LHON: a qualitative study on the experiences and hopes of patients and caregivers

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BACKGROUND

- LHON (Leber’s Hereditary Optic Neuropathy) is a rare orphan mitochondrial disease, maternally inherited, with an estimated prevalence 1:30,400-400,000, and predominantly affecting males (80%), typically from 15 to 35 years of age.
- In 70% of cases, LHON is due to mutations in one or more of the three mtDNA genes which encode proteins of the mitochondrial respiratory chain Complex I (COX). Disruption of the mitochondrial respiratory chain Complex I is the direct cause of apoptosis of retinal ganglion cells and atrophy of the optic nerve.
- The A11778G mutation is responsible for the majority of cases (70%). All mutations have incomplete penetrance.
- Vision loss manifests with acute to sub-acute decline in one eye, with rapid loss to near in days to weeks. Only 15-20% of patients have their 2nd eye clinically affected by LHON within 36 weeks of onset.
- Visual prognosis is very poor: between 75 and 98% of patients are reported to have final visual acuity of worse than 20/200, qualifying for legal blindness.
- There is currently no approved treatment and no cure for LHON.

PURPOSE

- Perceptions of affected patients with the A11778G mutation and caregivers about the diagnostic process, the consequences of LHON on their respective lives and expectations about future treatments have not yet been surveyed.
- The aim of the present study was to gain a deeper qualitative understanding around these themes.

METHODOLOGY

- A face-to-face exploratory qualitative focus group took place in November 2014 in 4 countries - the USA, the UK, Germany and France.
- 1 separate group with LHON patients and 1 separate group with caregivers were set up in each country, all audio and video recorded for further analysis.
- 17 LHON patients with the A11778G mutation in total were involved; 13 males and 4 females, aged between 18 and 57 years old.
- 17 caregivers whose relatives (husbands) have LHON participated; 9 mothers, 3 wives, 2 husbands and 1 father.
- Each group was conducted in a viewing facility and led by an experienced, independent moderator to guarantee anonymity and confidentiality.
- A semi-structured discussion guide was used and the collected data was analyzed to determine key qualitative trends.

CONCLUSIONS

- The findings highlight that disease awareness among physicians and referral pathways require improvements to lead to the right diagnosis more quickly.
- LHON strongly impacts patients and caregivers’ lives as it affects activities of daily living, emotional functioning, relationships, studies, work, recreation and finances.
- Increasing support to patients and caregivers would likely help both groups improve their quality of life.
- There is an urgent need for an effective drug improving LHON related visual failure to decrease dependency of patients and burden of caregivers.

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- We thank the patients and their relatives who participated in this study, as well as the Modo2G, neuro-ophtalmologists, and patient organizations who helped us to set up this project.

REFERENCES

- Caregivers typically describe relatives or partners providing part of full time support to a patient. We acknowledge that many caregivers typically fear for the family’s and caregivers’ autonomy and the possible role in, or contribution to, the LHON disease progression.

KEY FINDINGS

LENGTHY, WORRISOME, DIFFICULT JOURNEY BEFORE BEING DIAGNOSED

- Patients first noticed a relatively quick, but progressive, blurred vision for one eye that they usually related to fatigue, worsening of eye deficiency or ageing; including some patients aware of vision deterioration.
- When their vision did not improve, even deteriorated, and started affecting daily activities, patients worried more and usually first consulted an ophthalmologist (USA, UK) or a community-based ophthalmologist (France, Germany).
- After some visual tests, most patients were told the cause was unrelated, unclear or related to their lifestyle (e.g. smoking) - one was even misdiagnosed with a retinal-related condition.
- Others were referred to a hospital-based ophthalmologist for further investigation, a few to a neurologist, leading for one patient to diagnosis of multiple sclerosis.
- Patients consulted between 2 and 7 different doctors, often on their own initiative or pushed by relatives, before being finally referred to a LHON specialist, mainly when the 2nd eye became affected.
- After genetic testing, the LHON diagnosis was announced, devastating patients and caregivers who additionally learned that preventing visual loss progression or restoring vision was currently impossible.

Typical diagnosis journey reported by LHON patients

- All interviewed patients, including those recently diagnosed with some retained central vision, stress they feel locked in a worse condition that is described as gloomy, hopeless, painful, and frightening.
- This vision loss makes identification of people, objects and emotions very complicated for them, requiring permanent attention and vigilance, often leading to a mental stress.

LHON STRONGLY AFFECTS QUALITY OF LIFE FOR BOTH PATIENTS AND CAREGivers

- Several dimensions of the LHON patient’s quality of life are negatively influenced by the condition, which generates dependency on others, including for patients who underwent rehabilitation and use visual aids.

ACTIVITIES OF DAILY LIVING

- Patients of working age mention added costs for disabled people, reduced working hours and limited access to certain occupations. Early retirement or partial compensation in the form of disability pension for a long retirement time is often necessary.

STUDIES

- LHON patients, still at work or retired at the time of diagnosis, say they have experienced complications, social with their family, work, health, fear of losing of their job or becoming a burden to others.

EMOTIONAL FUNCTIONING

- Some patients suffer from a lack of will to live due to the vision loss, impact on family and societal change due to the reduced mobility, partner and generate depression. They feel isolated and have to face new challenges when managing their daily lives.

RELATIONSHIPS WITH OTHERS

- Friends are seen less often, or less, due to self-imposed isolation or patients’ dependency on caregivers who often assume the role of a nurse. Patients are often dependent on one caregiver, finding a partner or difficult to stay with one caregiver.

RELATIONSHIPS WITH FAMILY

- Most of patients meet more often with friends due to the vision loss, impact on family and societal change due to the reduced mobility, partner and generate depression. They feel isolated and have to face new challenges when managing their daily lives.

RECREATION

- Discrimination of most previous recreational activities such as sport, visiting friends, or activities in nature, such as reading books, watching TV, playing computer games or traveling in foreign countries.

FINANCES

- Most of patients have had a direct impact on their financial situation due to LHON, except those who stopped working or gained a job that their family has paid for.

Recreations

- Several caregivers stress they have to spend much time caring for their family members, especially with their daily activities that treated ones are not able to do, such as taking care of the elders or reading a book.

EXPECTATIONS TOWARDS TREATMENTS RELATE TO THE RESTORATION OF SOME AUTONOMY

- All patients and caregivers first wish for a cure. If that is not attainable, then a therapy should improve the vision enough to enable a minimal level of autonomy for patients, as well as less burden for caregivers.
- Enabling daily living activities, facilitating relationships with others in general and relatives in particular, plus being able to go back to previously abandoned recreational activities are the most expected benefits.
- A possible breakthrough in the future, which would correspond to their dream of a restored autonomy.

- Some recently diagnosed patients also wish that a new drug should hasten the progression of the vision loss or, even better, restore their initial vision if the medication is taken on time.
- Overall, interviewed respondents admit that just going from ‘unable to do’ to ‘can do with difficulty’ would be a big enough breakthrough for them to try out novel treatments.

LHON GENERATES BURDEN FOR CAREGIVERS

- Caregivers are strongly (in)involved in the LHON’s patient’s life and all had to fit their personal activities around the needs of the patient, sometimes, to the point of completely sacrificing such activities to be able to support their loved one.

Activities of daily living

- Patients face a daily burden to maintain their family’s autonomy and to face their job or career, as well as the financial stress due to the diagnosis. Moreover, they face the physical and psychological stress of taking care of their loved one.

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Expectations towards treatments relate to the restoration of some autonomy

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- Patients, 42 y/o: “It’s too hard keeping up now in pain of your face and in your head.”
- Patients, 56 y/o: “I’d like being able to wash your head.”

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