Capturing real-world rare disease patient journeys: Are current methodologies sufficient?

MSR19

Kristen A. Cribbs, Lucas T. A. Blackmore, Asia R. Banks, Da Sol Kim, Betsy J. Lahue

Alkemi LLC, Manchester Center, VT, USA

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Background

- Decision-makers increasingly prioritize patient perspectives and real-world outcomes to inform access and care guidelines[1,2]
- Data on patient disease journey experiences can be challenging to collect and evaluate for rare conditions[3,4]
- This study assessed the body of evidence for rare disease patient journeys

Methods

- A systematic literature review (SLR) was conducted to explore study designs, methods, and outcome trends in rare disease patient journeys
- PubMed and Google Scholar were queried using relevant keywords (journey, path, odyssey), and publications were selected based on pre-defined search parameters **(Table 1)**
- Data on disease state, study design, location, data collection methods, journey stage **(Figure 1)**, and reported outcomes was abstracted; Descriptive analyses were conducted
- GRADE quality assessments were performed

Table 1. PICOS Criteria

Parameter	Inclusion Criteria	Exclusion Criteria
Population	Patients diagnosed with a rare disease Caregivers of these patients Healthcare providers of these patients	Other populations
Outcomes	Patient journey-related outcomes (e.g., symptoms, QOL, HRU, costs, caregiver impact, treatment experience)	Non-patient journey outcomes
Study Design	Retrospective observational Prospective observational Open label Studies must focus on the patient journey and include at least one the following terms: • Journey • Path • Odyssey	Animal studies Case report Case series Narrative reports SLRs/Meta-analyses/Other reviews Randomized clinical trials
Publication Type	Peer-reviewed publications Congress proceeding (abstract, poster)	Opinion pieces Commentaries Editorials Grey literature
Date Range	January 1, 2014-April 30, 2024	Before January 1, 2014
Language	English	Non-English

Abbreviations: QOL, quality of life; HRU, healthcare resource utilization; SLR, systematic literature review

Figure 1. Patient Journey Schematic

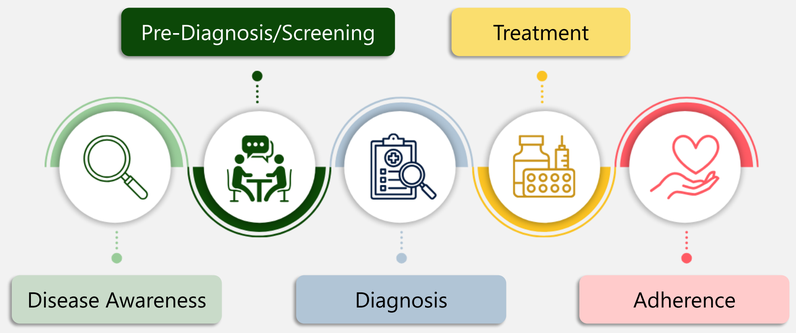
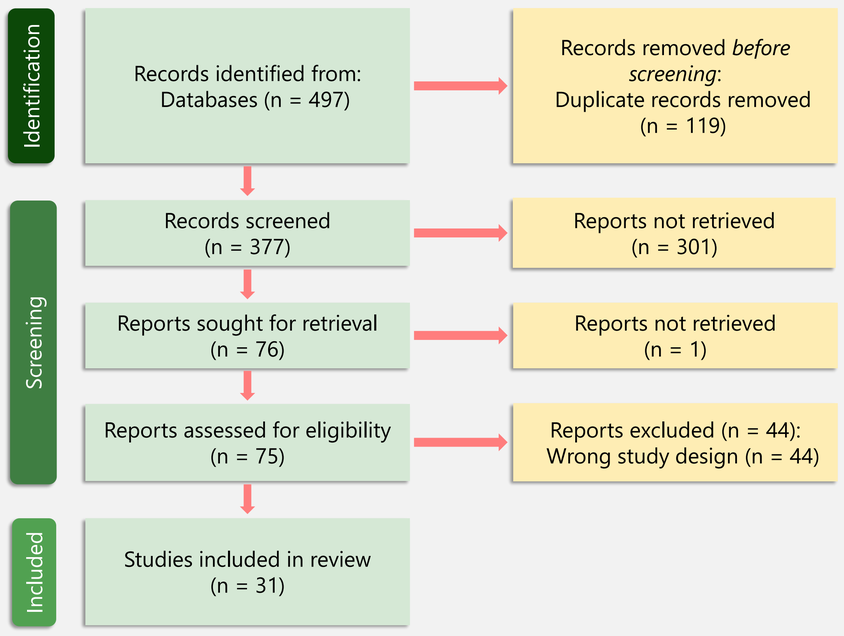


Figure 2. SLR PRISMA Flow Diagram



Results

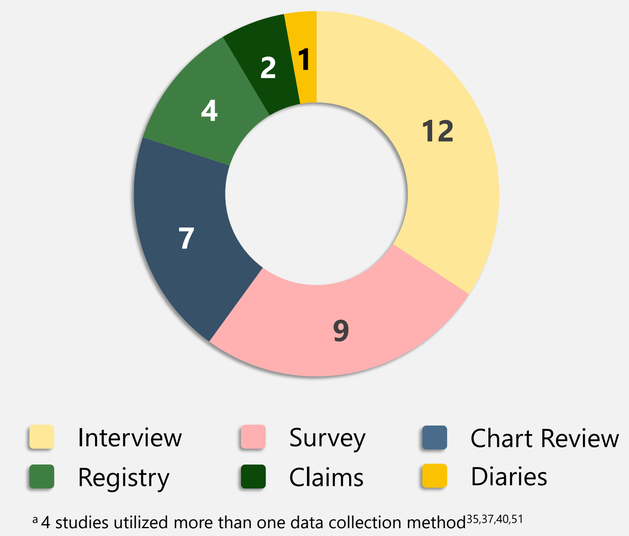
- 31 publications[5-35] **(Figure 2)** included 296,548 participants across 600+ rare diseases **(Table 2)**
- Most studies were prospective observational (61%)[6-8,10-12,15-18,22-25,27,30,31,33,35] and queried patients (87%)[5,6,9-21,23-26,28-35] **(Table 2)**
- Common methodologies included interviews (39%)[7-8,13,15-18,21,22,27,33,35] and surveys (29%)[6,10,12,20,23-25,30,31] **(Figure 3)**
- 'Pre-diagnosis/Screening' (97%)[5-27,29-35] was the most frequently studied journey stage, 'Adherence' (6%)[28,31] was least common **(Figure 4)**

Table 2. Study & Patient Characteristics

Variable	n (%)
Total Studies	31 (100)
Minimum Sample Size, n patients	*8*
Maximum Sample Size, n patients	*292,617*
Study Population[a]	
Patients[b]	27 (87)
Pediatric	*11*
Adult	*21*
Age not specified	*5*
Caregivers/Family	10 (32)
Healthcare Provider	3 (10)
Publication Type	
Original Research	30 (97)
Conference Proceeding	1 (3)
Study Location[c]	
Europe	14 (45)
North America	12 (39)
Asia & Pacific	9 (29)
South America	2 (6)
Study Design	
Observational	30 (97)
Prospective	*19 (61)*
Retrospective	*11 (35)*
Open Label	1 (3)
Disease Organ Systems	
Blood & Circulatory	1 (3)
Brain & Nervous System	3 (10)
Musculoskeletal	3 (10)
Integumentary	2 (6)
Endocrine	1 (3)
Immune	5 (16)
Multi-System	10 (32)
Not Specified	6 (19)
Diseases Assessed[d]	646

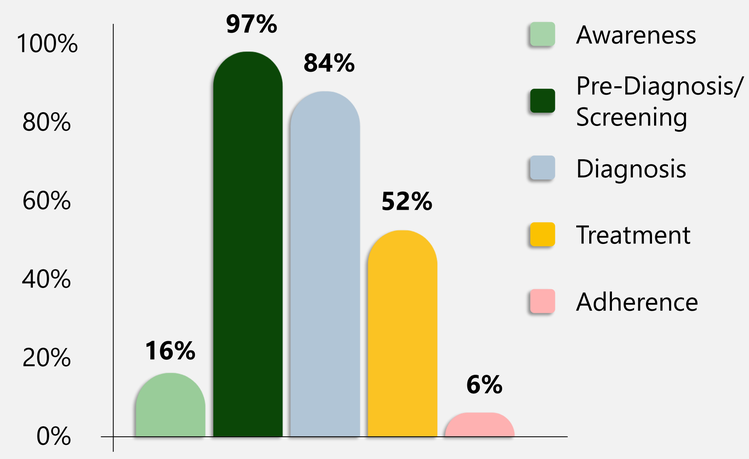
[a] 8 studies reported multiple study populations[12-15,20,24,33,35]
[b] 10 studies that included patients spanned multiple age demographics[6,12,15,20,23-25,29,30,33]
[c] 3 studies were multi-regional[11,21,24]
[d] 4 studies did not specify number of diseases assessed[7,8,26,33]

Figure 3. Frequency of Data Collection Methods Employed (n=31)[a]



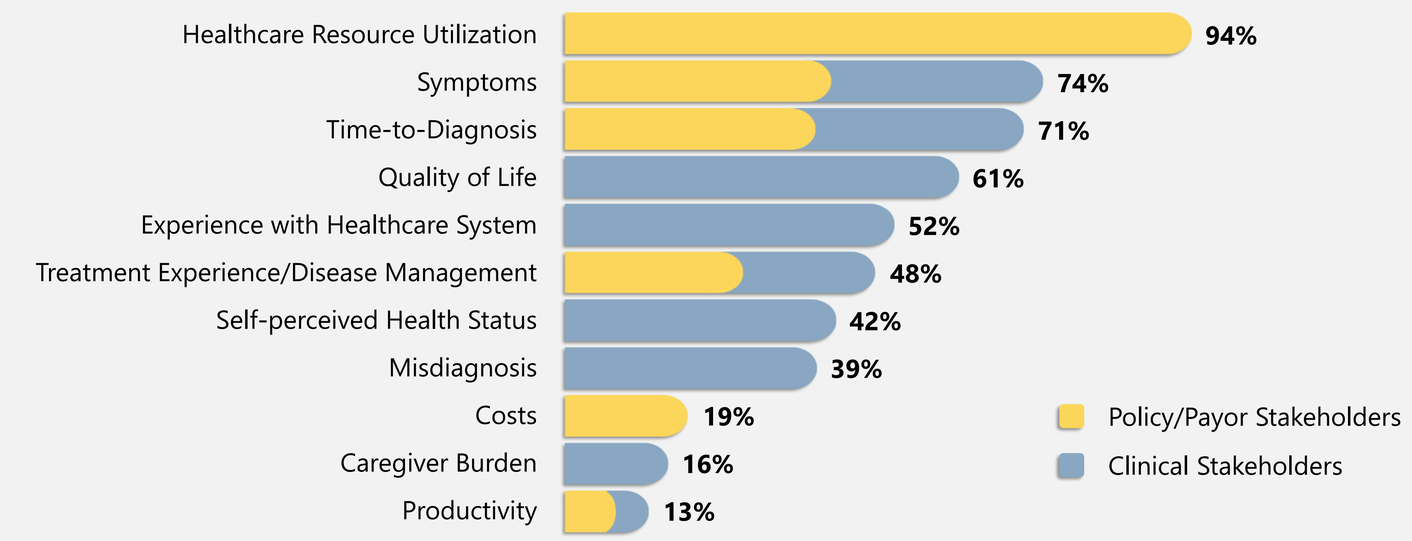
[a] 4 studies utilized more than one data collection method[35,37,40,51]

Figure 4. Proportion of Publications Investigating Patient Journey Stages (n=31)



- 164 outcomes were reported overall; 'Healthcare Resource Utilization' was most frequently reported (94%),[5-20,22-32,34,35] and 'Productivity' was least reported (13%)[9,11,31,33] **(Figure 5)**
- Due to observational designs, most studies (74%) were deemed 'low' quality[5,6,9-15,17-21,23-26,29,31,33-35]

Figure 5. Proportion of Publications Reporting Observed Outcomes and their Relative Importance to Healthcare Decision-Makers (n=31)



Conclusions

- Most SLR publications focused on pre-diagnosis and diagnosis journey stages and gathered data through interviews and surveys
- Researchers commonly reported resource utilization, patient symptoms, and time-to-diagnosis, while evidence gaps included treatment adherence, productivity, and caregiver burden
- Longitudinal assessments of real-world care and treatment experiences in rare disease, including caregiver perspectives, can enhance clinician and policymaker decision-making

References: **1.** FDA Real-World Evidence Program. 2018. **2.** NICE real-world evidence framework. 2022. **3.** Mesa et al. Cancer 2022. **4.** Zanello et al. Orphanet 2022. **5.** Aoust et al. Orphanet 2017. **6.** Baldell et al. Orphanet 2019. **7.** Baumbusch et al. J Genet Couns 2018. **8.** Bauskis et al. Orphanet 2022. **9.** Benito-Lozano et al. PLoS One 2023. **10.** Benson et al. Orphanet 2022. **11.** Bernthal et al. Orphanet 2021. **12.** Delgado-Garcia et al. Front Neurol 2022. **13.** Galvin et al. BMC Health Serv 2015. **14.** Grier et al. Neurol Genet 2018. **15.** Hausmann et al. Orphanet 2018. **16.** Hoffman et al. J Endocr Soc 2022. **17.** Isono et al. PLoS One 2022. **18.** Kinoshita et al. Value Health Reg Issues 2021. **19.** Kobayashi Takahashi et al. Brain Dev 2024. **20.** Lagler et al. JIMD Rep 2019. **21.** Lambert et al. Patient 2020. **22.** Luz et al. Acta Paulista 2015. **23.** Magerl et al. Orphanet 2020. **24.** Mehta et al. Mol Genet Metab 2017. **25.** Mengel et al. PLoS One 2020. **26.** Michaels-Igbokwe et al. Genet Med 2021. **27.** Somanadhan et al. Orphanet 2016. **28.** Tada et al. J Dermatol 2024. **29.** Tisdale et al. Orphanet 2021. **30.** Tsurumi et al. Genet Metab Rep 2022. **31.** Vargas Camaño et al. Rev Alerg Mex 2023. **32.** Vera-Llonch et al. Orphanet 2021. **33.** Witt et al. Orphanet 2023. **34.** Yan et al. J Environ Res Public Health 2020. **35.** Muir et al. Neuromuscular Disorders 2022.

Presented at ISPOR EU, November 2024, Barcelona, Spain
Please contact kristen.cribbs@alkemihealth.com for more information.

www.alkemihealth.com