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Leber hereditary optic neuropathy in Czechia and Slovakia: Quality of life and costs from patient perspective

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Introduction

Leber hereditary optic neuropathy (LHON) represents the most prevalent mitochondrial rare disease, manifesting as dyschromatopsia and progressive central vision loss. The majority of patients are young adults with the peak of onset in the second and third decades of life. Males are five times more affected than females (1-3). In Czechia, over 90 individuals with one of three prevalent mutations were diagnosed between 1992 and 2016 at the Institute of Inherited Metabolic Disorders of the First Faculty of Medicine, Charles University and General University Hospital, Prague, of which 20 patients were symptomatic (4). In Slovakia, 13 symptomatic patients were diagnosed with LHON at the Department of Paediatric Ophthalmology of the Faculty of Medicine, Comenius University Bratislava, and the National Institute of Children's Diseases since 2005.

Objective

The objective of this study was to quantify the direct non-medical and indirect costs incurred by LHON patients and their informal caregivers in Czechia and Slovakia, and to assess their quality of life.

Methods

- The study cohort comprised 27 adults and children with LHON from Czechia and Slovakia, who agreed to participate
- The following data were collected: age, education, family size, onset of LHON, severity of LHON, non-medical direct and indirect costs of LHON. Direct non-medical costs included out-of-pocket payments for transportation, education, informal care (adult patients), visual aids and modifications of domestic environment.
- Indirect costs (productivity loss) were calculated as absenteeism and presenteeism according to the WHO's Heath and Work Performance Questionnaire employing human capital approach. To assess quality of life, National Eye Institute 25-item Visual Function Questionnaire (VFQ-25, version 2000) for adults and Paediatric Eye questionnaire (PedEyeQ, version 2019) for children and their parents were used.

Results

1. Epidemiology

In this study of 27 patients with LHON, 21 were adults (average age 36.1 years) and six were children (average age 13.8 years). The most reported level of visual impairment was "some useful peripheral vision," followed by "good peripheral vision" and "moderate peripheral vision loss." A majority of participants (85%) received idebenone treatment, including both adults and children (Table 1).

Table 1 Demographics

	Adults	Children
No of individuals	21	6
Sex, n (%)		
Male	20 (95%)	6 (100%)
Female	1 (5%)	0 (0%)
Age, Years		
Mean (SD)	36.1 (13.1)	13.8 (1.83)
Range	18-70	11-16
Duration of LHON from onset, Years		
Mean (SD)	8.4 (6.0)*	3.3 (2.7)
Range	2-28	0.6-7
Severity of visual impairment, n (%)		
Some useful peripheral vision	7 (33.3%)	2 (33.3%)
Good peripheral vision	4 (19%)	1 (16.7%)
Light perception only (or shadows only)	2 (9.5%)	0 (0%)
I still have good overall vision	1 (4.8%)	0 (0%)
Good central vision	1 (4.8%)	0 (0%)
Moderate peripheral vision loss	1 (4.8%)	2 (33.3%)
Some useful central vision	1 (4.8%)	1 (16.7%)
Other	4 (19%)	0 (0%)
Treatment with idebenone, n (%)	17 (80.9%)	6 (100%)

* Duration of LHON was available only from 18 adult patients.

2. Loss of productivity costs and Non-medical direct costs

- Thirty-eight percent of adult patients were on disability pension, 14% worked part-time due to LHON, and only 24% held full-time jobs (Table 2).

Table 2 Frequency of employment status of adult patients and paediatric patients´ parents

Employment status	Adult patients	Children's parents
Disability pension	38%	9%
Full time work	24%	91%
Part time work	14%	0%
Student	19%	0%
Unemployed	5%	0%

- Employed adults missed 6% of their work time (about 97.8 hours per year), while parents of children missed 29% (around 264.5 hours annually).
- The cost of absenteeism was EUR 1,003 per adult employee per year and EUR 2,711 per parent. Total productivity loss due to both absenteeism and presenteeism was estimated at EUR 9,840 per adult and EUR 6,298 per parent annually (Table 3).

Table 3 Mean absenteeism and presenteeism (in %)

Adult patients	Mean	Median	Min	Max	Total costs per person/year
Relative absenteeism	6	0	-5	25	EUR 1,003
Relative presenteeism	56	50	25	100	EUR 8,659
Combination relative absenteeism and presenteeism	50	48	25	85	EUR 9,840
Informal care (missed hours per day)	1.83h	1.5h	0	5	EUR 4,502
Children's parents					
Relative absenteeism	29	25	-5	60	EUR 2,711
Relative presenteeism	82	90	60	100	EUR 3,542
Combination relative absenteeism and presenteeism	68	57	23	120	EUR 6,298

Note: The negative value represents extra hours worked by patients.

- 52.4% adult patients needed help with daily activities, mostly provided by relatives, who contributed an average of 1.83 hours daily. Informal care costs averaged EUR 4,502 per person/year. Most patients (62.9%) travelled over two hours to see specialists, with an average round-trip cost of EUR 41. Visual aids and home modifications cost about EUR 1,153 per person/year, while rehabilitation costs for those needing it averaged EUR 1,099.

3. Quality of life

The average VFQ-25 score for adult LHON patients was 43.47, with near vision scoring lowest (28.92). In the PedEyeQ for children, the functional vision domain averaged 37.5, social domain 61.25, and frustration/worry domain 52.5. The impact on parent and family domain scored 62.5 (Table 4).

Table 4 Mean score of VFQ-25 scale and PedEyeQ (Score 0 is the worst and 100 is the best)

Adult patients	Mean	SD	Median	Min	Max
Total VFQ25_score	*43.47*	*15.86*	*38.75*	*23.89*	*81.04*
General health	44.12	22.59	50.00	0.00	75.00
General vision	34.12	15.43	40.00	20.00	60.00
Ocular pain	64.71	21.76	62.50	37.50	100.00
Near activities	28.92	21.27	25.00	0.00	75.00
Distance activities	37.25	21.06	41.67	0.00	83.33
Social functioning	42.65	27.97	37.50	0.00	100.00
Mental health	50.74	24.40	50.00	0.00	93.75
Role difficulties	44.12	32.51	25.00	0.00	100.00
Dependency	52.94	27.63	50.00	8.33	100.00
Driving	87.50	5.89	87.50	83.33	91.67
Colour vision	61.76	20.00	50.00	25.00	100.00
Peripheral vision	50.00	21.65	50.00	25.00	75.00
Children and their parents					
Functional vision domain	37.50	19.36	42.50	10.00	55.00
Social domain	61.25	13.15	60.00	50.00	75.00
Frustration/worry domain	52.50	19.36	52.50	30.00	75.00
Impact on parent and family domain	62.50	23.98	60.00	40.00	90.00

CONCLUSIONS

This study highlights the significant socioeconomic challenges faced by LHON patients and their families. Early, timely access to diagnosis, treatment, financial support, and psychological counselling - ideally within treatment centers - can help patients and families better manage the effects of vision loss and adjust to living with this rare condition.

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