

The Impact of LHON on the Quality of Life of Patients and their Relatives

Benson S. Chen, MD^{1,2}, Erik Holzinger, MBA³, Magali Taiel, MD⁴, Patrick Yu-Wai-Man, MD PhD^{1,2,5,6}



¹Department of Clinical Neurosciences, University of Cambridge, Cambridge, United Kingdom; ²Cambridge Eye Unit, Addenbrooke's Hospital, Cambridge, United Kingdom; ³groupH, London, United Kingdom; ⁴GenSight Biologics, Paris, France; ⁵Moorfields Eye Hospital NHS Foundation Trust, London, United Kingdom; and ⁶Institute of Ophthalmology, University College London, London, United Kingdom.

Introduction

- The impact of Leber hereditary optic neuropathy (LHON) on interpersonal relationships, work, and finances of affected individuals has not been explored in detail¹
- Previous studies have focused on the emotional impact of LHON and on vision-related activities^{2,3}
- The impact of LHON on the relatives of patients is unknown and merits further study

The aim of this qualitative study was to explore the impact of LHON on patients' and their relatives' lives at the time of diagnosis and now, in four countries

Methods

- Eight focus group discussions conducted with individuals harbouring the m.11778G>A LHON mutation and their relatives in four countries [France, Germany, UK and USA]
- Separate focus groups for affected individuals and their relatives, conducted by an independent moderator using an interview guide
- Secondary analysis of the focus group findings conducted independently by two authors (BC and PYWM) using a pre-specified analytical framework organised into categories

LHON strongly affects quality of life for both patients and their relatives

LHON limits patient autonomy

- **Activities of daily living**: Total assistance required at onset. Adaptation and adjustment leading to partial assistance for some tasks only
- **Emotional**: Strong negative response at diagnosis; often followed by episodes of anxiety and frustration that can occur daily
- **Friendships**: Increased dependency on friends when socialising, sometimes with loss of friendships. Challenging interactions with strangers due to invisible disability
- **Relationships**: Closer relationship with family, tending to turn more practical than sentimental; sometimes generating tension
- **Work**: Adjustments to work routine, often with reduced hours and limited prospects for progression, creating uncertainty
- **Finances**: Reduced work-related income for some. Low-vision aids and idebenone costly and not universally subsidised. Inequitable access to disability benefit depending on country.

- All patients stressed they felt locked in a world apart, often described as gloomy, shapeless, and poorly coloured if at all
- Vision loss makes identification of people, objects and situations very complicated, requiring permanent attention and vigilance, often leading to mental fatigue



"Like it's always very bad weather – grey and few colours."

LHON places burdens on relatives

- **Activities of daily living**: Dedicate more vigilance, time, and efforts particularly for transportation and household chores leading to physical and mental fatigue
- **Emotional**: Stress arising from worry for the patient, especially the psychological impact of illness and uncertainty about the future including patient's ability to live independently
- **Relationships**: Closer relationship with patient sometimes at expense of others; maternal guilt
- **Work**: Most experienced no impact; some had to adjust their work schedule or stop working
- **Finances**: No direct impact on income, except those who adjusted their work schedule to support patient. Additional costs having to accompany patient to appointments

Focus Group Participants



17 individuals with LHON:

- Sex: 13 men and 4 women
- Current age (median[IQR]): 34 [24-56] years
- Age at diagnosis (median[IQR]): 28 [19-40] years
- Family history of LHON: (n [%]): 10 [58.8%]
- Country of residence: France (4); Germany (4); UK (5); USA (4)

17 relatives:

- 10 mothers (asymptomatic carriers) and 1 father
- Three wives and three husbands
- Country of residence: France (4); Germany (5); UK (4); USA (4)

Conclusion

- The impact of LHON extends beyond vision-related activity limitations
- Addressing the psychosocial impact of LHON and helping patients and their relatives adapt and cope with vision loss are vital

References

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The focus group interviews were conducted independently by groupH, a consulting and market research company, with funding from GenSight Biologics. No funding or inducements were received for the independently conducted secondary analysis of interview data.