

Patient-Centric Approaches in Rare Disorder Research: An in-Depth Systematic Review

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Verma A¹, Chinchapattanam D¹, Barathe P¹, Krishna A¹, Pruthi J¹, Rai MK², Prasanna R¹

¹EVERSANA, Mumbai, India, ²EVERSANA, Singapore, Singapore

INTRODUCTION

- Rare disorders, which collectively affect millions globally despite the rarity of individual conditions, often lack sufficient research, treatment options, and awareness, leading to delayed diagnoses and limited therapeutic interventions.
- In recent years, there has been a growing emphasis on integrating patient perspectives into research for rare disorders. This approach aims to ensure that treatments and interventions are more aligned with patient needs, improving overall quality of life and care experiences.

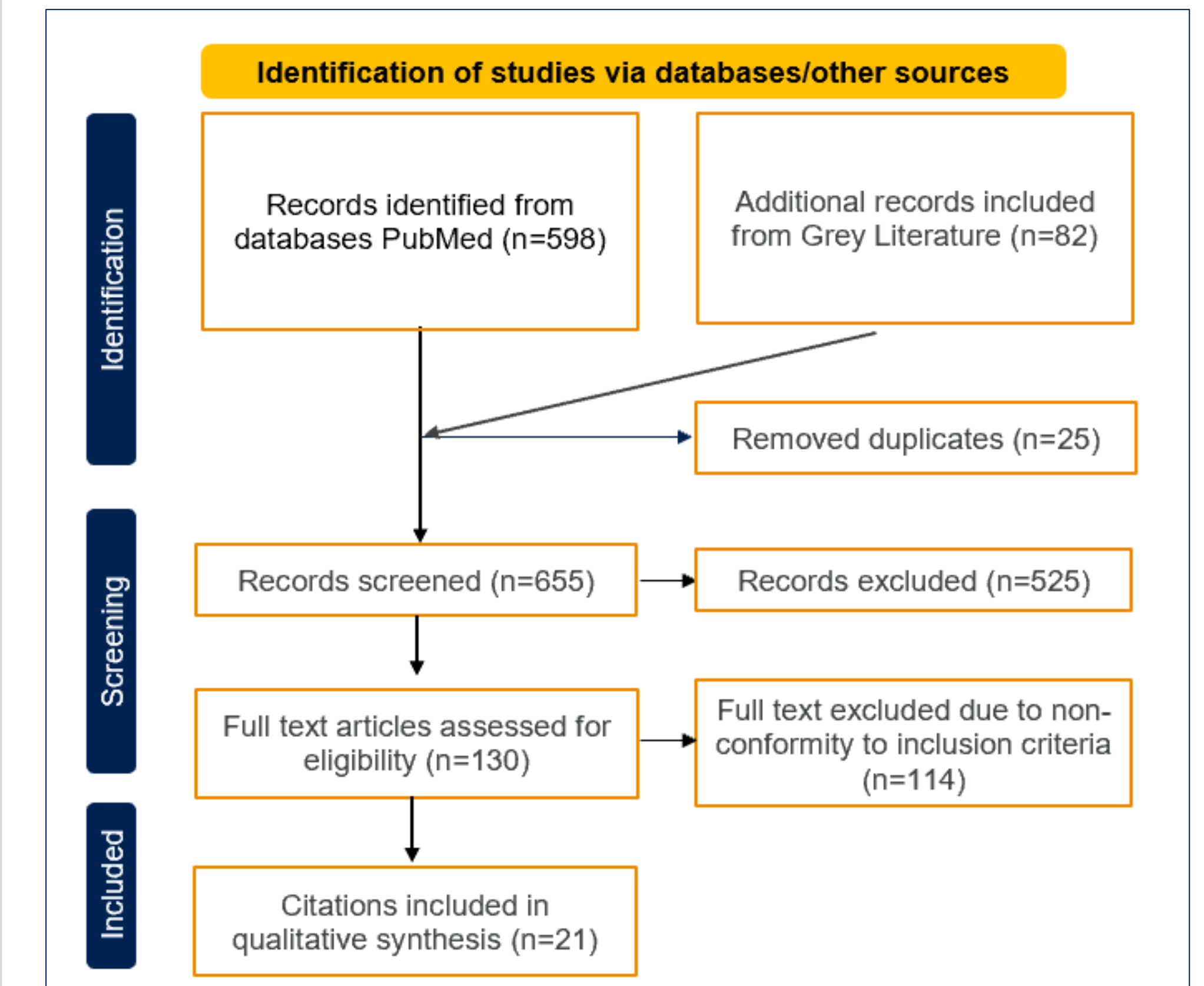
OBJECTIVE

- Patient-centric approaches are increasingly recognized as pivotal in rare disorder research, aiming to enhance care and outcomes tailored to individual patient needs. This systematic review explores various methodologies and outcomes of patient-centric approaches in rare disorder research.

METHOD

- A systematic literature search was conducted across PubMed and Google Scholar databases between 2014-2024 to identify studies focusing on patient-centric approaches in rare disorder research.
- Keywords included "Patient-Centric," "Rare Disorders," "Patient Engagement," and related terms.
- Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines were followed to conduct this systematic review (Figure 1).
- Studies that addressed patient involvement, engagement, or patient-reported outcomes in rare disorders were included.

Figure 1: PRISMA flowchart to summarize selection process



RESULTS

- A total of 680 studies from database search and grey literature were screened, with 130 undergoing secondary screening. Ultimately, 16 studies met inclusion criteria and were included in the final analysis (Figure 1).
- The studies were globally distributed, with a majority from the United States (n=6), followed by Europe (n=7), and global (n=3) (Figure 2).
- Methodologies and rare disorders covered included Duchenne muscular dystrophy (n=3), Cushing's syndrome (n=1), Castleman disease (n=1), and others/not specified (n=9) (Table 1).
- Patient-centric approaches in rare disorder research exhibited varied successes. Significant enhancements in healthcare efficiency were observed, particularly in reducing hospital stays and procedure times.

Figure 2: Geographical distribution of studies

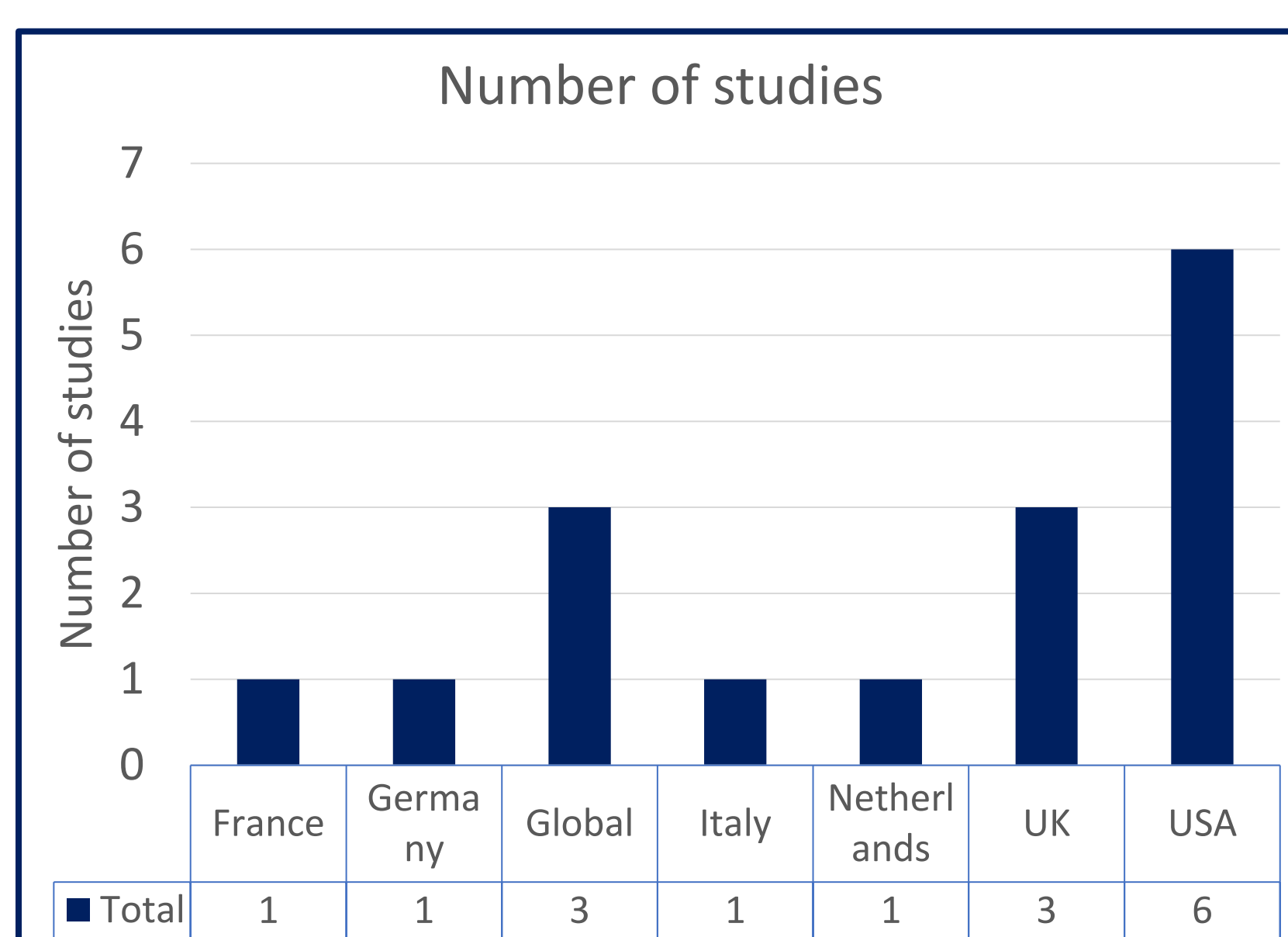


Table 1: Summary of studies on patient-centered research in rare diseases

Author	Year of publication	Study Type	Participants	Condition	Focus
Babac et al.	2019	Qualitative research	Patients, family, healthcare pros	Rare disease research	Patient-centered
Denger et al.	2019	Mixed method	Patients and caregivers	Duchenne Muscular Dystrophy (DMD)	Patient involvement
Courbier et al.	2019	Quantitative survey	Patients with rare diseases	Rare disease research	Patient engagement
Gaasterland et al.	2019	Qualitative research	Patient Think Tank (PTT)	Rare disease trial	Patient involvement
Bate et al.	2018	Mixed method	Patients with Ewing's Sarcoma	Ewing's sarcoma	Patient perspective
Gaasterland et al.	2018	Qualitative research	Patient representatives	Rare disease trial	Patient participation
Swezey et al.	2019	Qualitative research	Adults with Osteogenesis imperfecta	Osteogenesis imperfecta (OI)	Patient perspective
Stein et al.	2018	Qualitative research	Advocacy group; nine experts	Rare disease research	Patient-centered
Baldo et al.	2016	Qualitative research	Patients and TNGB	Rare disease biobanking	Patient-centered
Morel et al.	2016	Mixed method, Discrete-choice experiments (DCE)	Patients and families with rare diseases	Group of rare diseases	Patient-centered outcomes
Korsunska et al.	2023	Observational	Castleman disease network	Castleman disease	Patient-centered
Crossnohere et al.	2021	Cross-sectional	263 participants, USA or UK	Duchenne muscular dystrophy	Patient-centered
Smith et al.	2021	Community-advisor meetings	30 parents of children with DMD	Duchenne muscular dystrophy (DMD)	Patient engagement
Francisco et al.	2022	E-survey	209 CDG and 349 control participants	Congenital Disorders of Glycosylation (CDG)	Patient-centric
Ciesluk et al.	2022	Elicitation interviews	33 participants (13 patients, 20 caregivers)	NUT (nuclear protein in testis) carcinoma	Patient-centric
Vanderhout et al.	2021	Evidence review, Delphi survey, and workshop	Patients/caregivers of PKU or MCAD	Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) and phenylketonuria (PKU)	Patient engagement

- Crowd sourced initiatives proved effective in fostering patient-centered agendas but showed limitations in generalizability.
- Challenges such as logistical barriers and biased participant advocacy hindered engagement.
- E-research methods displayed potential but encountered ethical and participation challenges.
- Validation of patient-reported outcomes highlighted issues like small sample sizes and incomplete symptom assessments, underscoring the need for more comprehensive research efforts.

CONCLUSIONS

- Patient-centric approaches in rare disorder research show promising outcomes but face challenges such as limited generalizability and small sample sizes.
- Future research should focus on refining methodologies, improving inclusivity, and validating patient-reported outcomes to enhance the relevance and impact of patient-centric strategies in rare disorder management.

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CONTACT INFORMATION

Dr. Mahendra Rai,
Vice President & Regional Head, HEOR, RWE, Medical Affairs, EVERSANA APAC
Mahendra.Rai@Eversana.com