TTP	HUS	TMA
Steering committee members				*	*	*				
				*						
				*						
Healthcare professionals										
Patient representative										
Others (researchers, coordinators, epidemiologists, web designers, etc.)										
Regional coverage	*							*		
National		*	*		*	*			*	*
International				*			*	*		*
Clinical	*			*	*	*	*	*	*	*
Research-based	*	*	*	*	*	*	*	*	*	*
Epidemiological-based	*			*	*	*	*	*	*	*
Confirmed diagnosis of rare kidney disease	*	*	*	*	*	*	*	*	*	*
Related family history										
Age			*		*		*			*
Informed consent	*			*				*		
Other kidney disease			*	*	*	*			*	
Other (lack of family history, participation in blinded trials, adults, etc.)	*			*	*	*				
Not defined		*		*			*	*		*
Physicians		*	*	*	*	*	*	*		*
Nurses										*
Patient										
Other				*	*	*	*	*	*	*
Healthcare centres	*			*	*	*	*	*	*	*
Diagnostic centres		*	*	*	*	*	*	*	*	*
Research centres			*	*	*	*	*	*	*	*

(Continued)

Table 2. (Continued)

Reg. codes	Reg. 21	Reg. 22	Reg. 23	Reg. 24	Reg. 25	Reg. 26	Reg. 27	Reg. 28	Reg. 29	Reg. 30
Registries characteristics										
Starting year	2011	2021	2013	2010	2010	2010	2016	2009	2019	2016
Country	Germany	Iran	US	Germany	Spain	US	Germany	Germany	Germany	Germany
Disease	HUS	HUS	TMA	Membranous glomerulonephritis and C3G	Primary tubulopathy	Cystinosis	Primary hyperoxaluria	SRNS	Nephronophthisis	ARPKD
Steering committee members										Nephronophthisis Bardet-Biedl syndrome Hepatocyte factor beta-I nuclear nephropathy
Healthcare professionals	*									
Patient representative		*								
Others (researchers, coordinators, epidemiologists, web designers, etc.)										
Geographical coverage										
National		*	*	*	*	*	*	*	*	*
International	*			*	*	*	*	*	*	*
Clinical	*	*	*	*	*	*	*	*	*	*
Research-based	*	*	*	*	*	*	*	*	*	*
Epidemiological-based	*	*	*	*	*	*	*	*	*	*
Confirmed diagnosis of rare kidney disease	*	*	*	*	*	*	*	*	*	*
Inclusion criteria										
Related family history										
Age				*			*	*		*
Informed consent				*			*	*		*
Other kidney disease				*			*	*	*	*
Other (lack of family history, participation in blinded trials, adults, etc.)	*			*			*	*	*	*
Not Defined	*	*	*	*	*	*	*	*	*	*
Registrars										
Physicians	*	*	*	*	*	*	*	*	*	*
Nurses	*	*	*	*	*	*	*	*	*	*
Patient	*	*	*	*	*	*	*	*	*	*
Other	*	*	*	*	*	*	*	*	*	*
Healthcare centres	*	*	*	*	*	*	*	*	*	*
Diagnostic centres	*	*	*	*	*	*	*	*	*	*
Research centres	*	*	*	*	*	*	*	*	*	*

(Continued)

Table 2. (Continued)

Reg. codes	Reg. 31	Reg. 32	Reg. 33	Reg. 34	Reg. 35	Reg. 36	Reg. 37
Registries characteristics							
Starting year	2016	2012	2015	2006	2023	2015	2020
Country	Spain	Poland	Germany	France	Italy	Japan	Italy
Disease	ADPKD	Hereditary tubulopathies	ADPKD	DDD C3G	Fabry disease	Primary membranoproliferative glomerulonephritis	Stroke associated with Fabry disease
Steering committee members							*
Geographical coverage							
							*
Purpose							
Inclusion criteria							
Exclusion criteria							
Registrars							
Data sources							

aHUS: atypical hemolytic uremic syndrome. * Means "Yes".

criteria (registry codes: 5,10,33). Furthermore, age was a consideration for several registries (registry codes: 5,8,9,13,15,17,20,24,27,28,30,32,33,34,35,37). The inclusion criteria based on age varied depending on the type of rare kidney disease covered by each registry. Some registries focused on only paediatric patients (registry codes: 9,20,27,28,30,32), while others included only adults (registry codes: 8,24,34,37). The requirement for informed consent was an inclusion criterion in 11 registries (registry codes: 1,2,11,14,18,24,27,31,32,35,36). In all of these registries, an opt-in approach was applied. For paediatric registries, parental or guardian consent was obtained (registry codes: 11,14,27). The exclusion criterion across the registries was the presence of other kidney diseases (registry codes: 5,7,13,14,15,16,28,29,30,34,35,36). Conversely, 14 registries did not define any exclusion criteria (registry codes: 1,4,8,9,10,12,17,18,20,22,26,27,31,32).

Table 3 compares the datasets, data collection methods and quality assessment strategies across the registries. The datasets across all the registries are classified into eight categories: demographic data, clinical history, paraclinical data, therapeutic data (including interventions and medications), genetic testing, comorbidities, family history and outcomes. Almost all registries incorporated four essential data categories: demographic data, clinical history, paraclinical data and therapeutic data. Seventeen registries collected genetic testing data (registry codes: 1,3,4,5,6,10,14,15,24,27,28,29,30,32,33,34,35). Six registries recorded information on comorbidities (registry codes: 3,7,18,19,24,33) and 12 registries gathered data regarding family history (registry codes: 2,4,5,6,7,9,14,22,29,33,35,37). Six registries documented the stages of the disease (registry codes: 4,6,11,13,27,34), and biological samples were collected and stored in nine registries (registry codes: 2,5,17,20,23,24,28,30,34). Furthermore, eight registries employed patient-reported outcome measures (registry codes: 2,4,7,8,14,18,19,28).

Data quality assessment strategies were absent for 17 registries. In contrast, the remaining registries employed various methods to ensure quality control, including the involvement of physicians in data quality monitoring (registry codes: 1,2,5,6,7,14,15,22), adherence to established data quality guidelines (registry codes: 1,2,3,4,5,7,8,13,14,16,22,29,30,32), utilisation of data quality indicators (registry codes: 2,3,4,5,7,9,22,23,29,30), conducting data quality assessments and audits (registry codes: 2,3,4,7,9,16,19,22,23,29,30,31), providing periodic training sessions (registry codes: 3,16) and implementing automated checks on data entry (registry codes: 4,5,6,30).

A total of 10 registries utilised retrospective data collection methods (registry codes: 6,7,8,11,16,21,27,29,33,36). In contrast, nine registries exclusively employed prospective data collection methods (registry codes: 2,12,13,14,20,23,26,30,37). Furthermore, 17 registries used a combination of both retrospective and prospective approaches for data collection (registry codes: 1,3,4,5,9,10,15,17,18,19,22,24,28,31,32,34,35). The data collection method for registry number 25 was not reported. Table 4 summarises the frequency of each characteristic for the included registries.

Discussion

General characteristics of rare kidney disease registries

Steering committee members of the included registries, generally, comprised medical care specialists (primarily nephrologists) along with patient representatives. In certain registries, additional members such as web designers, researchers, information experts and other professionals were also included. Depending on the size and purpose of the registry, it is advisable to establish a steering committee comprising various internal and external experts. This body is responsible for overseeing daily operations and ensuring adherence to relevant laws and regulations, either directly or through dedicated working groups. Furthermore, independent advisory boards can offer essential technical expertise and uphold scientific neutrality (Tarride et al., 2024). Incorporating patient representatives into governing boards or committees promotes a patient-centred approach and augments their involvement in decision-making processes (Kodra et al., 2018). These findings suggest that future registries should adopt multidisciplinary governance structures, ensuring the inclusion of both clinical and non-clinical experts, as well as patient representatives, to enhance performance and transparency.

Regarding registry purposes, it was found that meeting the triad of clinical, research and epidemiological goals is essential for included registries. The prevalence of rare kidney diseases was also a key consideration. Clinical registries are generally designed for various purposes, including managing patient care, enhancing the understanding of the natural history of diseases, establishing suitable platforms for clinical trials and addressing specific research needs (Gliklich et al., 2014). Lazem et al. (2021) suggested that due to the limited understanding of the management of rare diseases and the scarcity of patient samples, it is essential to implement rare kidney disease registries focusing on clinical, research and epidemiological aspects. Kodra et al. (2018) noted that each registry should have a primary overarching objective alongside specific secondary goals. These objectives must be defined clearly, precisely and feasibly. Well-articulated goals are vital for determining what data to collect, as well as criteria for patient inclusion, necessary ethical and legal procedures, design of the study and analytical and reporting methods. Such clarity ensures that a registry effectively addresses significant scientific questions and collects data aligning with its specific objectives. Unclear objectives can lead to inflated costs without providing added value, result in excessive data collection compromise quality and completeness, and ultimately hinder the ability to effectively address critical questions (Kodra et al., 2018). Therefore, future rare kidney registries are recommended to clearly define primary and secondary objectives that integrate clinical, research and epidemiological goals.

Among the included registries, a physician's diagnosis is considered adequate for inclusion. This approach is necessary due to the limited understanding of rare diseases and the small size of patients, which makes broader inclusivity

Table 3. Comparison of data collection in registries of rare kidney diseases.

Reg. codes	Reg. 1	Reg. 2	Reg. 3	Reg. 4	Reg. 5	Reg. 6	Reg. 7	Reg. 8	Reg. 9	Reg. 10
Registries characteristics										
Data set	Demographic	*	*	*	*	*	*	*	*	*
	Clinical history	*	*	*	*	*	*	*	*	*
	Paraclinical measures	*	*	*	*	*	*	*	*	*
	Treatment	*	*	*	*	*	*	*	*	*
	Genetic test	*	*	*	*	*	*	*	*	*
	Comorbidities									
	Family history		*	*	*	*	*	*	*	*
	Outcomes	*	*	*	*	*	*	*	*	*
	Disease stage									
	Other (quality of life, parental kinship, etc.)		*	*	*	*	*	*	*	*
Biological samples	Physician's participation in data quality control		*	*	*	*	*	*	*	*
	Patient-reported outcomes	*	*	*	*	*	*	*	*	*
	Data quality assessment strategy									
	Following data quality guidelines	*	*	*	*	*	*	*	*	*
	Using of data quality indicators	*	*	*	*	*	*	*	*	*
	Evaluating and auditing data quality	*	*	*	*	*	*	*	*	*
	Training courses on data quality		*	*	*	*	*	*	*	*
	Real-time evaluation of data at the time of its entry		*	*	*	*	*	*	*	*
	Retrospective	*	*	*	*	*	*	*	*	*
	Prospective	*	*	*	*	*	*	*	*	*
Registries characteristics										
Data set	Demographic	*	*	*	*	*	*	*	*	*
	Clinical history	*	*	*	*	*	*	*	*	*
	Paraclinical measures	*	*	*	*	*	*	*	*	*
	Treatment	*	*	*	*	*	*	*	*	*
	Genetic test									
	Comorbidities									
	Family history		*	*	*	*	*	*	*	*
	Outcomes	*	*	*	*	*	*	*	*	*
	Disease stage	*	*	*	*	*	*	*	*	*
	Other (quality of life, parental kinship, etc.)									
Biological samples	Physician's participation in data quality control		*	*	*	*	*	*	*	*
	Patient-reported outcomes		*	*	*	*	*	*	*	*
	Data quality assessment strategy									
	Following data quality guidelines		*	*	*	*	*	*	*	*
	Using of data quality indicators		*	*	*	*	*	*	*	*
	Evaluating and auditing data quality		*	*	*	*	*	*	*	*
	Training courses on data quality		*	*	*	*	*	*	*	*
	Real-time evaluation of data at the time of its entry	*	*	*	*	*	*	*	*	*
	Retrospective		*	*	*	*	*	*	*	*
	Prospective		*	*	*	*	*	*	*	*

(Continued)

Table 3. (Continued)

Reg. codes	Reg. 21	Reg. 22	Reg. 23	Reg. 24	Reg. 25	Reg. 26	Reg. 27	Reg. 28	Reg. 29	Reg. 30
Registries characteristics										
Data set										
Demographic	*	*	*	*	*	*	*	*	*	*
Clinical history	*	*	*	*	*	*	*	*	*	*
Paraclinical measures	*	*	*	*	*	*	*	*	*	*
Treatment	*	*	*	*	*	*	*	*	*	*
Genetic test										
Comorbidities										
Family history		*	*	*	*	*	*	*	*	*
Outcomes	*	*	*	*	*	*	*	*	*	*
Disease stage										
Other (quality of life, parental kinship, etc.)			*	*	*	*	*	*	*	*
Biological samples										
Patient-reported outcomes										
Data quality assessment strategy		*								
Physician's participation in data quality control										
Following data quality guidelines		*	*	*	*	*	*	*	*	*
Using of data quality indicators		*	*	*	*	*	*	*	*	*
Evaluating and auditing data quality		*	*	*	*	*	*	*	*	*
Training courses on data quality										
Real-time evaluation of data at the time of its entry										
Data collection	*	*	*	*	*	*	*	*	*	*
Retrospective		*	*	*	*	*	*	*	*	*
Prospective		*	*	*	*	*	*	*	*	*
Registries characteristics										
Reg. codes	Reg. 31	Reg. 32	Reg. 33	Reg. 34	Reg. 35	Reg. 36	Reg. 37			
Data set										
Demographic	*	*	*	*	*	*	*	*	*	*
Clinical history	*	*	*	*	*	*	*	*	*	*
Paraclinical measures	*	*	*	*	*	*	*	*	*	*
Treatment	*	*	*	*	*	*	*	*	*	*
Genetic test		*	*	*	*	*	*	*	*	*
Comorbidities		*	*	*	*	*	*	*	*	*
Family history		*	*	*	*	*	*	*	*	*
Outcomes	*	*	*	*	*	*	*	*	*	*
Disease stage										
Other (quality of life, parental kinship, etc.)	*	*	*	*	*	*	*	*	*	*
Biological samples										
Patient-reported outcomes										
Data quality assessment strategy		*	*	*	*	*	*	*	*	*
Physician's participation in data quality control										
Following data quality guidelines		*	*	*	*	*	*	*	*	*
Using of data quality indicators		*	*	*	*	*	*	*	*	*
Evaluating and auditing data quality		*	*	*	*	*	*	*	*	*
Training courses on data quality										
Real-time evaluation of data at the time of its entry		*	*	*	*	*	*	*	*	*
Data collection	*	*	*	*	*	*	*	*	*	*
Retrospective	*	*	*	*	*	*	*	*	*	*
Prospective	*	*	*	*	*	*	*	*	*	*

* Means "Yes".

Table 4. Distribution of characteristics in the reviewed registries.

Registries characteristics	Frequency	Registry codes
National	20	3,7,8,9,12,13,15,16,19,22,23,24,25,31,32,33,34,35,36,37
International	15	1,2,4,5,6,10,14,17,20,21,26,27,28,29,30
Regional	2	11,18
Purpose		
Clinical	36	1,2,3,4,5,6,7,8,9,10,11,12,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37
Research-based	29	1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,20,22,23,24,26,27,28,29,30,31,34,37
Epidemiological-based	11	1,3,7,11,16,19,20,21,22,23,26
Steering committee members		
Healthcare professionals	8	1,4,6,7,15,16,22,37
Patient representative	5	1,2,4,14,16
Others (researchers, coordinators, epidemiologists, web designers, etc.)	8	1,2,4,6,14,16,22,37
Data set		
Demographic	37	1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37
Clinical history	36	1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,37
Paraclinical measures	36	1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,37
Treatment	33	1,2,3,5,6,8,9,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,30,31,32,33,34,35,36,37
Outcomes	25	1,8,9,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,27,28,30,31,32,34,37
Genetic test	17	1,3,4,5,6,10,14,15,24,27,28,29,30,32,33,34,35
Family history	12	2,4,5,6,7,9,14,22,29,33,35,37
Comorbidities	6	3,7,18,19,24,33
Disease stage	6	4,6,11,13,27,34
Other (quality of life, parental kinship, etc.)	10	2,3,4,7,8,15,19,24,27,31
Biological samples	9	2,5,17,20,23,24,28,30,34
Patient-reported outcomes	8	2,4,7,8,14,18,19,28
Data quality assessment strategy		
Following data quality guidelines	14	1,2,3,4,5,7,8,13,14,16,22,29,30,32
Evaluating and auditing data quality	12	2,3,4,7,9,16,19,22,23,29,30,31
Using data quality indicators	10	2,3,4,5,7,9,22,23,29,30
Physician's participation in data quality control	8	1,2,5,6,7,14,15,22
Real-time evaluation of data at the time of its entry	4	4,5,6,30
Training courses on data quality	2	3,16
Inclusion criteria		
Confirmed diagnosis of rare kidney disease	37	1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37
Age	16	5,8,9,13,15,17,20,24,27,28,30,32,33,34,35,37
Informed consent	11	1,2,11,14,18,24,27,31,32,35,36
Related family history	3	5,10,33
Exclusion criteria		
Other kidney disease	13	5,7,13,14,15,16,28,29,30,34,35,36,37
Other (lack of family history, participation in blinded trials, adults, etc.)	14	2,6,7,11,14,15,16,21,24,33,34,35,36,37
Not defined	14	1,4,8,9,10,12,17,18,20,22,26,27,31,32
Registrars		
Physicians	27	1,2,5,6,7,8,10,12,13,14,15,16,17,20,21,22,23,24,25,27,28,29,32,33,34,35,37
Nurses	2	2,3
Patient	2	26,31
Other	6	4,6,8,14,17,37

(Continued)

Table 4. (Continued)

Registries characteristics	Frequency	Registry codes
Data collection		
Retrospective	10	6,7,8,11,16,21,27,29,33,36
Prospective	9	2,12,13,14,20,23,26,30,37
Both	17	1,3,4,5,9,10,15,17,18,19,22,24,28,31,32,34,35
Data sources		
Healthcare centres	26	3,4,5,7,8,9,10,11,12,14,15,16,17,18,19,20,21,22,23,25,28,29,31,32,34,35
Diagnostic centres	6	1,2,3,7,13,29
Research centres	6	3,6,7,16,20,29

(Gliklich et al., 2014). Other common inclusion criteria typically include age, geographic location and informed consent (Kölker et al., 2022). In our review, age and informed consent were also among the inclusion criteria. Consistent with our findings, Lazem et al. (2021) underscored the importance of age and the confirmed diagnosis of HUS as key inclusion factors.

Exclusion criteria play a vital role as they help to eliminate individuals whose data may distort the findings or whose participation may not be in line with the objectives of a registry. Common exclusion criteria were the presence of comorbidities, misdiagnoses (such as patients with conditions not classified as rare diseases or those with rare forms of common diseases), and prior participation in studies (Hageman et al., 2023; Tarride et al., 2024). These criteria often vary based on the specific type of rare kidney disease under study. For example, in certain conditions, the absence of a family history resulted in exclusion, while studies focusing absolutely on paediatric populations excluded adults. For future registries, it is crucial to define accurate inclusion and exclusion criteria to ensure the integrity and reliability of the data collected. These criteria play a vital role in preventing bias and ensuring that only relevant and valid patient data are included in a registry. In the case of rare diseases, where patient populations are often limited, precise patient selection is essential for obtaining accurate results. Ill-defined inclusion criteria can lead to the inclusion of patients who do not exhibit the disease characteristics, distorting the findings. Similarly, well-defined exclusion criteria help eliminates data from patients who may have comorbidities or misdiagnoses, which could otherwise impact the validity of the study.

In the majority of included registries (27 cases), physicians were primarily responsible for patient enrollment. Various methods can be utilised to enrol patients. Patients may be reported by the diagnosing specialist or the treating physician, or they may choose to self-enrol through dedicated websites or patient support groups. Moreover, a combination of these methods can be employed (Pisa et al., 2023). Similarly, Lazem et al (2021) found that most registrars were nephrology specialists. Given that physicians often have demanding schedules, their obligation to enter data can pose significant challenges (Grant and Goddard, 2012). To address this concern, it is suggested that improving interoperability between registry software and other information systems, such as electronic health records (EHRs), could facilitate the automated transfer of various data into the registry (Lazem et al., 2021).

Data from rare kidney disease registries

Demographic data, clinical history, paraclinical data, treatment information, genetic testing, comorbidities, family history, outcomes and additional data are commonly collected by rare kidney disease registries. In their systematic review, Bernardi et al. (2023) established a minimum dataset crucial for RDRs, which includes the following components: eligibility (age at enrollment, date of encounter, etc.), identification (age, gender, etc.), diagnosis (clinical diagnosis, date, methods used to assess the diagnosis, etc.), treatment (start date, medication, etc.), medical consultation (mother’s age at delivery), comorbidity (diagnosis codes), outcome (cause of death, date of death, etc.), other (patients with ongoing follow up at this centre, etc.). These data elements provide a fundamental framework for the development of comprehensive and standardised RDRs. In the study conducted by Lazem et al. (2021), a dataset was proposed for HUS registries, which align with the minimum dataset identified in our study.

Increasingly, genetic testing is becoming a pivotal component of diagnostic tools with various applications such as confirming clinical diagnoses, establishing inheritance patterns, informing family planning decisions, identifying the causes of unexplained familial kidney disorders, evaluating the eligibility of family members for kidney donation, etc. (Aymé et al., 2017). Given these factors, genetic testing can play a major role in the evaluation of children and young adults, especially when clinical phenotypes do not yield a definitive diagnosis. Therefore, rare kidney disease registries may decide to include genetic data. In addition, Metjian et al. (2015) emphasised that data related to biological samples should also be documented to facilitate future research beyond the existing datasets. These data are essential for addressing research challenges and the scarcity of biological samples, particularly in identifying the relationship between genetic factors (through the storage of DNA and RNA samples) and the occurrence of rare kidney diseases (Kölker et al., 2022). According to Garcia et al. (2018), while independent registries are appropriate for understanding the natural history of diseases, developing best practices, and improving patient outcomes, they are limited in supporting fundamental research. Given the critical role of basic research in rare disease studies, scientists must first understand the disease pathways before developing suitable interventions. Conversely, rare disease biobanks provide the necessary infrastructure for basic research, enabling them to conduct new omics discoveries, identify and validate biomarkers, uncover novel genes and

develop new therapeutic strategies. Garcia et al. (2018) concluded that registries with biobanks offer a unique, practical, cost-effective and efficient approach to rare disease research.

Patient-reported outcomes (PROs) are also increasingly recognised as valid measures, including quality of life (QoL), pain, shortness of breath and physical function. Systematic collection of PROs is feasible and effective in clinical trials, patient registries and routine clinical care settings (Kölker et al., 2022). In their survey on RDRs in Australia, Ruseckaite et al. (2023) noted that PROs and QoL data were collected by only one subset of identified RDRs. Including PROs in clinical registries provides several advantages (Ahern et al., 2017). First, integrating patient's perspectives ensures that care outcome measurements remain patient-centred. Furthermore, symptom burden, QoL, and care satisfaction are dynamic variables that, if not captured 'at the moment', cannot be accurately reconstructed retrospectively and are effectively lost (Ruseckaite et al., 2023).

In nearly half of the registries reviewed, no data quality assurance methods were reported. Similarly, Hageman et al. (2023) noted that while RDRs typically describe registry design elements, they often place less emphasis on data quality. Likewise, in the study of Ruseckaite et al. (2023), quality improvement methods were not available for more than half of the identified RDRs. In general, unlike clinical trial research data, the quality of registry data is rarely evaluated at the source. Nevertheless, these data are frequently used for epidemiological studies, clinical research, quality improvement tracking and national or international comparisons. Pellen et al. (2018) proposed strategies for improving the quality of registry data, including training staff on measurement processes, organising data systematically, appropriately utilising patient information systems and defining and strictly applying clear rules for data transfer into the registry. Similarly, Lazem et al. (2021) recommended that managers of HUS registries adopt a combination of methods to ensure data quality. Therefore, it is strongly recommended that future rare kidney disease registries integrate clearly defined and systematically implemented data quality assurance procedures into their design. Ensuring data quality at every stage, from data entry to reporting, is essential to support the reliability of findings and to maximise the value of registry data.

Registries utilise various data collection methods, which can be broadly categorised into retrospective and prospective approaches. Retrospective designs are commonly employed in registries. Such studies offer advantages in terms of speed and cost-effectiveness, requiring fewer resources and less time compared to prospective studies. However, retrospective studies are more susceptible to biases, especially due to issues related to data collection, analysis and the impact of unrecognised confounding factors. In contrast, prospective studies present a more robust model for data collection because patients are selected based on predefined characteristics, with their outcomes subsequently recorded throughout the study. Prospective studies tend to be associated with lower levels of reporting and ana-

recommended that HUS registries should adopt both retrospective and prospective approaches, taking into account the feasibility of data collection, prioritisation of key variables and the limitations of available information resources. This is because retrospective data collection may result in the omission of critical information as patient records before the initiation of the registry may not be available. Conversely, a solely prospective approach would preclude the inclusion of patients already diagnosed with the condition, which could lead to the exclusion of numerous cases given the rarity of the disease.

The broad objective of registries is to collect data from a variety of potential sources (Gliklich et al., 2014). Rare kidney disease registries mainly collect the data from healthcare centres (hospitals) and diagnostic centres such as private clinics and academic research centres. Similarly, Lazem et al. (2021) reported that the majority of data sources were nephrology hospitals for HUS registries. They also noted that collaboration with a range of healthcare centres, diagnostic research facilities and nephrology units is also essential. On the other hand, registries may require data integration from various sources. Therefore, they recommended the use of EHRs as this process facilitates the compilation of case findings and the completion of patient records or follow-ups from diverse data sources. Accordingly, it is recommended that future rare kidney disease registries prioritise establishing formal collaborations with a wide range of data sources, including nephrology hospitals, diagnostic centres and academic research institutions. Moreover, the integration of EHRs should be considered a key strategy to facilitate data linkage, enhance case completeness and enable effective follow-up through access to longitudinal patient information across various healthcare settings.

Limitations

A limitation of this study is the incomplete data available for certain registries. Despite attempts to obtain additional information through correspondence with the authors, sufficient details were not acquired for some of the included registries. Another limitation is the potential omission of some rare kidney disease registries that may not have been captured in the search process. This may have occurred because some registries were not listed in scientific databases or Orphanet. These limitations should be taken into account when interpreting the results of this study.

Conclusion

This study emphasises the importance of establishing well-structured and clearly defined registries for rare kidney diseases. While national or international registries could offer the advantage of larger populations and more comprehensive data, the establishment of new registries remains essential for several reasons. Legal and ethical challenges regarding data sharing across different countries and institutions, as well as the varying healthcare systems, may hinder the creation of centralised databases (Roundtable on Environmental Health Sciences Research, and Medicine et al., 2016). Furthermore, differences in the prevalence and patterns of

rare kidney diseases across various regions often necessitate specific, localised data for more effective disease management and tailored treatment strategies. As a result, countries and institutions may be compelled to create their own registries. However, even when these registries are separate, adherence to standardised, transparent criteria – such as common data elements, well-defined inclusion and exclusion criteria, and consistent governance structures – can ensure that the data collected remain comparable across different registries. Such standardisation allows for the integration of data from multiple registries, enabling comprehensive analysis and enhancing the overall understanding of rare kidney diseases, even when the registries operate independently. In conclusion, the successful implementation of rare kidney disease registries depends on creating standardised frameworks that promote data consistency and interoperability.

Author contributions

ZM: Study conception and design, Data collection, Data analysis, Writing the draft, Revising the manuscript.

NH: Study conception and design, Data collection, Revising the manuscript.

AS: Study conception and design, Data collection, Data preparation, Data analysis, Writing the draft, Revising the manuscript, and Supervision.

All authors provided critical feedback and helped shape the research, analysis, and manuscript.

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Data availability statement

All data that support the findings are included within the manuscript, and Supplementary Material.

Supplemental material

Supplemental material for this article is available online.

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