Coexistence of Straatsma syndrome without macular extension and abnormal ellipsoid zone

Coexistência da síndrome de Straatsma sem extensão macular e zona elipsoide anormal

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ABSTRACT

Myelinated retinal nerve fibers are rare congenital anomalies that appear as gray-white patches. They may be present in a syndrome characterized by ipsilateral myelinated retinal nerve fibers, myopia and amblyopia. The author reported an ellipsoid zone defect on spectral domain optical coherence tomography in a case of Straatsma syndrome without macular extension.

RESUMO

Fibras nervosas retinais mielinizadas são anomalias congênitas raras que aparecem como manchas branco-acinzentadas. Eles podem se apresentar em uma síndrome caracterizada por fibras nervosas retinais mielinizadas ipsilaterais, miopia e ambliopia. O autor relatou um defeito na zona elipsoide na tomografia de coerência óptica de domínio espectral em um caso de síndrome de Straatsma sem extensão macular.

INTRODUCTION

Myelinated retinal nerve fibers (MRNFs) are rare congenital anomalies that appear as gray-white patches in the superficial retina. Myelinated retinal nerve fiber layers are usually unilateral; 7.7% of cases are bilateral.[1] It is usually asymptomatic and therefore it detected incidentally during an ophthalmic examination. Straatsma et al.[2] first described Straatsma syndrome in patients with myopia, amblyopia, and strabismus with unilateral MRNFs in 1979. Currently, the triad of unilateral myelinated nerve fibers, myopia and amblyopia is considered sufficient to diagnose Straatsma syndrome.[3] The author reported an ellipsoid zone (EZ) defect on spectral domain optical coherence tomography (SD-OCT) in a case of Straatsma syndrome without macular extension.

CASE REPORT

A 15-year-old girl referred to clinic for low vision in the right eye. Best corrected visual acuity (BCVA) was 20/200 and 20/20 in the right and left eyes. Cycloplegic autorefractometric measures revealed refractive error of 3.00 (3,00 at 10°) D, +2.25 (1.75 at 180°) D in the right and left eyes. Ocular motility was full in all directions of gaze. There was no relative pupillary defect (RAPD). Slit lamp examination was unremarkable in both eyes and intraocular pressure was within normal limits. Fundus examination of the right eye showed extensive myelination of the retinal nerve fiber layer. Systemic examination was normal (Figure 1).

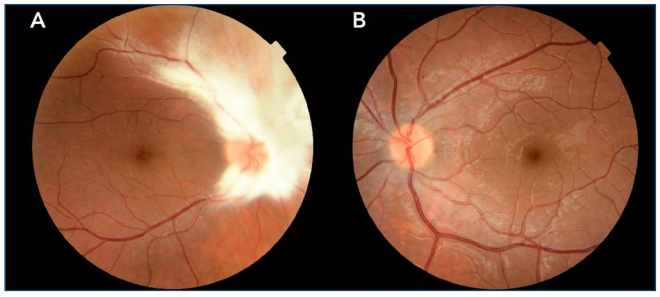

Figure 1. (A) Fundus photo of the myopic right eye showing extensive myelinated retinal nerve fibers. (B) Fundus photo of the normal left eye.

Spectral domain optical coherence tomography of the macula of the affected eye showed normal foveal contour; myelinated nerve fibers were not present in the fovea. There was an EZ and external limiting membrane (ELM) defect in SD-OCT. Left eye occlusion treatment for 4 to 5 hours a day was recommended. The patient was called for control periodically and followed up for 6 months. Although the occlusion treatment was performed regularly, no improvement in BCVA was observed compared to the first examination (Figure 2).

DISCUSSION

Myelinated retinal nerve fibers are congenital, non-progressive lesions that appear as gray-white spots on the retina. Myelinated retinal nerve fibers occur in 0.3 to 0.6% of the population.[4] It is most common in superior peripapillary region.[2]

The exact cause of nerve fiber myelination in retina is not known. Oligodendrocytes are glial cells responsible for myelination in the central nervous system. Normal myelination of the optic nerve begins in the lateral geniculate ganglion and ends in the lamina cribrosa. This is because astrocytes concentrated in the lamina cribrosa form a barrier to oligodendrocyte migration. Oligodendrocytes surrounding the MRNF were observed in histological and electron microscopic studies in patients with MRNF.[2,5]

It has been shown that MRNFs are not associated with age, gender, cardiovascular conditions other than paralysis or ocular parameters such as refraction, visual acuity, intraocular pressure or central corneal thickness.[6] Although MRNF is often seen as isolated, it can be associated with retinal vascular (telangiectasia, branch retinal artery or vein occlusion), retinal membrane (epiretinal membranes, retinal breaks), ocular developmental (coloboma, keratoconus, polycoria) and craniocephalic abnormalities, hamartoneoplastic (neurofibromatosis type 1, Gorlin's syndrome) and familiar disorders.[7] There were no ocular or systemic abnormalities in this case.

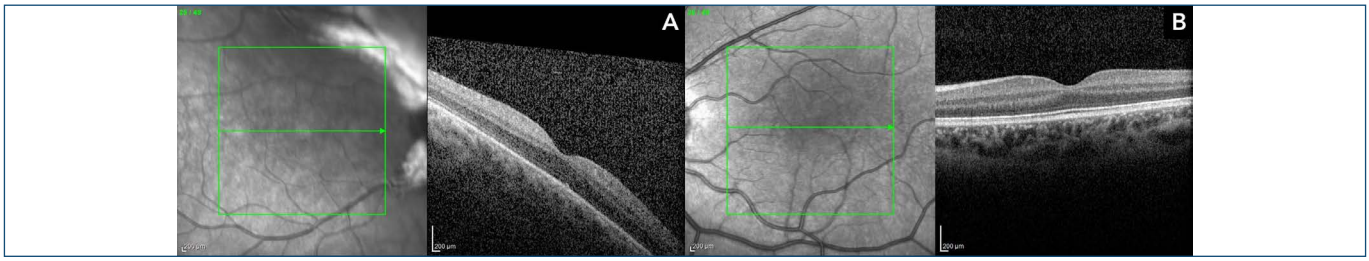

Figure 2. (A) Spectral-domain optical coherence tomography of the myopic right eye showing an ellipsoid zone and external limiting membrane defect in the macula. (B) Spectral-domain optical coherence tomography of the left eye showing a normal ellipsoid zone and external limiting membrane.

Straatsma syndrome was described initially in 1979, and it is responsible for only 0.3% to 10% of all cases of eyes with myelin fibers. It is characterized by the association of myelin fibers, myopia and amblyopia. Relative afferent pupillary defect can be seen even though it is not a common finding.[7] Some authors also describe the association of strabismus with this syndrome, which is not an indispensable condition for this syndrome. Strabismus was not observed in this case. It has been reported in the literature that the syndrome may occur with leukocoria in rare cases; therefore, it can be confused with retinoblastoma.[2] In this syndrome, organic amblyopia may occur due to anisometropic or ocular anomalies. The first step in the treatment of amblyopia patients with Straasma syndrome is full optical correction and occlusion therapy based on cycloplegic refraction. Due to structural abnormalities in the macula, visual results can be disappointing despite appropriate correction of refractive error and occlusion. There was only one case report in the literature showing the association of myelinated nerve fibers that do not include the macula and an abnormal EZ.[8]

In conclusion, macular SD-OCT evaluation of all patients presenting with Straasma syndrome can help determine the prognosis for success with visual therapy. More cases are needed showing abnormal EZ coexistence with myelinated nerve fibers that do not include the macula.

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