ORACLE Life Sciences

PCR208

Registry Participation in Rare Diseases: Insights from Stakeholders and an Adapted Targeted Literature Review (ATLR)

Nivantha Naidoo[1], Alissar Mousallem[1], Joseph Milce[1], **Lysel Brignoli[1]**, Alexa Berk[2], Thomas Neumann[3], Bertold Ries[4], Camilla Wensing[5], Perrine Le Calvé[1]

[1]Oracle Life Sciences, Paris France, [2]Oracle Life Sciences, US, [3]Division of Rheumatology, Cantonal Hospital St. Gallen, Switzerland, [4]Independent (at the time of the research), Switzerland, [5]Swiss Haemophilia Society, Switzerland

Background

Registries are an essential research tool for advancing rare disease diagnosis, management, treatment, and long-term outcomes.

Registry engagement and participation can pose important administrative, logistical, and privacy-related burdens.[1–3]. Nevertheless, patients with rare diseases, and their caregivers, experience meaningful benefits, including contributing to research and advocacy, gaining access to information, and connecting with support networks.

Understanding the characteristics of the burdens and benefits is essential to informing registry design, improving user experience, and ultimately strengthening participation and retention.

Objectives

We aimed to explore key themes related to registry engagement and participation, including motivators, benefits, facilitators, and enablers of recruitment and retention, as well as burdens, barriers, and challenges.

Methods

Our approach consisted of two complementary steps:

1. Stakeholder Workshop: As part of a Rare Disease Summit, Oracle Life Sciences (OLS) hosted a workshop titled *"Unlocking Registry Success – Best Practices for Patient Engagement"*. The session explored effective strategies for identifying, recruiting, and retaining patients in rare disease registries, with an emphasis on fostering meaningful and sustained engagement. Seven summit attendees participated, representing a diverse mix of stakeholders: patients, leaders from patient advocacy organizations, a clinician, a representative from a life sciences company, and OLS experts in registry design and operations.

2. Adapted Targeted Literature Review (ATLR)[4,5,6]: An ATLR was conducted to rapidly identify, analyze and synthesize the most recent and relevant evidence on patient and caregiver perspectives regarding participation in rare disease registries. A targeted search algorithm was applied to MEDLINE to identify English-language, peer-reviewed journal articles published since 2020. In addition, a targeted web-based sites search was performed using predefined keywords. The EURORDIS ***"Voice of Rare Disease Patients"*** (2013) report[7] was used as a foundational resource to inform a preliminary thematic framework and guide data extraction.

Results

Insights from the Stakeholder Workshop

The workshop was conducted in two 90-minutes sessions, guided by several open questions that stimulated discussions around recruitment, long-term engagement, and co-creation. During these discussions, four thematic domains were identified as critical motivators to drive recruitment and sustain participation in rare disease registries:

- **Trust and credibility**: patients are more likely to engage in registries when introduced by trusted sources such as physicians. Clear communication on data privacy and data protection, equitable incentives and ability to provide feedback are also key motivators.
- **Personal benefits:** participants emphasized the need for a clear understanding of community benefits and individual gains; as well as relevance of the data collected for the participants.
- **Ease of participation:** A simplified, user-friendly experience (e.g., short follow-up surveys, gamification, flexible ways to participate) was seen as essential to lowering burden and encouraging long-term participation.
- **Humanistic connection:** A sense of belonging and being valued was highlighted through regular communication, community engagement opportunities, and recognition of the importance of the patients' perspective.

Detailed motivators within each thematic domain are illustrated in **Figure 1.**

Figure 1: Findings from the Stakeholder Workshop

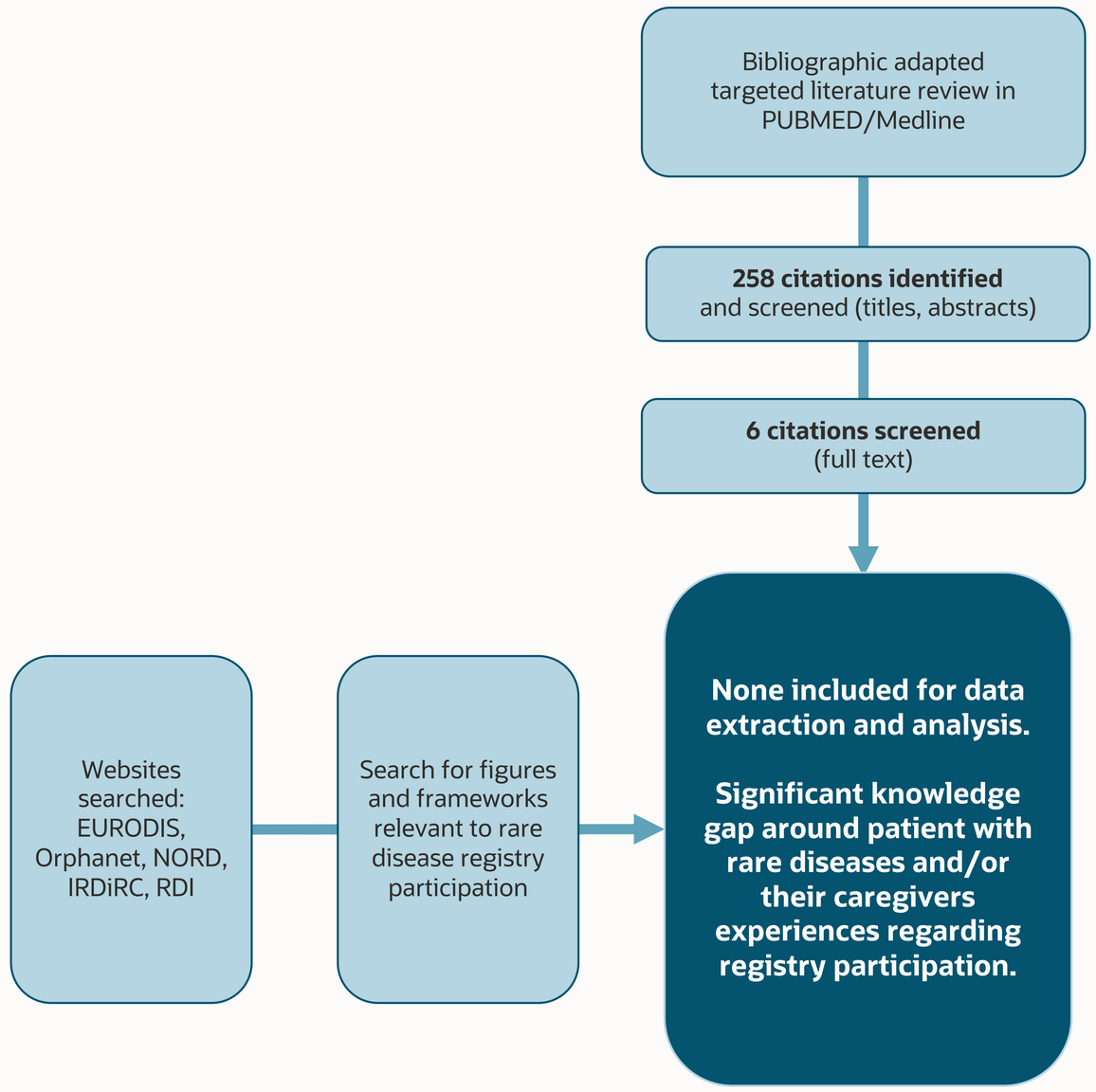
ATLR findings

The targeted searches identified 258 records for screening. Six studies were eligible for full-text review; however, none were included for extraction.

Notably, two studies examined patient experiences in the context of rare disease research participation, but their focus was limited to clinical trial settings, not registry participation. The other four studies focused on rare disease registry building, maintenance, and other operational aspects. All these studies lacked direct insight into the lived experiences of patients with respect to registry engagement specifically.

Despite refining the search strategy to be more sensitive (i.e., reducing specificity and inclusion thresholds), no additional eligible studies were identified (**Figure 2**).

Figure 2: Output from the multi-resource review (ATLR and web-based review)



Conclusion

These findings highlight a critical knowledge gap: there is little to no published evidence on the experiences of patients with rare diseases and their caregivers, in registry participation and engagement. Combined with insights from the workshop, this indicates the need to collect patient and caregiver perspectives. These perspectives are essential to inform patient-centered engagement strategies that support recruitment, retention, and the long-term success of rare disease registries.

Disclosure

N.S-N., A.M., J.M., L.B., A.B. and P.L.C are employees of Oracle Life Sciences. **T.N.** received financial support from Oracle for participation in the workshop related to the subject of this abstract. **B.R.** is employee of Celgene International Sàrl, a Bristol Myers Squibb company. **C.W.** is affiliated with the Swiss Haemophilia Society.

Acknowledgments

The authors would like to thank Tim Irfan, for organizing the Oracle Life Sciences Rare Disease Summit and all participants for their valuable contributions to all workshops and discussions.

References

1. Stratton C, et al. JBI Evid Synth. 2025;23(3):546–53.
2. Hageman IC, et al. Orphanet Journal of Rare Diseases. 2023;18(1):106.
3. Boulanger V, et al. Pharm Med. 2020;34(3):185–90.
4. Subiron-Naidoo N, et al. Value in Health, 2024;27(12):S2
5. Subiron-Naidoo N, et al. Value Health. 2024;27(12):S2
6. Tagliabue S, et al. Value Health. 2024;27(12):S2
7. EURORDIS. The Voice of Rare Disease Patients. 2025. Available from: https://www.eurordis.org/publications/the-voice-of-rare-disease-patients-experiences-andexpectations-of-over-3000-patients

Scan to download a copy of this poster

Copies of this poster and its content, obtained through this QR code, are for personal use only and may not be reproduced without written permission from the authors