

Experiences of people living with a rare disease with healthcare:

A SCOPING REVIEW FOR THE H-CARE PROJECT

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Background

People living with a rare disease (PLWRD) are a heterogeneous population faced with complex, and specific healthcare challenges. Patient experience is an increasingly valued healthcare quality indicator, with patient reported experience measures (PREMs) emerging as serviceable instruments^{1,2}. To capture the experiences of PLWRD within secondary and/or tertiary healthcare settings, we conducted a scoping literature review.



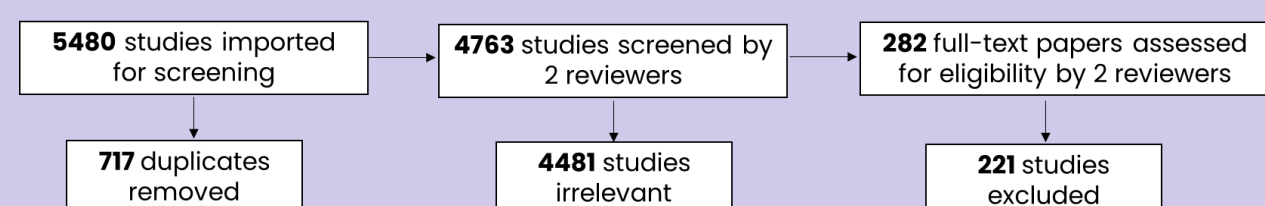
Methodology

Medline, Embase, and CINAHL were searched using :
(a) keywords for rare or complex diseases, and
(b) keywords for patient experience or healthcare quality
Papers from 2005 to September 2022 were retrieved and analysed. Paper screening, selection and data extraction were performed on Covidence by two blinded reviewers.

Aims

- 1 Identify and extract the domains and categories of healthcare experience included in empirical studies exploring the views of PLWRD.
- 2 Inform a first definition of healthcare experience for PLWRD, defining whether and/or which rare disease-specific aspects it includes.
- 3 Map and assess existing questionnaires against the identified domains and categories to determine whether they allow to fully capture PLWRD healthcare experience in a comprehensive way, and if proved insufficient, establish what should be added or developed.

Results



1. Study design



61% (37/61): Quantitative methods
21% (13/61): Qualitative methods
10% (6/61): Mixed methods
8% (5/61): Qualitative-quantitative



67% (41/61) performed validation activities



49% (30/61) involved patients and/or carers in instrument design and implementation.

2. Study scope

59% (36/61) are single disease 85% (52/61) are country-specific

61
included
papers

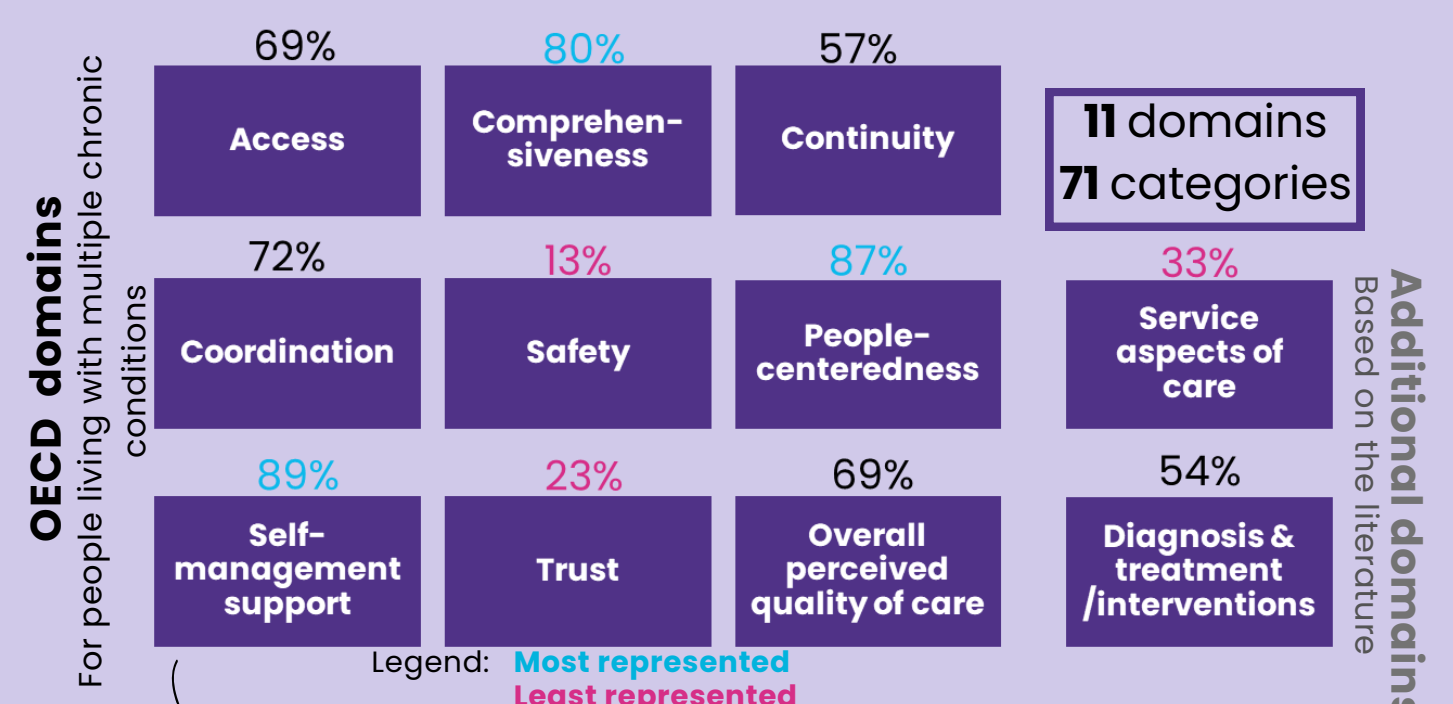
3. Target population

43% (26/61) Patient-only experiences
(mostly adult patients)

18% (11/61) Proxy-only experiences
(carers usually parents)

4. Healthcare experiences of PLWRD

Healthcare experience domains distribution (%) among the 61 included papers



42 questionnaires were mapped:

- the Cystic Fibrosis Patient Experience and Satisfaction with Care Services questionnaire covers all 11 domains and the highest number of categories (31/71)

Conclusion

This study outlines rare disease secondary and tertiary healthcare experience from a patient lens, putting forward a preliminary definition of what healthcare experience for PLWRD is and entails. Our findings must be discussed and validated with representatives from diverse groups from the rare disease community.

References

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