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## SUMMARY

### OBJECTIVES

- Severe visual impairment affects not only patients but also their informal caregivers, whose quality of life (QoL) is often overlooked.
- Approximately 900 rare diseases can lead to sight loss, many without effective treatments, creating significant psychosocial and practical burdens for caregivers<sup>1</sup>.
- The Objective of this research is to systematically review the association between caregiver QoL and the care provided to adults with severe visual impairment, with a focus on rare eye diseases such as Retinitis Pigmentosa (RP) and Leber's Hereditary Optic Neuropathy (LHON)<sup>2,3</sup>.

### METHODS

- A PRISMA-adherent systematic literature review<sup>5</sup> was conducted using Embase, MEDLINE, and Cochrane Library databases. Studies published before June 2024 were included if they explored caregiver burden and QoL in adults (≥18 years) with sight loss. Data were analysed using qualitative content analysis<sup>6</sup>.

### RESULTS

- Six publications met the eligibility criteria. Key themes included:
  - Negative association between severity of visual impairment and caregiver anxiety, spousal strain, and intensity of informal care.
  - Influence of patient and caregiver demographics on caregiving experience.
  - High prevalence of depressive symptoms and overcontrolled coping among caregivers, with female caregivers most at risk.
- Survey data from LHON patients<sup>4</sup> published in a NICE technology appraisal corroborated these findings, revealing significant emotional, financial, and social challenges for caregivers.

### CONCLUSIONS

- Caregiver QoL is profoundly impacted by adult sight loss yet remains under-researched.
- Current evidence highlights urgent need for interventions addressing caregiver mental health, coping strategies, and financial support.
- Future Research Directions:
  - Development and evaluation of targeted psychosocial interventions for caregivers.
  - Longitudinal studies to assess changes in caregiver burden over time.
  - Exploration of cultural and socioeconomic factors influencing caregiver experiences.
  - Integration of caregiver QoL metrics into health technology assessments (HTAs) for rare eye disease treatments.

## BACKGROUND & AIMS

- Severe visual impairment in adulthood has profound implications not only for affected individuals but also for their informal caregivers. Rare eye diseases such as Retinitis Pigmentosa (RP) and Leber's Hereditary Optic Neuropathy (LHON) are progressive, lifelong conditions with limited disease modifying treatment options, resulting in sustained reliance on informal care. Across Europe, approximately 900 rare diseases are associated with sight loss, many of which fall under the remit of the European Reference Networks for rare eye diseases<sup>1</sup>.
- While patient reported outcomes in visual impairment are increasingly recognised, caregiver quality of life (QoL) remains underrepresented in clinical, economic, and health technology assessment (HTA) decision making. Informal caregiving in this context extends beyond physical assistance, encompassing emotional support, navigation of healthcare systems, financial management, and social adaptation. Understanding how caregiver QoL interacts with the nature and intensity of care provided is critical to informing holistic care models and value assessments, particularly for rare diseases where societal burden extends well beyond the patient.
- The objective of this research was to systematically review the evidence examining the association between caregiver quality of life and the care provided to adults with severe visual impairment, with specific emphasis on rare inherited eye diseases such as RP and LHON.
- Secondary objectives included:
  - Identifying key psychosocial and demographic drivers of caregiver burden
  - Characterising patterns of informal care provision in adult sight loss
  - Assessing the extent to which caregiver outcomes are captured in existing evidence relevant to HTA decision making

## METHODS

- A systematic literature review (SLR)<sup>7</sup> was conducted in accordance with PRISMA 2020 guidelines. Searches were undertaken in MEDLINE, Embase, and the Cochrane Library, including studies published up to June 2024.
- Eligibility criteria:
  - Adult patients (≥18 years) with severe visual impairment or blindness
  - Studies reporting caregiver burden, caregiver QoL, or caregiving intensity
  - Observational, qualitative, and mixed methods studies
  - English language publications
- Studies focused exclusively on paediatric populations or non informal care settings were excluded. Data extraction focused on caregiver demographics, caregiving activities, psychosocial outcomes, and reported associations with disease severity.
- Qualitative content analysis was applied to synthesise findings and identify recurring thematic patterns using publish guidelines<sup>8</sup>

## RESULTS

- Six publications met the inclusion criteria. Evidence consistently demonstrated a negative association between severity of visual impairment and caregiver QoL. (figure 1)
- Key themes identified:
  - Psychological burden and emotional strain** - Increasing severity of sight loss was associated with elevated caregiver anxiety, depressive symptoms, and emotional exhaustion. Spousal caregivers in particular reported high levels of strain linked to continuous caregiving demands.
  - Intensity and scope of informal care** - Caregivers provided extensive unpaid support, including daily functional assistance, mobility support, and administrative tasks. Higher care intensity correlated with poorer reported QoL outcomes.

- Demographic influences on caregiver experience** - Caregiver gender, age, employment status, and relationship to the patient influenced reported burden. Female caregivers were disproportionately affected, with higher prevalence of depressive symptoms and constrained coping strategies.
- Corroboration from NICE submitted patient evidence<sup>4</sup> - Survey data from LHON patient submissions included in NICE technology appraisals highlighted significant emotional, social, and financial impacts experienced by caregivers, reinforcing findings from the published literature.
  - NICE TA1093<sup>3</sup> gave considerable insight into the impact on care givers for Adults with sight loss. Patient and caregiver quotes give real strength and depth of understanding to the impact and caregiver burden (figure 2)
  - Patient research for the technology appraisal also concluded that "When asked the number of carers people had the respondents show an average of 1.58 carers, on an average of 24.8 days out of 31 per month (83% of the time). This would reasonably translate to an average of 1.30 carers continually. This finding is consistent across adults and children.

## DISCUSSION

- This review highlights a critical gap between the lived experience of caregivers supporting adults with severe visual impairment and the way value is currently assessed in healthcare decision-making. Although patient-reported outcomes in visual impairment are increasingly incorporated into clinical trials and HTAs, caregiver outcomes remain peripheral, despite caregivers providing essential, unpaid care that sustains functional independence and quality of life for affected adults.
- The findings suggest caregiver burden is not solely driven by the presence of visual impairment, but by its severity, progression, and associated loss of patient autonomy, which collectively increase both the intensity and emotional salience of caregiving. Demographic factors particularly gender, spousal relationship, and employment disruption further exacerbate vulnerability to poor caregiver QoL outcomes.
- The small number of eligible studies reflects broader structural challenges in rare disease research. Heterogeneity of conditions, limited sample sizes, and a historical focus on patients rather than caregivers have constrained evidence generation. Nevertheless, triangulation with NICE committee testimony and patient surveys strengthens the validity and relevance of the findings.
- From a health policy and HTA perspective, these results reinforce the need to move beyond a patient-only lens. Interventions that slow disease progression or maintain functional vision may deliver indirect but meaningful benefits by reducing caregiver burden, preserving household productivity, and mitigating downstream psychosocial costs. Incorporating caregiver QoL metrics into evidence generation and HTAs would support more holistic and realistic assessments of value particularly for rare diseases where informal care represents a substantial component of total disease burden.

Figure 1. PRISMA flow diagram.

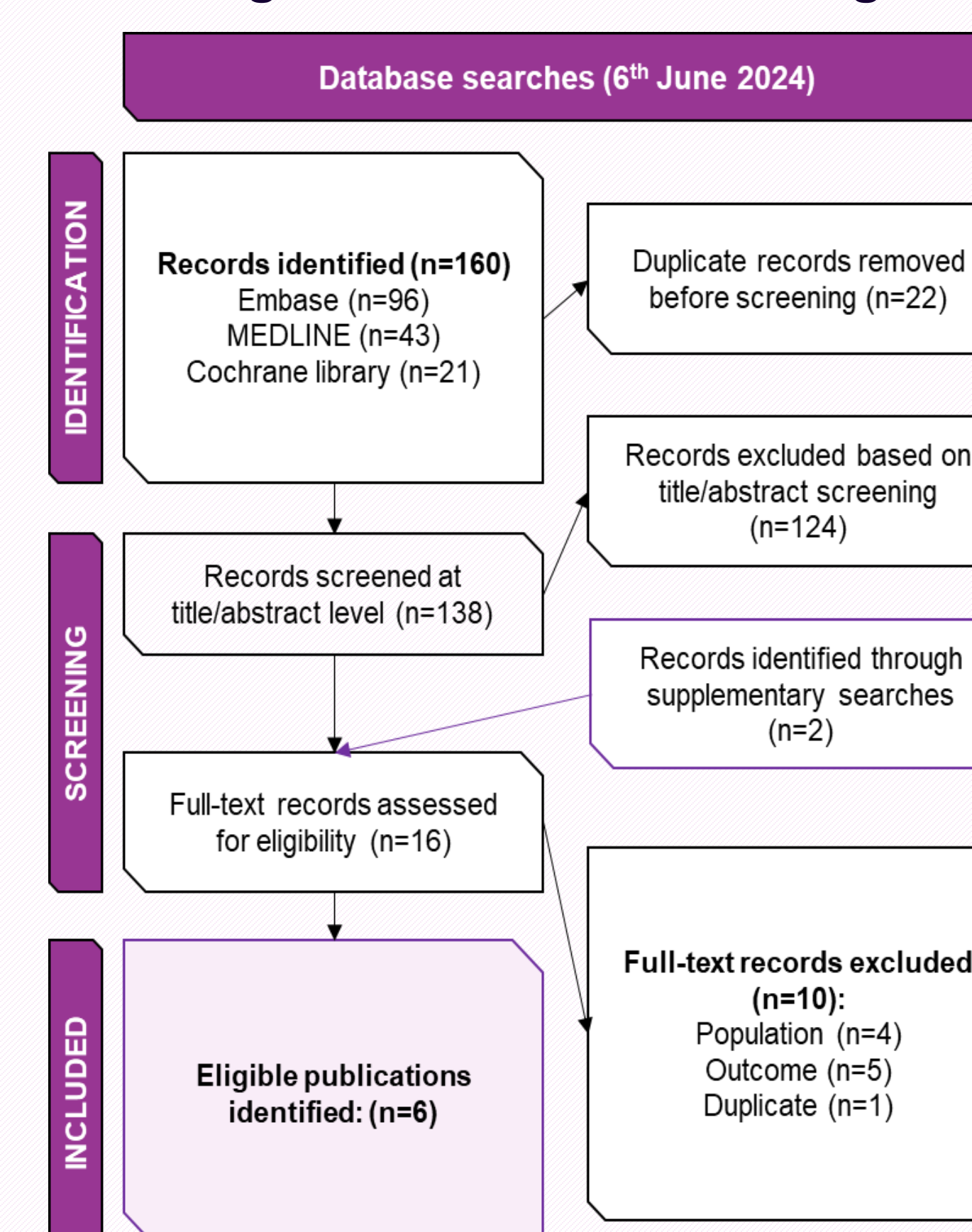


Figure 2. Patient and carer impact as reported in NICE TA1093

**Living with sight loss, what help do you need from others?**

- "I need help and support from my family. I need help with shopping, housework, personal finances, cooking and getting around."
- "Support includes every aspect of daily life and limits bigger opportunities."
- "I can't live without the help of others."
- "I require support from family with day to day living with banking and food shopping, hard to see prices on clothes and food. Travelling, navigating my way around need help from others."

**What is the impact on those who help people with sight loss?**

- "Financially, emotionally, every way possible it's such an awful disease."
- "Not only I had to stop working, but my wife has struggled at work because of the amount of additional responsibility. Before the tasks were shared as well as raising the family and working. Now she has to handle all of it, plus support me."
- "Emotionally painful to see your loved one no longer able to do basic daily life tasks as they once could and not able to do activities, they loved anymore."

## CONCLUSIONS

- This systematic review demonstrates a clear and consistent association between severity of adult visual impairment caused by rare diseases and reduced caregiver quality of life. Across the limited but convergent evidence base, caregivers experience substantial psychological, emotional, and functional burden that increases in parallel with care intensity and disease progression.
- Caregiving in rare eye diseases such as Retinitis Pigmentosa and Leber's Hereditary Optic Neuropathy extends well beyond episodic support. Informal carers frequently provide sustained assistance with daily living, mobility, administration, and emotional support, often for the majority of days each month. This level of engagement is associated with heightened anxiety, depressive symptoms, role strain, and reduced participation in employment and social life particularly among spousal and female caregivers.
- Importantly, patient and caregiver reported evidence submitted to NICE technology appraisals corroborates the published literature, highlighting significant emotional, financial, and social impacts that are rarely captured within formal assessments of clinical benefit and cost-effectiveness.
- Overall, caregiver quality of life is profoundly impacted yet systematically under-represented in research, policy, and HTA decision-making related to rare eye diseases. Failure to account for caregiver outcomes risks underestimating the full societal burden of sight-threatening rare diseases and the broader value of interventions that preserve or stabilise vision.**

## References

- European Reference Networks (ERNs), Eye Diseases. <https://www.ern-eye.eu/ern-eye-a-european-reference-network-dedicated-to-rare-eye-diseases/>
- NICE. Available at: <https://www.nice.org.uk/guidance/hst11> (Accessed: 6th January 2026)
- NICE. Available at: <https://www.nice.org.uk/guidance/ta1093> (Accessed: 6th January 2026)
- LHON patient survey results in NICE committee papers available at: <https://www.nice.org.uk/guidance/ta1093/documents/committee-papers-4> (Accessed: 6th January 2026)
- Page et al. (2021). The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. *bmj*, 372.
- Vaismoradi (2016). Theme development in qualitative content analysis and thematic analysis. *Journal of Nursing Education and Practice* 6(5):100-110
- Ahmadu et al. (2024) Association between caregiver quality of life and the care provided to adults with severe visual impairments in rare disease: A systematic review, *ISPOR EU 2024*