

Abstract Template

Title of Abstract

Therapeutic trajectories of patients and families with rare diseases: a narrative literature review

[Carla Campaña]^{1*}, [Báltica Cabieses]², [Gabriela Repetto]³,

¹ Universidad del Desarrollo, Santiago, Chile

² Universidad del Desarrollo, Santiago, Chile

³ Universidad del Desarrollo, Santiago, Chile

*Presenting author: cacampanac@udd.cl

Background & Objectives

Rare diseases (RD) are a large group of highly heterogeneous disorders, defined according to incidence. Given their rarity and complex manifestations, the patient and family experiences are unique. Research on patients' therapeutic trajectories is important to understand these experiences and improve health care. Research on therapeutic trajectories from the perspective of patients and families has grown in the past decade, yet studies on RD are limited and diverse. Our objective was to develop a narrative synthesis of existing scientific evidence in order to answer the question: which are the most frequent therapeutic trajectories and/or odysseys of patients and families (or caregivers) with RD, according to existing scientific evidence?

Method (s) and Results:

We conducted a narrative literature review. The search was performed in PubMed (May 2021), including studies with trajectories of RD patient and their families or caregivers, without filters. Of the 574 titles identified, 53 studies met the inclusion criteria and were selected for the narrative review. Most studies were from Europe (n=20), case study (n=23) and cross-sectional (n=16). Twenty were quantitative and 27, qualitative and 6 mixed studies. We identified six main themes: diagnosis, treatment, cost, quality of life, key informant, and technology contributions. The main topics are time, diagnosis, information, new diagnosis, and treatment strategies.

Conclusions (Significance and Impact of the Study):

Findings from this narrative review suggests that understanding patients and families therapeutic trajectories could help recognize effective time-to-diagnosis, treatments, and the responsiveness of healthcare systems at point of care. Findings indicate that the perspective of patients and families with RD must be included as part of the research and policy agenda, as their experience is vital to shed light on global healthcare quality and equity goals. Generating this information in other countries and contexts is relevant to develop public policies for rare diseases, from the experience of patients and.

Conflict of interest disclosure: The authors declare no potential conflicts of interest, whether scientific, financial and personal.

Keywords: rare disease, patient trajectory, patient odyssey, health system.