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Case Report

Straatsma syndrome: A case report

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Abstract

The aim of our article is to report a case of Straatsma syndrome, a rare disease characterized by characteristic triad of axial myopia, unilateral myelinated retinal nerve fibres and amblyopia. The patients will be usually asymptomatic, however few cases may present with significant visual abnormalities, commonly axial myopia and amblyopia, constituting the Straatsma syndrome. We report a case of 13 year old boy who presented to us with this rare syndrome. A 13 year old boy presented with a history of decreased vision in right eye since childhood. On examination his BCVA in right eye was 6/24, N6, while left eye it was 6/6, N6. Cycloplegic refraction revealed refractive error of -6.50DS, -1.50DC at 168° in right eye. Fundoscopy was fundamental in diagnosing this case. Dilated fundus examination of right eye revealed blurred optic disc margin and presence of myelinated retinal nerve fiber layer along superior- temporal arcade and inferio- temporal arcade, contiguous with optic nerve head, more extensive superiorly. B scan report of right eye revealed vitreous detachment. Straatsma syndrome may be included in the differential diagnosis of leucokoria and can be suspected in patients presenting with refractive errors.

Though the prognosis is poor to moderate, better results may be achieved with an early diagnosis and managing amblyopia. It can yield good results, despite high degree of anisometropia and low visual acuity.

Keywords: Straatsma syndrome, Myelinated retinal nerve fibers (MRNF), Anisometropia, Amblyopia, High myopia.

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1. Introduction

Straatsma Syndrome is a rare clinical condition outlined in 1979 by Straatsma et al., in a case series of 4 patients with unilateral myopia, amblyopia, and strabismus with myelinated retinal nerve fibers (MRNF).[1] The clinical triad of MRNF, myopia and amblyopia is now regarded as Straatsma Syndrome.[2] However, additional findings such as strabismus, nystagmus, hypoplastic optic nerve, and heterochromia iridum have also been proclaimed and do not preclude the diagnosis of this syndrome.[2-4] Though it is generally unilateral, bilateral cases of traditional and reverse Straatsma syndrome have also been reported.[2,5] Treating amblyopia pose a major challenge in managing cases of this syndrome. Several factors are reported to be associated with poor visual outcomes after occlusion therapy, including a high degree of anisometropia, strabismus, extensive myelination, and macular involvement.[3,6,7]

2. Case Report

A 13 year old boy presented to ophthalmology OPD at district hospital, Tumakuru with the chief complaint of decreased vision in right eye since childhood. Patient was advised optical correction for refractive error 3 years ago, but he is not using spectacle correction. There was no significant family history. On examination his BCVA was 6/24, N6 in right eye and 6/6, N6 in left eye. Cover-uncover and alternate cover tests were normal for both distant fixation and near fixation. The extraocular movements were normal in all cardinal gazes. Cycloplegic refraction showed refractive error of -6.50DS, -1.50DC at 168° in right eye. Pupillary reflexes were normal in both eyes. Anterior segment evaluation was done through slit lamp examination which was normal in both eyes. IOP was recorded using rebound tonometer and right eye recorded 13 mm Hg and left eye 11 mmHg. Dilated fundus examination of right eye revealed

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blurred optic disc margin and presence of myelinated retinal nerve fibre layer along the superior-temporal arcade and inferio- temporal arcade contiguous with optic nerve head, more extensive superiorly. Macula was spared with normal foveal reflex. Dilated left eye fundal examination was normal. Axial length of right eye was 23.86 mm while left eye axial length was 22.38mm. USG B scan report of right eye showed vitreous detachment, (**Figure 3**) while left showed no abnormality. MRI brain of the patient was done, which was normal.

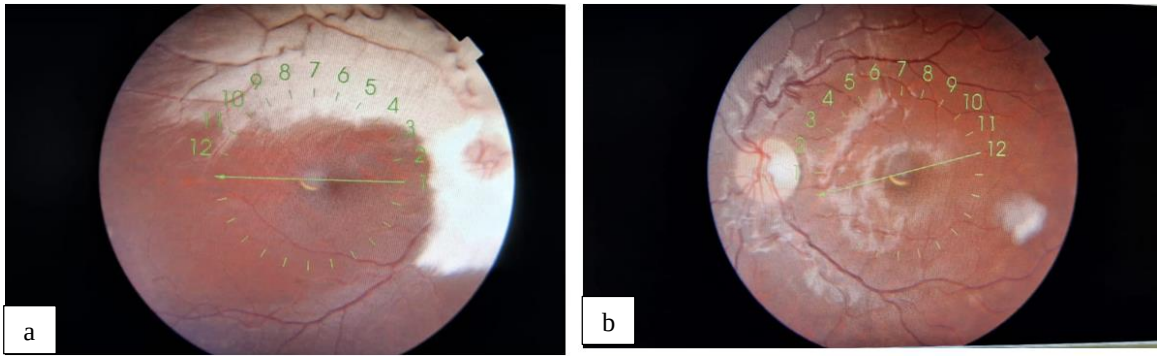

Figure 1: a): Photography of right eye showing extensive MRNF along the superior and inferior temporal arcade; **b):** Fundus photography of the left eye within normal limits

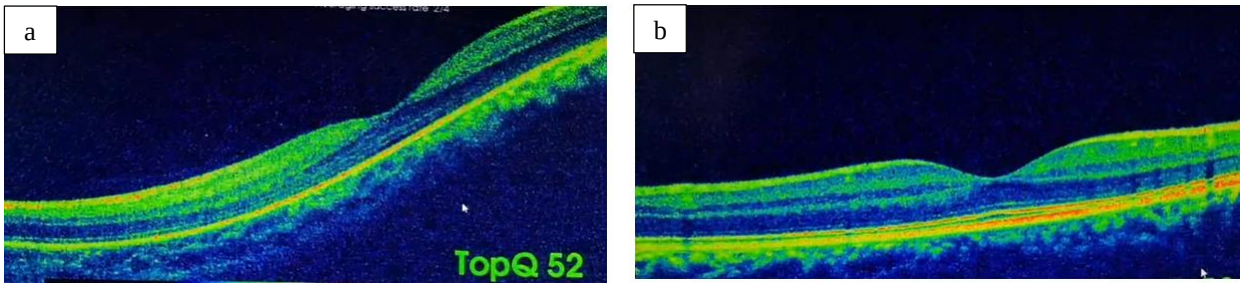

Figure 2: a): SD-OCT scan of right eye centered at fovea within normal limits showing normal foveal contour; **b**): SD- OCT scan of left eye centered at fovea within normal limits showing normal foveal contour

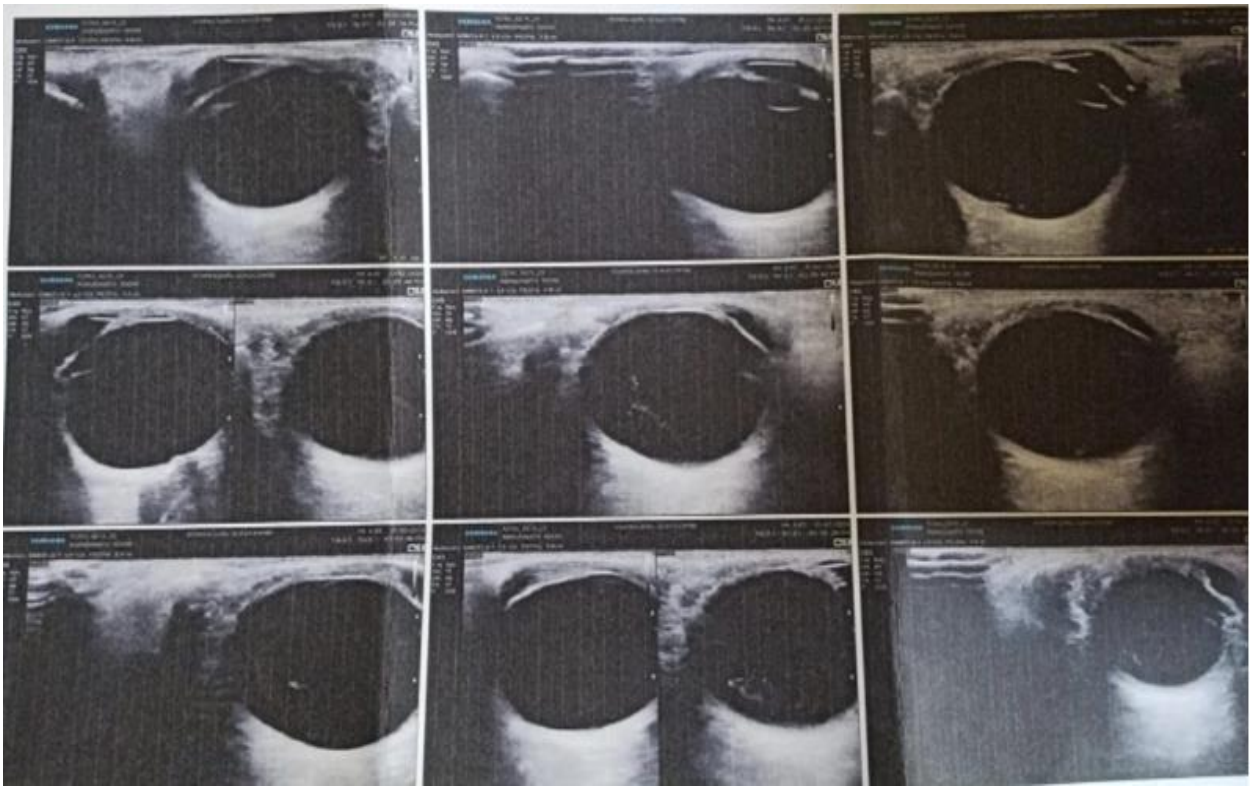

Figure 3: USG B scan showing vitreous detachment of right eye

3. Discussion

Myelinated retinal nerve fibers (MRNF) are considered as rare anomalies that appear as grey-white opaque lesions with feathery edges in retina that conceal the retinal details. They may be located on the disc, more often at temporal inferior, followed by temporal superior sector of the optic nerve head. Though majority of cases are congenital, there are few reports of acquired cases as well.[8] Myelinated retinal nerve fibers are present in about 1% of patients and were first reported by Virchow.[3,9] A myelin sheath is normally present posterior to the lamina cribrosa. In contrast, MRNF appears anterior to the lamina cribrosa, as a white striated patch with feathery borders in the peripapillary area.[10] MRNF is usually a benign condition, but it may affect visual function variably based on the location and extension of myelination plaques and macular involvement.

For this abnormality is still not fully understood and a little is known about the pathogenesis of this clinical entity. One of the theories mention that it may be related to the failure of the lamina cribosa in stopping the anterograde optic nerve myelination, while few others attribute it to the presence of heterotopic oligodendrocytes- like cells as the primus movil.[11] It is assumed to be an imbalance between the formation of the lamina cribrosa and myelination from the lateral geniculate body.[7]

Oligodendrocytes play a vital role in myelination of axons of ganglion cells, and it is postulated that astrocyte dysfunction promotes the migration of these cells through the lamina cribrosa, potentially contributing to the process. Myopia has been observed in 35% to 58% of individuals with myelination of fibers in prior investigations, with 83% exhibiting refractive errors up to 6 diopters.[12]

One theory regarding this correlation suggests that a blurred image on the retina during a critical period of ocular development may lead to visual deprivation, potentially prompting axial elongation of the eye and onset of myopia. This, in turn, could delay the maturation of the lamina cribrosa, thus facilitating the extension of fiber myelination across the retina.[566] Optic nerve myelination originates from the lamina cribrosa, which serves as a protective barrier at term. Abnormalities in the lamina cribrosa or the presence of ectopic oligodendrocyte progenitor cells can result in myelinated retinal nerve fiber layer. In our case, myopia was evident. In the right eye, with a best-corrected visual acuity (BCVA) of 6/24 N6, while the left eye exhibited visual acuity of 6/6, N6. Cycloplegic refraction revealed a refractive error of -6.50DS, -1.50DC at 168° in right eye.

There are three types of MRNF, based on their location. Type 1 MRNF is situated along the superior-temporal arcade with continuity with the optic disc; in type 2, MRNF exists along both the arcades with continuity with the optic disc; Type 3 MRNF lacks any connection to the optic disc. Type 1 of MRNF is the most frequently encountered, while type 2 carries worse prognosis in terms of visual outcomes.[7,10,12] Our case was categorized as type 2 MRNF. In a study involving 3698 autopsies, Straatsma et al. discovered MRNF in 39 cases, with 4 of them had ipsilateral myopia, amblyopia, or strabismus.[12] Earlier studies have shown the association of myopia ranging from 35% to 58% among patients with myelination of fibers, and 83% of them had refractive error upto 6 diopters.[12] It may be attributed to a blurred image on the retina during a critical stage of ocular development may lead to visual deprivation, subsequently triggering axial elongation of the eye and the development of myopia, which in turn is responsible for delayed development of the lamina cribrosa facilitating the extension of myelination of fibers through the retina.

The prevalence of MRFN was recently reported to be 0.4% among the 4439 patients enrolled in the Beijing Eye Study [15]. This demonstrated no significant association amongst factors such as age, gender, refractive error, visual acuity (VA), intraocular pressure, cataract, glaucoma, or age-related macular degeneration. However, associations with other ocular conditions such as Keratoconus or Coloboma,[13] [and systemic diseases such as neurofibromatosis, Down syndrome, and Craniofacial Dysostosis were reported.[7] Additionally, familial inheritance was also described. In this case there were no systemic associations. The type and degree of refractive error i.e., anisometropia and amblyopia are known to cause a significant effect on visual acuity of patient than the mere presence of myelinated nerve fiber.[4] In fact, scotomas in myelinated retinas are smaller than predicted by the extent of myelination, indicating that light manages to penetrate the photoreceptor layer despite the myelin interference.[3,4,12] Our case had BCVA of 6/24, N6 in right eye and 6/6, N6 in left eye. Cycloplegic refraction revealed refractive error of -6.50DS, -1.50DC at 168°. This finding is suggestive of amblyopia in right eye.

The prognosis remains uncertain in this syndrome, due to the multifactorial nature of amblyopia. There is even a documented variation of the triad involving hyperopia instead of myopia, referred syndrome to as "reverse straasma."[5]

Recently, optical coherence tomography (OCT) imaging has been conducted in these patients.[9,14] Central macular thickness is shown to be significantly thicker, while the outer ring macular thickness is significantly thinner.[13] In Stratsma Syndrome, the latter is believed to be unrelated to the degree of myopia, but rather attributed to the shadowing of the outer layers of the retina induced by the medullated nerve fibers, leading to imprecise OCT measurements.[14] Previous studies have suggested the presence of an organic cause (abnormal foveal appearance on fundoscopy with a disruption of the ellipsoid zone on OCT) as a prognostic factor for a poor response to occlusion therapy in patients with MRNF.[10,12] The SD-OCT scan centered at the fovea was found to be within normal limits for both eyes in our patient. Despite the

challenging optic nerve analysis in eyes with peripapillary MRNF, associated optic nerve involvement, namely in the form of optic nerve hypoplasia, can rarely occur.[4] In our cases, good optic nerve function was confirmed by the absence of RAPD. While the USB B-scan report of the right eye indicated vitreous detachment, the left eye B-scan was reported as normal.

3.1. Prognosis

In straasma syndrome, the visual prognosis is poor, despite a prompt occlusion therapy and full refractive correction, particularly in cases of a high degree of anisometropia, strabismus, extensive myelination, and macular involvement.[10] Treating amblyopia can pose challenges. The extent of anisometropia emerges as crucial prognostic indicator. This implies that a lower degree of anisometropia may result in good visual outcomes with an early and proper optical correction and occlusion therapy. Strabismus in Straatsma syndrome may result in poor visual prognosis in these patients.[4,6] The prognosis for this condition ranges from poor to moderate, especially if it remains undiagnosed during the critical amblyogenic period, resulting in severe amblyopia. Straatsma syndrome frequently eludes diagnosis because to its rare presentation and clinical features may be missed. Thus, the inclusion in the differential diagnosis of leukocoria is very important.[4] Furthermore, given its association with high degrees of myopia in most cases, vigilant monitoring for rhegmatogenous retinal detachment and other complications commonly associated with high myopia, such as macular hole, is imperative. This is because amblyopia may potentially mask these conditions, warranting a high level of suspicion.

3.2. Treatment

When diagnosed early in life patients should be given prompt ammetropia correction and aggressive therapy for amblyopia. A positive visual outcomes have been reported by Kee and Hwang,[7] with an early and proper management at mean age of years. It has been concluded that the refractive error, degree of anisometropia, extent of myelination, and macular appearance are the main prognostic factors. Measures aimed at detecting and enabling appropriate treatment to maximize the visual potential of such patients are crucial. Therefore, governmental programs aimed at reducing the prevalence of amblyopia are essential for fostering a competitive society in a global context. In our patient, optical correction was prescribed in the form of glasses, followed by vision therapy. Occlusion therapy was initiated in the left eye with 2 hours of patching per day. He was reviewed every 2 months for the period of 6 months. There was no improvement in the visual acuity after 6 months of amblyopia treatment.

4. Limitations of Study

This is a descriptive study which lack comparison group and limited generalizability of findings. It may be difficult to extrapolate results to larger population groups.

5. Conclusion

High degree of anisometropia, extensive myelination with macular involvement, strabismus, late presentation are considered as poor prognostic factors of Straatsma syndrome. Though it is associated with poor visual prognosis, a timely intervention with optical correction, vision therapy and occlusion therapy should always be attempted in addition to regular follow up.

6. Source of Funding

None.

7. Conflict of Interest

None.

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