CASE REPORT OLGU SUNUMU

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Macular Hole Associated with Straatsma Syndrome

Straatsma Sendromu ile Birlikte Gözlemlenen Makular Delik

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ABSTRACT Myelinated retinal nerve fibers appear as white or gray-striped patches within the distribution area of the retinal nerve fibers. They are generally asymptomatic and are often discovered incidentally during a fundoscopic examination. Straatsma syndrome is a rare condition characterized by the presence of myelinated retinal nerve fibers, axial myopia, and amblyopia. The syndrome may be accompanied by other findings such as nystagmus, strabismus, and optic nerve hypoplasia. The extent and type of myelinated retinal nerve fibers, other accompanying ocular findings, and the time of visual rehabilitation affect the visual prognosis. In this study, a rare case of Straatsma syndrome accompanied by epiretinal membrane and macular hole without any visual rehabilitation is presented.

Keywords: Amblyopia; epiretinal membrane; macular hole; myelinated retinal nerve fibers; Straatsma syndrome

ÖZET Miyelinli retina sinir lifleri, retina sinir liflerinin dağılım alanında beyaz veya gri çizgili yamalar olarak görünür. Genellikle asemptomatik olup göz dibi muayenesi sırasında tesadüfen tespit edilirler. Straatsma sendromu, miyelinli retinal sinir lifleri, aksiyel miyopi ve ambliyopinin birlikte görüldüğü nadir bir sendromdur. Sendrom, nistagmus, şaşılık ve optik sinir hipoplazisi gibi diğer bulgularla birlikte olabilir. Miyelinli retinal sinir liflerinin yaygınlığı ve tipi, eşlik eden diğer okular bulgular, görsel rehabilitasyon zamanı gibi durumlar görsel prognozu etkiler. Bu çalışmada nadir görülen, epiretinal membran ve makular delik eşlik eden ve herhangi bir görsel rehabilitasyon almayan bir Straatsma sendromu olgusu sunulmuştur.

Anahtar Kelimeler: Ambliyopi; epiretinal membran; makular delik; miyelinli retinal sinir lifleri; Straatsma sendromu

In the majority of cases, myelinated retinal nerve fibers are discovered by accident in children or adults who are otherwise healthy. In most cases, they are unilateral and do not exhibit any symptoms.[1] On the other hand, there have been reports of other abnormalities, including strabismus, nystagmus, hypoplastic optic nerve, and heterochromia of the iris.[2-4] A case series of four patients with unilateral myopia and amblyopia accompanied with myelinated retinal nerve fibers was the starting point for the initial description of Straatsma syndrome, which was published in 1979 by Straatsma et al.[5] This case report presents a patient who was diagnosed with Straatsma syndrome, which was accompanied by epiretinal membrane and macular hole.

CASE REPORT

A female patient who was 37 years old submitted an application to the ophthalmology clinic in order to get a standard eye checkup. On the other hand, she indicated that she had no other concerns and that she

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had been experiencing decreased visual acuity in her left eye since childhood. The Snellen chart indicated that the best-corrected visual acuity for the right eye was 1.0 (+0.75, -1.25x95 degrees), whereas the best-corrected visual acuity for the left eye was 0.1 (-6.00, -1.75x40 degrees). The axial length of the left eye was 26.97 millimeters, whereas the right eye measured 22.95 millimeters. In both eyes, the intraocular pressure was measured at 14 mmHg. The evaluation of the anterior portion was found to be normal in both eyes. When the fundus of the right eye was examined, there were no major findings. An epiretinal membrane, a macular hole, and a myelinated retinal nerve fiber were seen in the left eye (Figure 1). The optic disc was the origin of the nerve fiber, and it extended over the nasal hemifield and temporal arcades. A full-thickness macular hole was discovered during imaging that was carried out using spectral domain optical coherence tomography (OCT-1 Maestro, Topcon, Tokyo, Japan) (Figure 2). It was determined that the patient had Straatsma syndrome, which was accompanied with epiretinal membrane and a macular hole. For the purpose of this case study, a written informed consent form was obtained from the patient.

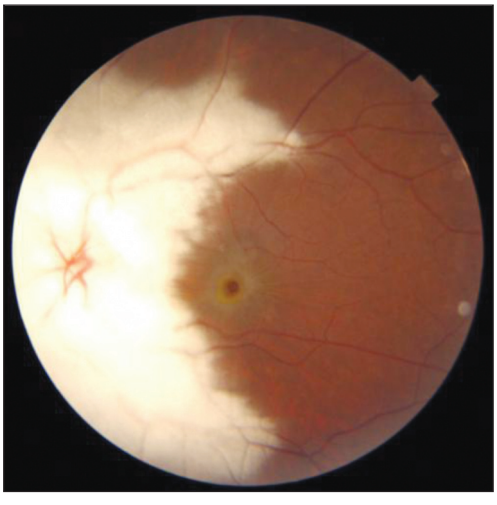

FIGURE 1: The color fundus photograph centered on the fovea of the patient's left eye.

DISCUSSION

Myelinated retinal nerve fiber is present in 0.57% to 1% of the population and can be seen bilaterally in approximately 7% of the cases.[3] Normally, myelin sheaths cover the retinal nerve fibers posterior to the lamina cribrosa. However, in cases of myelinated retinal nerve fibers, myelination appears abnormally in front of the lamina cribrosa as a feather-edged white striped patch in the peripapillary region.[5] The majority of patients who have myelinated retinal nerve fibers do not experience any symptoms, and the impact on their visual function may differ depending on the location and amount of the lesion. There are three different types of myelinated retinal nerve fibers that have been described, and they are as follows: Type 1 nerve fibers, which are the most common, are located along the superior temporal arcade; Type 2 nerve fibers, which are the rarest, are located along both temporal arcades; and Type 3 nerve fibers are discontinuous with the optic disc. A worse visual prognosis has been generally related with Type 2 myelinated retinal nerve fiber, which is one of these types of nerve fibers.[5,6] According to this classifica-

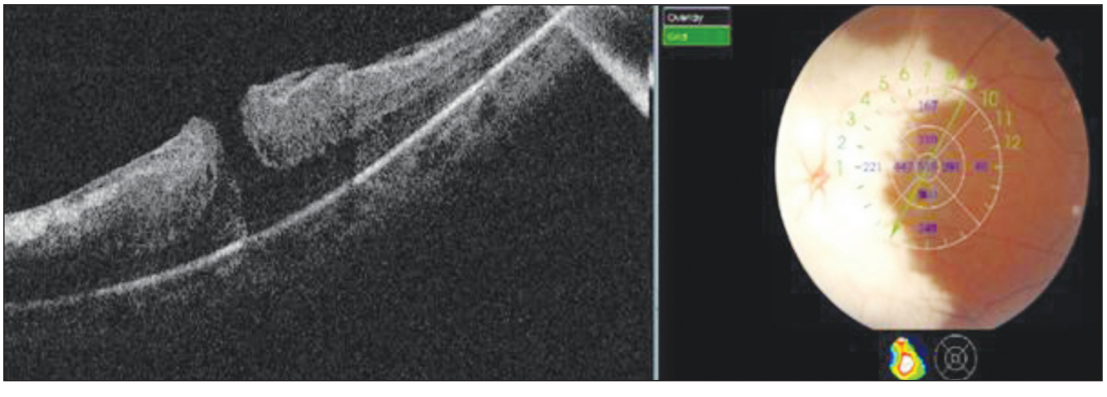

FIGURE 2: A spectral domain optical coherence tomography cross-sectional image passing through the fovea of the patient's left eye.

tion, the patient presented in this current case had Type 2 myelinated retinal nerve fiber with nasal hemifield involvement.

A higher degree of myopia and a lower level of visual acuity may be connected with larger patches of myelinated nerve fibers in the retina, according to scientific speculation. In a study that involved twelve patients, it was discovered that the degree of myelination surrounding the fovea was connected with the visual prognosis of the patients. The results of this study indicate that patients with retinal involvement of five clock hours or less showed the greatest improvement in their visual acuity through the use of occlusion therapy and spectacle correction. On the other hand, patients with retinal involvement of nine clock hours or more showed the worst outcomes.[7] Vitreoretinal interface diseases such as epiretinal membrane, vitreomacular traction, and macular hole accompanied by myelinated retinal nerve fibers have been described in the literature.[8-10] Minami et al. reported a case of failed surgical closure of macular hole associated with myelinated retinal nerve fiber around the optic disc.[9] Yang and Chen presented a case of macular hole and retinal detachment, in which artificial posterior vitreous detachment could not be induced due to tight vitreoretinal adhesion in the myelinated retinal nerve fiber area.[10] These reports suggest that the visual prognosis of myelinated nerve fiber cases may be worse when accompanied by findings of vitreoretinal interface diseases. In addition, when vitreoretinal interface diseases accompany Straatsma syndrome cases, which by definition have amblyopia, visual prognosis may be even more affected. Posterior segment findings such as myopic foveoschisis and dome-shaped maculopathy associated with high myopia, have also been described in Straatsma syndrome.[11,12] These pathologies occuring due to myopia may also worsen the visual prognosis. The challenge seen in the patients with Straatsma syndrome is treating the amblyopia. The presence of conditions such as profound anisometropia, strabismus, Type 2 myelination, extensive myelination, and macular involvement has been associated with poor visual prognosis despite treatment. Although these factors affect the visual function, occlusion therapy in Straatsma syndrome may be beneficial in the early period.[13] Usui-Ouchi et al. reported a case of macular hole retinal detachment associated with Straatsma syndrome.[14] In their case, it was noted that after surgery, some of the myelinated nerve fibers regressed, and the patient's vision improved from 20/600 to 20/300. The authors suggested that the loss of myelinated nerve fibers might be due to nerve fiber layer damage occurring during retinal detachment or internal limiting membrane peeling procedures. Bleicher et al. also reported that a macular hole associated with Straatsma syndrome closed after surgery; however, they observed the development of serous choroidal effusion postoperatively.[15] The authors argued that the presence of an unusually strong vitreoretinal adhesion in the region of the myelinated nerve fibers, as well as the development of postoperative serous retinal detachment, suggests that myelination in Straatsma syndrome might contribute to vitreoretinal adhesion and the formation of macular holes . In this current case, vitreoretinal surgery was recommended to the patient, but the patient declined. Therefore, findings during and after the surgery could not be discussed.

In conclusion, in Straatsma syndrome, where a visual impairment is observed due to profound anisometropia and lack of visual rehabilitation, additional findings such as Type 2 myelination, diffuse myelination and vitreoretinal interface diseases may be associated with worsening prognosis.

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Conflict of Interest

No conflicts of interest between the authors and/or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Halil İbrahim Ateşoğlu;* ***Design:*** *Halil İbrahim Ateşoğlu;* ***Control/Supervision:*** *Halil İbrahim Ateşoğlu,*

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