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An Investigation of Patient Journey Assessment in Rare Disease

Kristen A. Cribbs, PhD, MPH, Da Sol Kim, MS, Betsy J. Lahue, MPH
Alkemi LLC, Manchester Center, VT, USA

Background

- Understanding the patient journey, or spectrum of disease-related events experienced by a patient from symptom onset through diagnosis and treatment, is critical to informing patient-centered care
- People living with rare disease face distinct challenges that arise from the infrequency of their medical conditions such as long diagnostic journeys, inadequate clinical management, and limited access to effective treatments
- The goal of this study was to investigate how patient journeys are characterized in rare disease

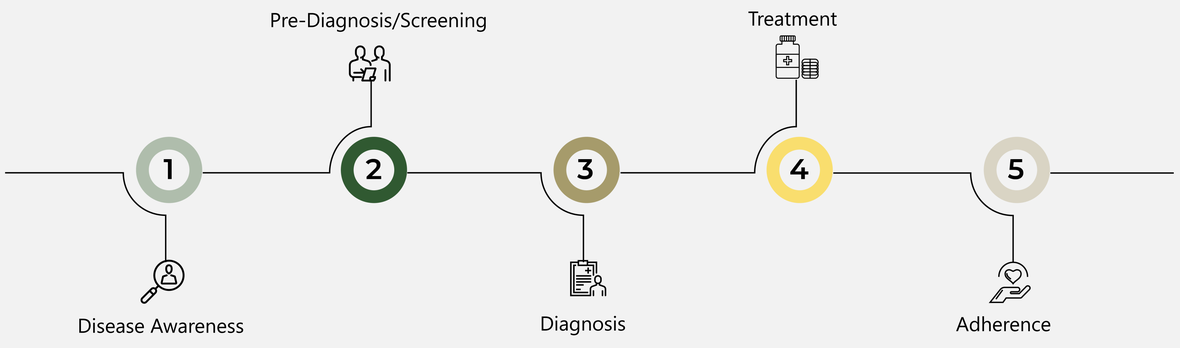
Methods

- We conducted a targeted literature review to explore rare disease patient journey study designs, methods, and outcome trends
- We queried PubMed and Google Scholar using relevant keywords (patient journey, rare disease, patient experience) and selected articles based on pre-defined search parameters **(Table 1)**
- We abstracted data on disease state, study design, location, data collection methods, journey stage **(Figure 1)**, and reported outcomes from identified articles; Descriptive analyses were conducted

Table 1. Literature Review Parameters

Parameter	Inclusion	Exclusion
Population	Patients diagnosed with a rare disease	Other patient populations
Outcomes	Patient journey-related aspects (e.g., screening, diagnosis, disease awareness, treatment, adherence, and management)	Non-patient journey outcomes
Study Design	Randomized clinical trials Retrospective observational studies Prospective observational studies Systematic Literature Reviews/Meta-Analyses/Other Reviews	Animal studies
Publication Type	Peer-reviewed publication Congress proceeding (abstract, poster)	Opinion pieces Editorials Grey literature
Date Range	January 1, 2018 – October 31, 2023 (newer articles prioritized)	Before January 1, 2018
Language	English	Non-English

Figure 1. Patient Journey Schematic



Results

- Ten publications were identified,[1-10] encompassing 16 rare diseases
- Immune-related conditions were the most studied (3/16, 19%)
- Most studies (n=6)[2-5,7,8] employed a prospective observational design and were conducted in Europe (n=7)[1-3,6-8,10]
- Patient survey was the most common data collection method used (n=4)[1,2,4,7,8] **(Figure 2)**; Two studies reported more than 1 data collection method[1,5]
- For journey stages, all 10 studies investigated 'Pre-Diagnosis/Screening' and 'Diagnosis,' and none investigated 'Awareness' or 'Adherence' **(Figure 3)**
- All studies reported multiple outcomes (44 reported overall). Pre- and post-diagnosis 'Symptoms' were assessed most frequently in all 10 studies,[1-10] followed by 'Quality-of-Life' (n=9)[1-8,10] and 'Healthcare Resource Utilization' (n=7)[2-5,7-9] **(Figure 4)**
- Few studies assessed economic burden outcomes, such as 'Costs,'[1,3] 'Productivity,'[1,3] or 'Caregiver/Family Burden'[1,10] (n=2 each) **(Figure 4)**

Figure 2. Data Collection Methods Employed (n=10)

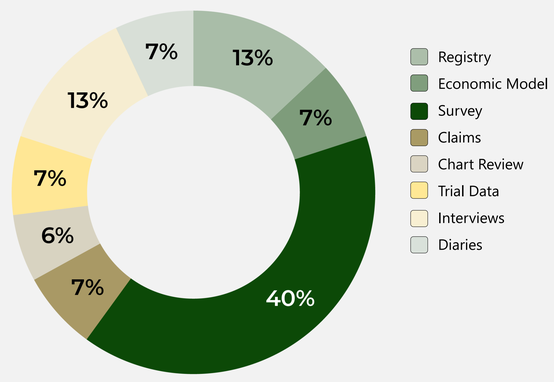


Figure 3. Frequency of Patient Journey Stages Assessed (n=10)

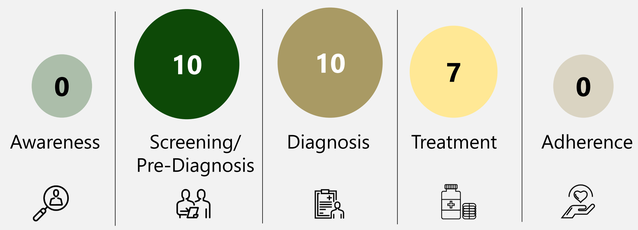
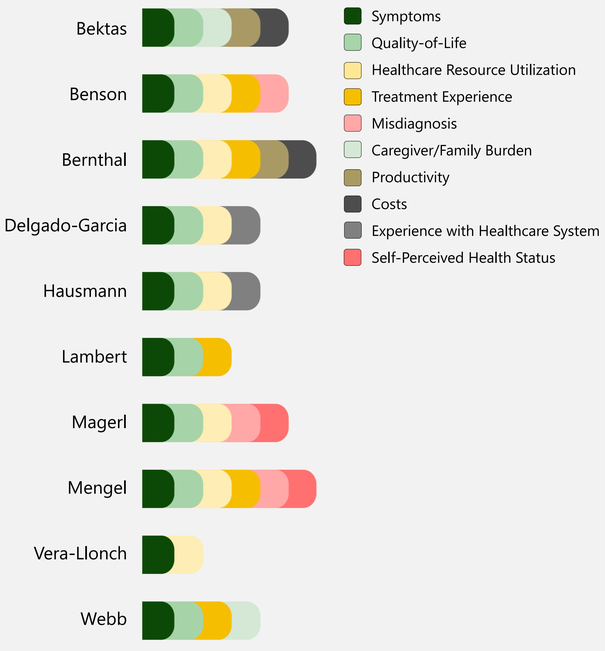


Figure 4. Reported Outcomes by Article



Conclusions

- This literature review revealed that rare disease patient journey assessments primarily use patient surveys, emphasizing symptoms and quality of life through diagnosis
- Treatment adherence, productivity, and family impact are more infrequently explored and may warrant further research to facilitate comprehensive patient care

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Acknowledgements: Alkemi thanks Lucas Blackmore for his assistance preparing this poster

Presented at ISPOR, May 2024, Atlanta, Georgia. Please contact betsy.lahue@alkemihealth.com for more information.
www.alkemihealth.com