Low Vision Aids in Pediatric Leber Hereditary Optic Neuropathy: Experience of a Tertiary Center

Auxiliares de Baixa Visão na Neuropatia Óptica Hereditária de Leber Pediátrica: Experiência de um Centro Terciário

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ABSTRACT

INTRODUCTION: Leber hereditary optic neuropathy (LHON) is the most common inherited mitochondrial disorder. It typically affects young males, but its onset in childhood is rare. Treatment is limited and include the use of idebenone. Furthermore, to improve visual performance and academic and social adaptation, low vision aids are crucial. We report our experience with pediatric LHON and the low vision aids we recommend for these patients.

METHODS: Retrospective study of a Portuguese pediatric LHON cohort with confirmed pathogenic mitochondrial mutation, followed in our Pediatric Ophthalmology and Low Vision Center.

RESULTS: We enrolled 3 patients, all male, of 8, 14 and 15 years old. Vision loss and diagnosis occurred at 5, 10 and 14 years old, respectively. The variants *m.13094T>C* (patient 1, Leber plus) and *m.3460G>A* (patients 2 and 3, Leber) were present. Best-corrected visual acuity (BCVA) was: 20/20 in the right eye (OD), 20/30 in the left eye (OS), with subjective cecocentral and peripheral visual field loss (patient 1), 20/100 OD, 20/200 OS, with cecocentral and diffuse defects on perimetry (patient 2) and 20/400 in both eyes (OU) with severe visual field constriction and central islands of vision sparing (patient 3).

Neuroimaging, multimodal ophthalmic imaging and blood analysis were performed before definitive diagnosis. Since diagnosis, all the children started idebenone and were evaluated by the low vision multidisciplinary team (ophthalmologists, pediatricians, physiatrists, geneticists, psychologists, orthoptic technicians, social workers and special education teachers) along with the low vision team to promote concomitant visual rehabilitation.

Patient 1 only requires individual school support. Patient 2 uses a monocular, focusable, hand-held Keplerian telescope for far with improvement of the BCVA to 20/20 OS and video magnifier systems with good adaptation and reading speed improvement. Patient 3 also benefits from video magnifiers and a hand-held magnifier for near, with improved visual tasks.

CONCLUSION: Along with medical treatment and follow-up, the approach to pediatric LHON must include low vision rehabilitation. As shown here, the availability of a wide range of aids for near and far vision greatly improves visual performance and allows for greater participation in school and social activities, facilitating the healthy development of these children.

KEYWORDS: Child; Optic Atrophy, Hereditary, Leber/therapy; Optical Devices; Visual Acuity.

RESUMO

INTRODUÇÃO: A neuropatia óptica hereditária de Leber (LHON) é a doença mitocondrial hereditária mais comum. Embora afete tipicamente jovens do sexo masculino, o seu aparecimento na infância é raro. O tratamento é limitado, e inclui o uso de idebenona. Além disso, para melhorar o desempenho visual e a adaptação escolar e social, os auxiliares de baixa visão são essenciais. O presente artigo relata a nossa experiência com LHON pediátrica e apresenta as ajudas visuais que promovemos para estes doentes.

MÉTODOS: Estudo retrospetivo de uma coorte pediátrica portuguesa com LHON confirmada por mutação patogénica, seguida no nosso Centro de Oftalmologia Pediátrica e Baixa Visão.

RESULTADOS: Foram incluídos 3 pacientes do sexo masculino, com idades de 8, 14 e 15 anos. A perda de visão e o diagnóstico ocorreram aos 5, 10 e 14 anos, respetivamente. As variantes *m.13094T>C* (doente 1, Leber *Plus*) e *m.3460G>A* (doentes 2 e 3, Leber) foram identificadas. A melhor acuidade visual corrigida (BCVA) foi: 20/20 no olho direito (OD), 20/30 no olho esquerdo (OE), com perda subjetiva de campo visual cecocentral e periférica (doente 1), 20/100 OD, 20/200 OE, com defeitos cecocentrais e difusos na perimetria (doente 2) e 20/400 em ambos os olhos (ODE) com constrição severa do campo visual e ilhas centrais de visão poupada (doente 3).

Neuroimagem, imagiologia oftalmológica multimodal e análises sanguíneas foram realizadas antes do diagnóstico definitivo. Desde o diagnóstico todas as crianças iniciaram idebenona e foram avaliadas por uma equipa multidisciplinar de baixa visão para promover a reabilitação visual.

O doente 1 necessita apenas de apoio escolar individualizado. O doente 2 usa um telescópio monocular focável de Kepler para longe, melhorando a BCVA para 20/20 OE, e sistemas de vídeomagnificadores com boa adaptação e melhoria da velocidade de leitura. O doente 3 também beneficia de vídeomagnificadores e de uma lupa manual para perto, com melhoria nas tarefas visuais.

CONCLUSÃO: Além do tratamento médico e follow-up regular, a abordagem à LHON pediátrica deve incluir reabilitação de baixa visão. A disponibilidade de uma ampla gama de ajudas para perto e para longe melhorou significativamente o desempenho visual, permitindo uma maior participação nas atividades escolares e sociais, promovendo o desenvolvimento saudável destas crianças.

PALAVRAS-CHAVE: Acuidade Visual; Atrofia Óptica Hereditária de Leber/tratamento; Criança; Dispositivos Ópticos.

INTRODUCTION

Leber hereditary optic neuropathy (LHON) is the most common mitochondrial disorder causing bilateral optic neuropathy, typically affecting young males between 15 and 35 years. It results from point mutations in mitochondrial DNA, most commonly *m.11778G>A*, *m.3460G>A*, and *m.14484T>C*, which impair complex I of the respiratory chain and lead to selective degeneration of retinal ganglion cells.[1-4] Patients usually present with acute, painless central vision loss that may rapidly progress to legal blindness, though spontaneous partial recovery can occur, especially within the first year.[2-7] The disease follows maternal inheritance and is influenced by genetic and environmental modifiers such as smoking and alcohol.[5] Despite advances in research, including idebenone and gene therapy, no curative treatment is currently available.[1,5]

Given the limited treatment options and the typically irreversible nature of vision loss, low vision rehabilitation plays a central role in managing LHON. In particular, low vision aids (LVAs) are crucial in helping patients remain functionally independent and socially integrated. They aim to maximize residual vision by enhancing reading ability, distance viewing, or performing daily activities.[8]

Despite the importance of such interventions, data regarding their use in pediatric LHON remains scarce. One of the most comprehensive reports to date, by Gopalakrishnan *et al* recently reported the largest cohort study to date assessing low vision rehabilitation in LHON, analyzing data from 74 patients. Their findings confirmed that LVAs significantly improve visual function, especially for near tasks. Notably, they observed that patients under 18 years of age showed greater improvements in reading acuity and reading speed, and had distinct preferences in device type,

favoring simpler optical aids. Their study also highlighted the need for pediatric-centered rehabilitation strategies and tools to address the unique developmental and educational needs of this population.[9]

The value of LVAs in both adult and pediatric low vision populations has been emphasized in several systematic reviews.[10,11] Barker *et al*[10] conducted a Cochrane review which, although it found no high-quality randomized controlled trials in children, underscored the need for robust evidence using objective outcomes like reading speed and quality of life measures. More recently, Perrault *et al*[12] compared low vision aid use in children and adults, revealing that optical devices (e.g., magnifiers and high-add spectacles) were more frequently prescribed to children, who also showed better improvements in visual acuity and greater independence after rehabilitation. These findings support the notion that early access to individually tailored visual aids can have a substantial impact on functional vision and daily life, especially in pediatric patients.[8,9,12]

This study aims to report the visual rehabilitation experience with LVAs in pediatric patients with LHON at a tertiary care center, emphasizing functional outcomes and device selection in this specific population, given the significant impact of early-onset vision loss on independence and quality of life.

METHODS

This retrospective case series included all pediatric patients with LHON who were followed at the Pediatric Ophthalmology and Low Vision Center (LVC) in collaboration with the Metabolic Diseases and Genetics Department of Unidade Local de Saúde de Coimbra between January 2020 and September 2024.

Inclusion criteria were: (1) age under 18 years at the time of diagnosis; (2) confirmed mitochondrial mutations associated with LHON; and (3) regular follow-up by both the ophthalmology and low vision teams.

Before their referral to the LVC, each patient underwent a thorough neuro-ophthalmic evaluation by trained neuro-ophthalmologists to rule out other causes of optic neuropathy, leading to their LHON diagnosis. Multimodal ophthalmic imaging and neuroimaging were performed and genetic testing confirmed the diagnosis.

Following diagnosis, all patients began treatment with idebenone and were enrolled in a low vision rehabilitation program tailored to their specific needs. The rehabilitation was managed by a multidisciplinary team consisting of ophthalmologists, geneticists, physiotherapists, psychologists, orthoptists, and special education teachers. Visual rehabilitation involved personalized programs, such as training in keyboard use and reading skills. Visual aids were prescribed and customized to each patient based on their individual visual requirements.

Upon referral to the LVC, a detailed low vision assessment was performed for each patient. Distant visual acuity was measured using the Snellen chart and LH symbols, and near visual acuity was assessed with the MNREAD acuity chart. Visual acuity, contrast sensitivity and reading speed were recorded at the time of diagnosis and during follow-up after the use of LVAs.

Data collected included the patients' presenting complaints, specific visual needs, presenting visual acuity, final visual acuity, and the details of the prescribed low vision devices. All assessments and fittings of the LVAs were conducted by a specialized team comprising a low vision ophthalmologist and an orthoptic technician at the LVC.

Ethical approval was obtained from the Ethics Committee of Unidade Local de Saúde de Coimbra. As this was a retrospective study with anonymized data, the need for informed consent was waived in accordance with institutional policies and the Declaration of Helsinki.

RESULTS

Three pediatric patients were included in this study (Table 1 summarizes main information).

Table 1. Summary of the features of the patients and main results.

Patient	1	2	3
Sex	Male	Male	Male
Age (at diagnosis) in years	8 (5)	14 (10)	15 (14)
Variant (gene)	*m.13094T>C (MT-ND5)*	*m.3460G>A (MT-ND1)*	*m.3460G>A (MT-ND1)*
BCVA for far at diagnosis	OD: 20/100 OS: 20/500	OD: 20/250 OS: 20/800	OD:20/400 OS: 20/160
Final BCVA for far	OD: 20/20 OS: 20/32	OD: 20/100 OS: 20/20	OD: 20/640 OS: 20/400
BCVA for near in M (reading speed in WPM) at diagnosis	2.5 (10)	5 (10)	8 (unmeasurable)
Final BCVA for near in M (reading speed in WPM)	0.6 (40)	3.2 (80)	6.3 (50)
Low vision aids used	Handheld EVES Typoscope	Portable handheld EVES Keplerian telescope Desktop video magnifier	Desktop video magnifier Portable handheld EVES

BCVA, best corrected visual acuity; M, M-value; WPM, words-per-minute; EVES, electronic vision enhancement systems.

PATIENT 1

An 8-year-old male diagnosed with LHON at 5 years old. Genetic testing revealed the presence of the variant *m.13094T>C* in heteroplasmy (56%) in the *MT-ND5* gene, classified as pathogenic and associated with Leber plus syndrome. The patient had a history of Duane type 1 syndrome and developmental delay, but no relevant family history.

Fundoscopy revealed optic disc pallor in both eyes (OU), more prominently in the left eye (OS). The assessment of visual fields using computerized static perimetry was not performed in this patient due to the young age at diagnosis and the difficulty in obtaining adequate cooperation for the examination.

At the time of diagnosis, best corrected visual acuity (BCVA) was 20/100 in the right eye (OD) and 20/500 OS, with contrast sensitivity measured at 20/100 (2.5%). The patient was initially prescribed a typoscope (Fig. 1), provided with individualized school support, and introduced early to a handheld electronic vision enhancement system (EVES). Following three years of idebenone therapy combined with low vision rehabilitation, the patient exhibited significant visual improvement, with BCVA reaching 20/20 OD and 20/32 OS. Binocular distance acuity improved to 20/20, and near vision progressed from 2.5M to 0.6M without the need for low vision aids. Reading speed also increased from 10 to 40 words per minute. Given the extent of spontaneous visual recovery, the use of EVES was discontinued, and the patient now relies solely on individualized educational support.

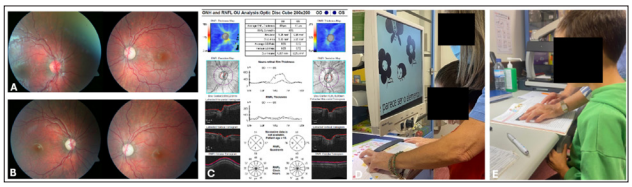

Figure 1. Patient 1: (A) Fundus photography at diagnosis showing congestive optic disc with telangiectasias OD and optic disc pallor OS. (B) Fundus photography at the last evaluation showing optic disc pallor OU. (C) Optical coherence tomography (OCT) demonstrating decreased retinal nerve fiber layer (RNFL) thickness. (D) Training in reading with the use of a desktop EVES magnifier during a low vision consultation (at one of the initial evaluations). (E) Training in reading with the use of a typoscope during a low vision consultation (at one of the final evaluations).

PATIENT 2

A 14-year-old male diagnosed with LHON at 10 years old. Genetic testing confirmed the presence of the *m.3460G>A* mutation, in homoplasmy, in the *MT-ND1* gene, classified as pathogenic. The patient had an ocular history of myopia and astigmatism, but no other relevant personal or family history.

Ophthalmologic evaluation revealed optic disc pallor at fundoscopy. Perimetry showed a centrocecal scotoma along with diffuse visual field defects (Fig. 2).

At initial evaluation, BCVA was 20/250 OD and 20/800 OS for distance, with near vision measured at 5M. Contrast sensitivity was 20/200 (5%). Following diagnosis, the patient was prescribed a portable handheld EVES for near tasks (Fig. 2), which led to a marked improvement in visual performance – near vision improved from 5M to 3.2M, and reading speed increased from 10 to 80 words per minute. Consequently, the patient transitioned to using primarily distance vision aids. For far vision, a 6×16 Keplerian monocular telescope and a portable handheld EVES adjusted for distance were provided. With the telescope, distance BCVA improved to 20/100 OD (20/200 unaided) and to 20/20 OS (20/100 unaided). These functional gains were attributed to the effective use of low vision aids rather than to spontaneous visual recovery.

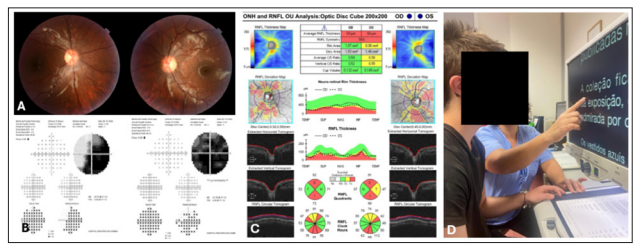

Figure 2. Patient 2: (A) Fundus photography at diagnosis showing temporal optic disc pallor OU. (B) Perimetry showing centrocecal scotoma along with diffuse visual field defects. (C) OCT demonstrating decreased RNFL thickness. (D) Training in reading with the use of a desktop EVES magnifier with inverted contrast during a low vision consultation.

PATIENT 3

A 15-year-old male diagnosed with LHON at age 14 years old. Genetic testing revealed the *m.3460G>A* mutation in the *MT-ND1* gene, classified as pathogenic. The patient had a history of accommodative endotropia and a family history of LHON, with a maternal uncle also affected by the condition. No other significant personal medical history was reported.

During the ophthalmic evaluation, fundoscopy revealed optic disc pallor with residual edema, and perimetry showed severe visual field constriction with central islands of spared vision (Fig. 3). The patient's initial BCVA was 20/400 OD and 20/160 OS. Contrast sensitivity was 20/400 (2.5%). Although distance vision further deteriorated to 20/640 OD and 20/400 OS, with binocular vision measured at 20/250, near vision improved from 8M to 6.3M.

The patient is currently using a desktop EVES magnifier for near vision tasks (Fig. 3), as well as a portable handheld EVES magnifier for both near and far tasks. The use of these aids resulted in a significant improvement in reading speed, increasing from unmeasurable to 50 WPM.

DISCUSSION

Early-onset vision loss has significant functional and psychological implications.[8] In LHON the bilateral progressive loss of central vision patients leads to reading disability at an early stage, severely affecting quality of life and in-

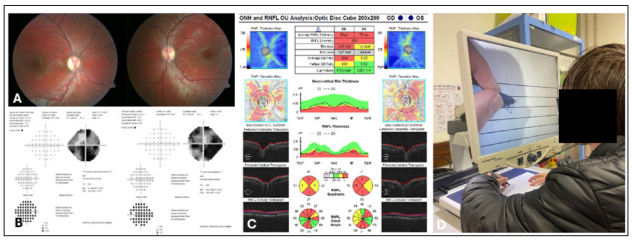

Figure 3. Patient 3: (A) Fundus photography at diagnosis showing temporal optic disc pallor with residual edema OU. (B) Perimetry severe visual field constriction with central islands of spared vision. (C) OCT demonstrating decreased RNFL thickness. (D) Training in writing with the use of a desktop EVES magnifier during a low vision consultation.

dependent living.[9] A study by Kirkman *et al* evaluated 196 LHON patients using the well-validated Visual Function Index (VF-14) questionnaire and found that LHON has a profoundly negative impact on quality of life, yielding the lowest VF-14 scores when compared to other ophthalmic disorders. Reading small print and reading a book were identified as the most challenging tasks for these patients in the VF-14 questionnaire.[13]

While promising research into potential treatments is ongoing, the primary approach for managing LHON remains supportive rather than curative. This approach primarily focuses on visual rehabilitation and the use of optical aids, which are particularly beneficial for individuals experiencing central vision loss due to LHON.[14]

There is limited literature on how LVAs improve visual acuity in LHON patients,[9,15,16] particularly in the pediatric population, where diagnosis is less frequent.[2,5,6] Despite this, finding appropriate low vision aids for these patients is crucial due to the limited treatment options for this debilitating condition.

In our cohort, two of the three pediatric patients showed improvement in distance visual acuity, as measured with LVAs, after starting low vision rehabilitation. All three patients demonstrated significant improvements in near vision and reading speed, highlighting the effectiveness of tailored low vision devices in enhancing functional vision. Notably, reading speed served as an objective outcome measure - widely recognized as a direct functional correlate of near visual acuity and reading ability. As supported by Barker *et al*,[10] it offers a reliable and quantifiable alternative to subjective quality-of-life questionnaires, especially in pediatric populations, given that reading performance has been found to be one of the best predictors of patient-reported visual ability and vision related quality of life. The most commonly used low vision devices were the desktop EVES magnifier for near vision and the portable handheld EVES for both near and far vision. Only one child required an optical aid, specifically a 6x16 Keplerian telescope.

Our findings are consistent with the study by Gopalakrishnan *et al*[9] in 2023, which retrospectively reviewed 74 LHON cases divided into three age groups (<18, 18-40, >40 years old). They found no statistically significant differences in distance visual acuity but did report significant improvements in near vision with LVAs across all age groups. The study also highlighted that different low vision aids can be recommended depending on the stage of LHON. In the early stages, when peripheral vision remains intact, monocular and binocular telescopes are useful for distance vision. As central vision loss progresses, stand magnifiers and electronic magnifiers become more relevant. In advanced stages of the disease, non-optical aids and electronic head-mounted devices may offer significant benefits. Additionally, Gopalakrishnan *et al*[9] noted that for patients under 18 years old, stand magnifiers and electronic magnifiers are the most commonly prescribed aids, while older patients tend to require a broader range of aids, including non-optical options and telescopes, to address their diverse visual needs.

Rudanko *et al*[15] reported in 1995 that, out of 20 LHON patients, 19 benefited from LVAs, despite many of them adopting eccentric fixation due to central scotomas. Given the large central scotomas that can extend 15º-40º from the central fixation point, many LHON patients need to rely on eccentric viewing, consciously fixating to the side of the object of interest.[9,16,17]

In our study, all patients showed improvements in near visual acuity after initiating visual rehabilitation with the use of LVAs. However, visual recovery also occurred in the absence of LVAs, potentially attributable to spontaneous recovery or the use of idebenone. Patient 3 experienced further visual decline, but given the short follow-up period (less than one year post-diagnosis), further observation is necessary before drawing definitive conclusions.

Although most individuals with LHON experience permanent vision loss, spontaneous recovery – particularly within the first year – is well documented and influenced by genotype.[3,7,17] Recovery is more frequent in cases with the *m.14484T>C* mutation, less so with *m.11778G>A*, and intermediate with *m.3460G>A* (present in our second and third patients).[4,5,18] The rare *m.13094T>C* mutation, seen in patient 1, has limited literature, but some reports suggest a potential for early recovery, possibly influenced by age and hormonal factors.[19] Favorable prognostic indicators include younger age at onset, subacute progression, and specific OCT features.[20-22]

Idebenone has been explored as a treatment option, with mixed results. Some retrospective studies suggest that early and prolonged use may improve visual outcomes, particularly when started within the first year.[23] However, randomized trials have not demonstrated statistically significant efficacy.[24] Despite being safe and well-tolerated, further research is required to clarify its role, especially in asymptomatic carriers or chronic cases.

The efficacy of low vision aids has also been demonstrated in hereditary retinal dystrophies such as retinitis pigmentosa (RP). In a Brazilian study of 30 RP patients, Castro *et al* found that 90% benefited from optical or electronic aids, including telescopes, hand-held magnifiers, prism spectacles, and closed circuit televisions (CCTV) systems. These interventions significantly improved both distance and near visual acuity, enabling tasks such as reading, blackboard copying in school-aged children, and enhanced mobility for adults. Patients also reported high satisfaction with the visual rehabilitation program and regained functional independence in daily activities.[25] Another study involving

RP patients showed that 59.4% achieved functional reading with near LVAs, particularly spectacles and stand magnifiers. Interestingly, while better visual acuity favored success, contrast sensitivity at low spatial frequencies was more strongly correlated with reading speed, emphasizing its role in evaluating LVA outcomes.[26] These studies illustrate that, as in pediatric LHON, individualized LVA strategies can restore functional vision and independence, particularly when accompanied by structured training and early intervention.

Despite the positive outcomes observed in this study, the small sample size is a significant limitation. A larger, longitudinal study is necessary to assess long-term compliance with LVAs and to evaluate the evolving needs of patients as their visual function changes. Additionally, assessing both quantitative and qualitative improvements in quality of life with LVAs, beyond visual acuity, would provide a more comprehensive understanding of the impact of these aids on daily activities.

While ongoing research into therapeutic interventions for LHON continues, the role of low vision aids and visual rehabilitation should not be underestimated. More studies focused on the importance of LVAs and visual rehabilitation are needed to further support their use, particularly in pediatric populations.

CONCLUSION

The results of this study highlight the importance of early low vision rehabilitation in pediatric patients with LHON. Despite the lack of curative treatments for LHON, timely intervention with appropriate LVAs can substantially improve visual performance by enhancing remaining vision, facilitating both distance and near vision improvements. As demonstrated by the cases, a multidisciplinary approach tailored to each patient's specific visual needs and social context allows for significant improvements in quality of life.

Early use of LVAs, such as telescopes, video magnifiers, and specialized reading tools, not only enhanced the children's functional vision but also enabled greater participation in academic and social settings. These children need to be able to see, and by providing early visual support, we can normalize their performance. This helps them continue their studies and personal life while also boosting their self-esteem and mental health.

This study underscores that along with medical treatment and a close follow-up, the approach to pediatric LHON must include low vision rehabilitation at an early stage.

The wide range of visual aids available can be adapted to each child's needs, allowing for effective management of vision loss and promoting healthy development and independence. Future studies with larger cohorts are needed to further explore the long-term outcomes of pediatric LHON patients undergoing low vision rehabilitation.

CONTRIBUTORSHIP STATEMENT / DECLARAÇÃO DE CONTRIBUIÇÃO

NC: Responsible for gathering data, presenting results, conducting literature searches, and writing the manuscript.

LC, SF and CP: Responsible for performing clinical evaluations of the patients.

CC, LC, SF, JM and CP: Responsible for formal analysis, review and supervision of the manuscript.

All authors approved the final version to be published.

NC: Responsável pela recolha de dados, apresentação dos resultados, pesquisa bibliográfica e redação do manuscrito.

LC, SF e CP: Responsáveis pela realização das avaliações clínicas dos doentes.

CC, LC, SF, JM e CP: Responsáveis pela análise formal, revisão e supervisão do manuscrito.

Todos os autores aprovaram a versão final a ser publicada.

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Confidencialidade dos Dados: Os autores declaram ter seguido os protocolos da sua instituição acerca da publicação dos dados de doentes.

Proteção de Pessoas e Animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pela Comissão de Ética responsável e de acordo com a Declaração de Helsínquia revista em 2024 e da Associação Médica Mundial.

Proveniência e Revisão por Pares: Não comissionado; revisão externa por pares.

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REFERENCES

1. Sundaramurthy S, SelvaKumar A, Ching J, Dharani V, Sarangapani S, Yu-Wai-Man P. Leber hereditary optic neuropathy-new insights and old challenges. Graefes Arch Clin Exp Ophthalmol. 2021;259:2461-72. doi: 10.1007/s00417-020-04993-1.
2. Liutkeviciene R, Sidaraite A, Kuliaviene L, Glebauskiene B, Jurkute N, Aluzaite-Baranauskiene L, et al. A Typical Case Presentation with Spontaneous Visual Recovery in Patient Diagnosed with Leber Hereditary Optic Neuropathy due to Rare Point Mutation in MT-ND4 Gene (m.11253T>C) and Literature Review. Medicina. 2021;57:202. doi: 10.3390/medicina57030202.
3. Fraser JA, Biousse V, Newman NJ. The neuro-ophthalmology of mitochondrial disease. Surv Ophthalmol. 2010;55:299-334. doi: 10.1016/j.survophthal.2009.10.002.
4. Mascialino B, Leinonen M, Meier T. Meta-analysis of the prevalence of Leber hereditary optic neuropathy mtDNA mutations in Europe. Eur J Ophthalmol. 2012;22:461-5. doi: 10.5301/ejo.5000055.
5. Meyerson C, Van Stavern G, McClelland C. Leber hereditary optic neuropathy: current perspectives. Clin Ophthalmol. 2015 Jun 26;9:1165-76. doi: 10.2147/OPTH.S62021.
6. Hudson G, Yu-Wai-Man P, Chinnery PF. Leber hereditary optic neuropathy. Expert Opin Med Diagn. 2008;2:789-99. doi: 10.1517/17530059.2.7.789.
7. Newman NJ. Hereditary optic neuropathies: from the mitochondria to the optic nerve. Am J Ophthalmol. 2005;140:517-23. doi: 10.1016/j.ajo.2005.03.017.
8. Kavitha V, Heralgi MM, Parkar M, Harogoppa S. Quality of life in children with low vision following use of low vision aids. Taiwan J Ophthalmol. 2019;10:203-7. doi: 10.4103/tjo.tjo_94_18.
9. Gopalakrishnan S, Bhaskaran G, Jayasankar O, Selvakumar A, Raman R. Changes in visual acuity using low vision devices in patients with Leber hereditary optic neuropathy: A retrospective study. Indian J Ophthalmol. 2023;71:402-7. doi: 10.4103/ijo.IJO_1130_22.
10. Barker L, Thomas R, Rubin G, Dahlmann-Noor A. Optical reading aids for children and young people with low vision. Cochrane Database Syst Rev. 2015;2015:CD010987. doi: 10.1002/14651858.CD010987.pub2.
11. Virgili G, Acosta R, Bentley SA, Giacomelli G, Allcock C, Evans JR. Reading aids for adults with low vision. Cochrane Database Syst Rev. 2018;4:CD003303. doi: 10.1002/14651858.CD003303.pub4.
12. Perrault MA, Lauer G, Voss S, Seitz B, Käsmann-Kellner B. Visual Impairment and Low Vision Aids-A Comparison between Children and Adults. J Pers Med. 2023;13:1608. doi: 10.3390/jpm13111608.
13. Kirkman MA, Korsten A, Leonhardt M, Dimitriadis K, De Coo IF, Klopstock T,et al. Quality of life in patients with leber hereditary optic neuropathy. Invest Ophthalmol Vis Sci. 2009;50:3112-5. doi: 10.1167/iovs.08-3166.
14. Stelmack JA, Tang XC, Reda DJ, et al. Outcomes of the Veterans Affairs Low Vision Intervention Trial (LOVIT). Arch Ophthalmol. 2008;126:608-17. doi:10.1001/archopht.126.5.608
15. Rudanko SL. Visual rehabilitation of persons with Leber's hereditary optic neuropathy. J Vis Impair Blind. 1995;89:153 6.
16. Becker SL, Nowakowski RW. Leber optic atrophy low vision case report. Am J Optom Physiol Opt. 1988;65:209 11.
17. Zaky AG, El Bayoumi B, Elghoubashy ES, Sarhan AE, Zaky MA. Eccentric viewing training for low vision rehabilitation in patients with central scotoma. Menoufia Med J. 2020;33:1186.
18. Yu-Wai-Man P, Griffiths PG, Hudson G, Chinnery PF. Inherited mitochondrial optic neuropathies. J Med Genet. 2009;46:145-58. doi: 10.1136/jmg.2007.054270.
19. Emperador S, Vidal M, Hernández-Ainsa C, Ruiz-Ruiz C, Woods D, Morales-Becerra A, et al. The Decrease in Mitochondrial DNA Mutation Load Parallels Visual Recovery in a Leber Hereditary Optic Neuropathy Patient. Front Neurosci. 2018;12:61. doi: 10.3389/fnins.2018.00061.
20. Barboni P, Savini G, Valentino ML, Montagna P, Cortelli P, De Negri AM, et al. Retinal nerve fiber layer evaluation by optical coherence tomography in Leber's hereditary optic neuropathy. Ophthalmology. 2005;112:120-6. doi: 10.1016/j.ophtha.2004.06.034.
21. Barboni P, Savini G, Valentino ML, La Morgia C, Bellusci C, De Negri AM, et al. Leber's hereditary optic neuropathy with childhood onset. Invest Ophthalmol Vis Sci. 2006;47:5303-9. doi: 10.1167/iovs.06-0520.
22. Ramos Cdo V, Bellusci C, Savini G, Carbonelli M, Berezovsky A, Tamaki C, et al. Association of optic disc size with development and prognosis of Leber's hereditary optic neuropathy. Invest Ophthalmol Vis Sci. 2009;50:1666-74. doi: 10.1167/iovs.08-2695.
23. Carelli V, La Morgia C, Valentino ML, Rizzo G, Carbonelli M, De Negri AM, et al. Idebenone treatment in Leber's hereditary optic neuropathy. Brain. 2011;134:e188. doi: 10.1093/brain/awr180.
24. Klopstock T, Yu-Wai-Man P, Dimitriadis K, Rouleau J, Heck S, Bailie M, et al. A randomized placebo-controlled trial of idebenone in Leber's hereditary optic neuropathy. Brain. 2011;134:2677-86. doi: 10.1093/brain/awr170.
25. de Castro CT, Berezovsky A, de Castro DD, Salomão SR. Reabilitação visual em pacientes com retinose pigmentária. Arq Bras Oftalmol. 2006;69:687-90. doi: 10.1590/s0004-27492006000500013.
26. Moya STF, Oliveira A, Fernandes L. Retinitis pigmentosa and low vision: functional use of near low vision aids and correlation with visual functions. Rev Bras Oftalmol. 2003;62:679–88.

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