

Low vision center evaluation of twin patients with retinal dystrophy

Avaliação do Setor de Baixa Visão de pacientes gemelares com distrofia retiniana

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ABSTRACT

In this paper, we report a two identical female twin patients with retinal dystrophy in investigation. The main diagnostic hypothesis is the leber congenital amaurosis. The patients were evaluated by the Low Vision Center at the Hospital Oftalmológico de Sorocaba, São Paulo- Brazil, using optical and non-optical resources for mainly patient's socio-educational relationship improvement.

Keywords: Vision, low; Leber congenital amaurosis; Congenital retinal dystrophy

RESUMO

É relatado o caso de duas pacientes gemelares idênticas do sexo feminino portadoras de distrofia retiniana em investigação. A principal hipótese diagnóstica é a amaurose congênita de leber. Foi realizada avaliação pelo setor de visão subnormal em centro oftalmológico, com orientação de uso de recursos ópticos e não ópticos para melhoria principalmente das relações socioeducativas das pacientes.

Descritores: Baixa visão; Amaurose congênita de Leber; Distrofia retiniana congênita

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INTRODUCTION

Leber congenital amaurosis (LCA) was first described by Theodore Leber in 1869 as patients with severe vision loss soon after the first years of birth, jerk nystagmus, diminished pupillary reflex, and normal-appearance retina early in life progressing to a classic appearance of pigmentary retinosis. ⁽¹⁾ Several genes may be mutated in the LCA, also generating several phenotypes. ^(1,2) It shows recessive autosomal inheritance in most cases, although in rare cases an autosomal dominant pattern can be seen. ⁽¹⁾ Patients with GUCY2D mutations usually have a stable clinical course; those with RPE65 mutations show a period of limited improvement followed by progressive deterioration with gradual progression over several decades. Koenekoop summarized the longitudinal evaluation studies of the visual function of LCA in 90 patients, and found that gradual worsening was observed in 15%, stability in 75%, and improvement in 10% of cases. ⁽¹⁾

Generally, the clinical course begins in the early years of life, most often before one year of age. ⁽³⁾ The prevalence is around 1:33,000 born in the world ⁽⁴⁾, being 1:80,000 when considering only the United States. ^(3,4) In Brazil, LCA is present in 9% of patients with retinal dystrophy. ⁽⁵⁾ Symptoms are severe congenital visual loss in early childhood, usually before 5 years of age, nystagmus, strabismus, reduced pupillary reflexes, and markedly reduced or undetectable full-field electroretinogram. ⁽⁶⁾ Associated clinical signs and symptoms may include oculodigital sign, keratoconus, cataract, photophobia, and hyperopia in most patients. ^(1,7,8)

Retinal alterations may include normal appearance on examination, bone spicules, salt-and-pepper retina, vascular attenuation, and coloboma. ⁽¹⁾

Visual acuity in most LCA patients varies from 20/200 to light perception, but some cases are reported to have 20/50 or better acuity early in life. ⁽⁹⁾

Gene therapy has been promising, although further studies are needed to begin large-scale use in humans. ⁽¹⁰⁾

Visual impairment in childhood usually has lifelong implications. In order to achieve the highest levels of functioning, participation, and quality of life, children shall be entitled to effective rehabilitation programs. ⁽¹¹⁾

The present case report aims to describe the low vision center approach of this rare entity in two patients with typical LCA ocular alterations with salt-and-pepper retinal dystrophy, nystagmus, hyperopia, and low vision. They are being followed and guided on optical and non-optical visual aids. Extraocular manifestations are currently under investigation at the Pediatric center, with diagnostic confirmation in the retinal center.

Case report

L.R.H. (patient 1) and identical twin sister L.R.H. (patient 2), 10 years and 8 months old, born in Botucatu, SP, Brazil, and coming from Taguaí, SP, Brazil, have been followed for 5 years in this service. The children's father has the same retinal ophthalmic pattern, with low vision and nystagmus. But the mother has no ophthalmic complaints. They deny consanguinity. In the first ophthalmic appointment at age five, according to medical records, they came to the low vision center due to the mother's complaint of photophobia, esotropia, and low vision since five months of age. She also reported difficulty in distinguishing the colors perceived since they were three. The mother reports uneventful pregnancy with obstetric follow-up, but they were born by cesarean section

with gestational age of 35 weeks and two days due to premature amniorrhexis. Examination of both children showed nystagmus, photophobia, esotropia, and hyperopia.

Patient 1 visual acuity using Lea's symbols was 4/50 in the right eye and 4/40 in the left eye with correction. Static refraction was +6.50 DS -1.00 CD at 180° in both eyes, with refraction similar to the glasses in use.

Patient 2 had visual acuity measured with Lea's symbols of 4/50 in the right eye, and 4/30 in the left eye with correction. Static refraction was +7.00 DS -1.00 CD at 180°, also similar to refraction already in use.

They were guided to wear glasses, and referred to start activities at the visual rehabilitation center of the service. There they had follow up with an educator, a psychologist, and an occupational therapist. At the first appointment in the specialized retinal ambulatory at the same time, no retinal alterations were seen, but retinal dystrophy needed clarification.

As instructed in the first appointment, the children started the literacy period with the presence of class assistants for both who dictated to the children what was on the board, besides the inclined writing table (they bring their faces closer to 15 cm for writing and reading), enlarged material, they sat on the first row, the board was white written with black marker, and the colored pencils have names written on the tip for use. The mother reports good pedagogical development.

At six years of age, they returned for appointment with 5/80 Snellen visual acuity in both eyes of patient 1 and 5/60 of patient 2, measured with refraction which did not change in any patient. They presented acuity for near sight with correction from 1.0 M to 15 cm.

In the following year, the mother reports that the school suggested that each child stay in a separate class, with each one having a class assistant. She reports that learning has improved, and they were able to follow the activities well. They are using the same school material as the other children, with good reading and writing. Both patients presented visual acuity with correction of 20/200 in both eyes.

They had appointments in the low vision department every six months, with eye examination similarly described.

At 10 years of age, they came for a new appointment in the low vision department, with acuity of patient 1 with static refraction in the right eye being +7.75 DS -1.75 CD at 170° (20/100), and in the left eye +6, DS -2.25 CD at 170° (20/150). Patient 2 had refraction in the right eye of + 5.50 DS -2.50 at 180° (20/200), and in the left eye +5.75 DS -2.00 CD at 175° (20/150), and these were prescribed. values.

They walk without difficulty, can perform school activities similarly to other children, sit in the first desk in the middle row, have difficulty reading the board, but say that teacher dictates what is written to them. They no longer have class assistants. They report that classmates help them with the most difficult activities, such as those that need to differentiate colors.

The following exams were carried out:

Chromatic Tests

- Binocularly Ishihara Test: all responses that identify wrong dyschromatopsias. Patient 1 was tested with a red filter (Segment® FC-425) without improvement, and patient 2 had improvement with this filter, with 5 plaques that were previously wrong. Other filters were tested on both children without improvement.

- Panel 16 Quantitative Color Vision Test: patient 1 had only

five rights on the green color spectrum, and patient 2 had four rights on the same spectrum.

- Text reading with font 8 at a distance of 20 cm with correction:

Patient 1 - 52 words per minute

Patient 2 - 53 words per minute.

Direct and consensual photomotor pupillary reflexes were reduced, 1+/4+ and had no relative afferent pupillary defect.

Flicker electroretinogram (ERG) and reverse visual evoked potentials (VEP) and flashes of light were carried out, showing low amplitude and latency in visual responses with severely reduced ERG and altered diffuse photoreceptor function.

They are being monitored in the retina department waiting diagnostic confirmation with genetic testing.

DISCUSSION

It is estimated that 19 million children under the age of 15 have low vision (1% of the total population in this age group), of which 1.4 million have irreversible blindness.⁽¹²⁾

The activities carried out at the rehabilitation center were of visual stimulation, with pedagogical support and following the activities related to the educational growth of the patients (Figure 1). They had good neuropsychomotor development, meeting well the age-specific developmental milestones. Monitoring with visual stimulation and early detection programs for visual problems are important factors for proper visual rehabilitation.⁽¹³⁾

The definitive diagnosis of retinal dystrophies is particularly complex, as there are overlapping phenotypes with similar signs and symptoms,⁽²⁾ but with funduscopy with few findings, early onset of the disease, genetic character, esotropy, high hyperopia, usually ($\geq 5.0D$)⁽³⁾, nystagmus, greatly decreased pupillary reflex, and almost no response in the ERG, Leber Congenital Amaurosis is most likely diagnostic hypothesis.

Longitudinal studies of visual performance show that most patients with Leber congenital amaurosis remain stable, with acuity maintained over time, but there may also be progression or even improvement in some cases.⁽²⁾ In the present report, both patients had improvement over time; in the first appointments they had an acuity lower than 20/200, with better acuity in the last appointment.

Ganesh et al. provided training to low-vision school patients with optical aids such as telescopes and magnifying glasses, and non-optical devices such as lighting lamps, reading stands, writing guides, and books with enlarged fonts. They have found significant improvements in functionality, especially in academic activities such as copying from the board, reading textbooks, writing along a straight line, and activities such as locating lost objects in the classroom.⁽¹⁴⁾ The patients in the present report started to use non-optical devices since the beginning of their school life, such as inclined writing table, good lighting, and books with enlarged fonts, as well as the use of optical correction of hyperopia, achieving good grades and following the other students in relation to learning.

Reading training activities at the rehabilitation center were important for this good school performance. Boonstra et al. showed that reading training, both in patients with low vision who made use of optical aids such as magnifying glasses and for those who did only training without optical aid, improved reading



Figure 1: Patients carrying out educational activity and visual stimulation in the rehabilitation center with pedagogical follow-up

performance.⁽¹⁵⁾

When the suspicion of the disease is detected, there is also the importance of multidisciplinary assessment, with referral to the Pediatrics service to rule out possible extraocular alterations such as renal dysfunctions, and in some cases renal failure, as well as cognitive alterations and neurological symptoms such as hypotonia or ataxia.^(6,16) Therefore, the ophthalmologist shall be aware of the need to refer the child to other specialties.

Early diagnosis of low vision, multidisciplinary follow-up with visual stimulation, adequate optical correction, and the prescription of non-optical reading aids have all proved to be indispensable factors for good learning and adequate school follow-up for both patients.

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