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SUMMARY

OBJECTIVES

- The use of patient reported outcomes (PROs) in Leber's hereditary optic neuropathy (LHON) research offers a direct window into the experiences and challenges faced by those affected by the condition.
- This research aims to provide an overview of the PROs used in LHON research and the insights they offer into the life of affected individuals.

METHODS

- We conducted a targeted literature review of PROs reported in LHON patients up to 6th June 2023.
- Embase, MEDLINE, Econlit, and EBMR (Cochrane) databases were searched for terms related to LHON, PROs, and quality-of-life (QoL) data.
- Identified articles were assessed for inclusion by a single reviewer based on title and abstract screening, and subsequently full text screening.
- Data was extracted from included studies regarding the PROs used and the impact of LHON as assessed by instrument use.

FINDINGS

- Research on PROs in LHON has mainly employed standardised questionnaires/surveys to assess vision-specific QoL and psychosocial wellbeing.
- These instruments include the 25-Item National Eye Institute Visual Function Questionnaire (NEI-VFQ-25), the Visual Functioning Questionnaire (VFQ-25), and the Vision-related Quality of Life questionnaire (VR-QOL).
- All currently available PROs lack specificity to LHON, showing that there is a need to develop LHON-specific measures that ensure the unique factors of LHON are fully described and captured.

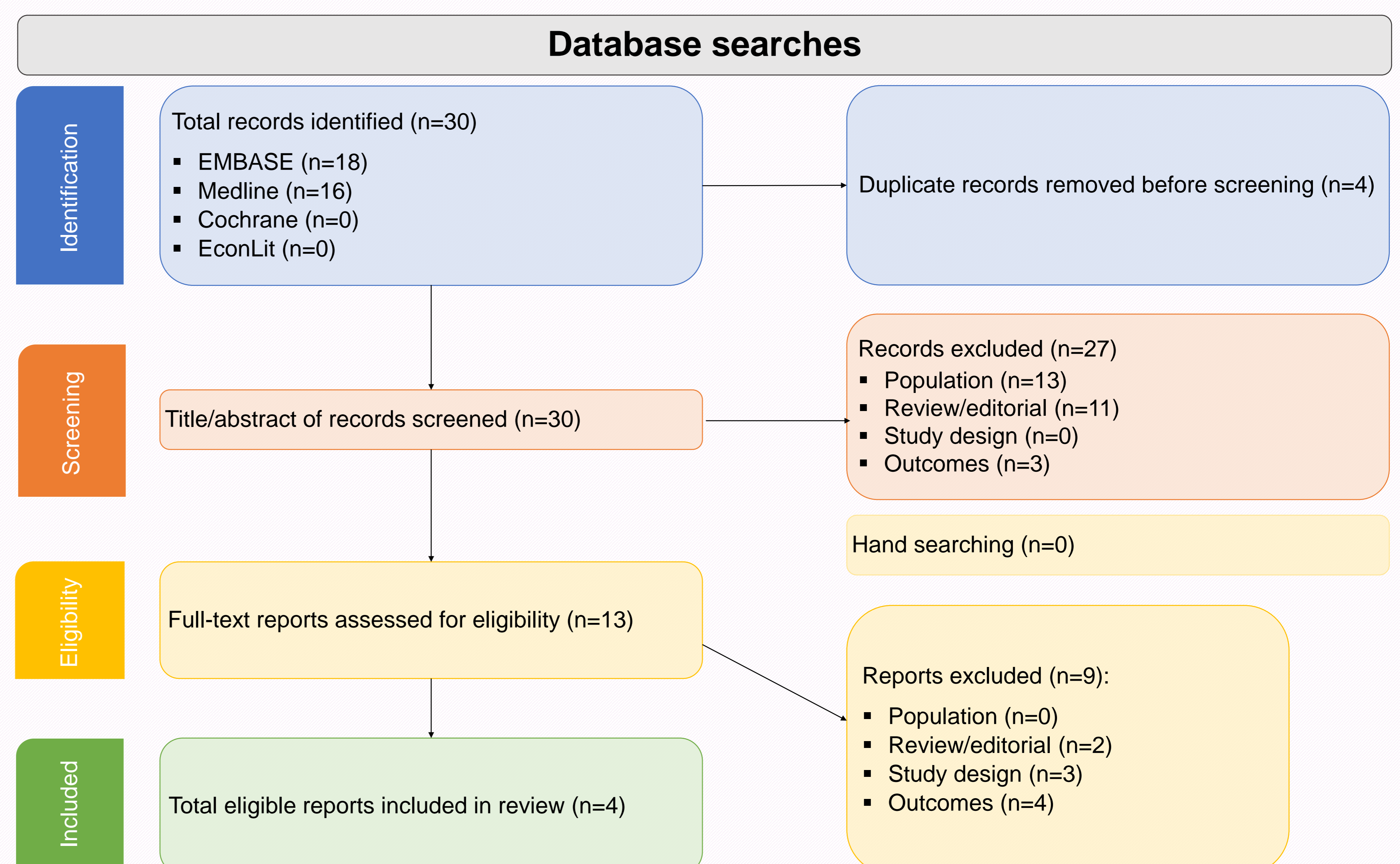
BACKGROUND & AIMS

- LHON is a rare genetic disorder which causes retinal ganglion cell degeneration
- This degeneration can result in severe vision loss or blindness.
- Patient-reported outcomes (PROs) play a crucial role in assessing the impact of diseases on quality of life (QoL), and understanding patients' experiences and needs.
- The use of PROs in LHON research is particularly significant because it offers a direct window into the experiences and challenges faced by those affected by this condition. Given the unique and multifaceted impact LHON has on visual function and psychosocial well-being, traditional clinical metrics may not capture the full scope of patient experiences. PROs, in contrast, allow individuals to express their perspective regarding how LHON influences their quality of life, psychological well-being, and daily functioning.
- This targeted literature review sought to compile and synthesise the existing body of research concerning the application of PROs in LHON. By examining this literature, it aimed to provide a comprehensive understanding of how PROs are used to assess the impact of LHON on patients' lives, furthering our insights into this rare genetic disorder and its implications for patient care and quality of life.

METHODS

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- Identified articles were assessed for inclusion by a single reviewer based on title and abstract screening, and subsequently full text screening.
- Data were extracted from included studies, regarding the PROs used, and the impact of LHON as assessed by the instrument used.
- By employing this rigorous methodology, this targeted literature review aims to provide a comprehensive and insightful analysis of how PROs have been utilised in LHON research, shedding light on the experiences and needs of individuals affected by this rare genetic disorder. This methodology ensures a robust foundation for the examination of the impact of LHON on patients' quality of life and psychosocial well-being.

Figure 1: PRISMA diagram



RESULTS

- 30 papers were identified in the initial search, with 4 included after abstract and full-text screening (Figure 1).
- Selection was based on whether the study aimed to measure either PRO or QoL in LHON.
- Research on PROs in LHON has mainly employed standardised questionnaires/surveys to assess vision-specific QoL and psychosocial well-being.
- These instruments include the 25-Item National Eye Institute Visual Function Questionnaire (NEI-VFQ-25), the Visual Functioning Questionnaire (VFQ-25), and the Vision-related Quality of Life questionnaire (VR-QOL). All of these lack specificity to LHON.
- Studies using these tools consistently report significant LHON-related impairments in vision-related functioning and QoL domains, such as general vision, near and distance activities, colour vision, and social functioning.
- PROs also highlight the psychosocial impact of LHON, including feelings of frustration and depression, and reduced overall life satisfaction.
- Additionally, PROs show that people with LHON often employ adaptive strategies and coping mechanisms to deal with the burden of their disease.
- Studies exploring patient narratives and qualitative interviews have featured themes of resilience, acceptance, and the importance of support systems in adjusting to life with the disease.

CONCLUSIONS

- The integration of PROs into LHON research is important for informing clinical management and identifying areas for intervention.
- There is a clear need for PROs specific to LHON to be developed.
- By capturing patients' perspectives and priorities, PROs help to guide reimbursement and treatment decisions.
- Furthermore, PROs allow emerging therapies and interventions to be evaluated on the basis of patient-centred outcomes, enhancing the development of patient-centred care approaches.

References

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