Use of optical tools in a patient with Leber's Hereditary Optic Neuropathy: A case report

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Introduction

- To report the case of a 4-year-old patient with Leber's Hereditary Optic Neuropathy who has difficulties in the literacy process and, with the help of the low vision and visual rehabilitation team of the Hospital Oftalmológico de Brasília, showed significant improvement in visual acuity and quality of life.

Case Report

- Patient BLD, 4 years old, male, referred to the Low Vision Service of the Hospital Oftalmológico de Brasília for evaluation of pediatric low vision due to severe low visual acuity (BAV) in both eyes, both far and near; and difficulty in the literacy process. Previous pathological history: Preterm patient aged 33 weeks and birth weight of 1980 grams, 8 days of hospitalization in the neonatal ICU due to respiratory failure. At 3 months of age, the parents observed the presence of strabismus. They sought specialized care at another service in Brasília, where they started using glasses at 10 months of age.
- After assistance in conjunction with a geneticist, the hypothesis of mitochondriopathy associated with bilateral optic nerve hypotrophy was raised. Imaging: March 2018 MRI showing optic nerve narrowing at the chiasm level. Ophthalmological examination shows better fixation with the left eye and bilateral horizontal nystagmus. Uncorrected visual acuity of 20/800 in both eyes and 20/400 with eyeglasses prescribed for 18 months. After performing dynamic refraction: RE: +11.00DE – 1.50DC at 10° and LE: +11.00DE – 0.75DC at 160° showed improvement in corrected visual acuity to 20/150 in both eyes.

- Medicinal filters were tested to decrease Glare, but the patient did not report improvement. Kepler telescopic system tested, manual monocular Teleloupe with 6x magnification and presented corrected visual acuity: 20/60 both eyes. Near visual acuity: 1.2M (Metric reading table) at 25 cm with mild difficulty; and with the help of a stone-type manual magnifier, it evolved to 0.8M at 25 cm and better reading speed.
  Features Krimsky with correction: 50 DP exotropia. Unrestricted extrinsic eye movement; Anterior chamber biomicroscopy: with calm eye; transparent cornea and wide anterior chamber, formed and without reaction; photomotor reflex present. Fundus denoting normal scleral ring, pale optic nerve 2+/4+ suggesting optic nerve atrophy; macula and vessels without changes in both eyes.

Figure 1: Magnibrite Loupe

Figure 2: Monocular Teleloup

Discussion

- Leber's Hereditary Optic Neuropathy (LHON) is an optic neuropathy resulting from a point mutation of maternally inherited mitochondrial DNA. It is more common in males and the age of onset of HLON is usually between 15 and 35 years. However, visual deterioration can arise at any age. When the presentation of the disease occurs before the age of 12 years, it is called infantile NOHL. In this pathology, there is a degeneration of retinal ganglion cells, in particular those that contribute to the papillomacular bundle. The preferential involvement by the papillomacular bundle is justified by the high energy requirement of this bundle, which is dependent on good mitochondrial functioning. Thus, the presence of a central or pericentral scotoma and a papillary pallor is characteristic.

- The reported case demonstrates the importance of specialized ophthalmic follow-up in low vision and visual rehabilitation, mainly because it deals with a pediatric patient under 7 years of age and in the literacy process; since BAV significantly compromises their pedagogical training and quality of life. With this, we observed that the correct refraction and the use of the appropriate optical aid, it was possible to significantly improve the patient's visual acuity and his quality of life.