



# Straatsma Syndrome: Should Visual Prognostic Factors Be Taken into Account? A Case Report

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## Abstract

Straatsma syndrome is the triad of myelinated retinal nerve fibers, myopia, and amblyopia and may be associated with strabismus, nystagmus, hypoplastic optic nerve, and heterochromia iridum. The degree of anisometropia, presence of strabismus, extent of myelination, and macular involvement have been reported to be associated with poor visual acuity after occlusion therapy for amblyopia in this syndrome. Here we present two cases of Straatsma syndrome with different responses to occlusion therapy and discuss their treatment responses according to prognostic factors for post-occlusion visual acuity.

**Keywords:** Amblyopia, myelinated retinal nerve fibers, straatsma syndrome, prognostic factors

## Introduction

Straatsma syndrome was originally described by Straatsma et al.<sup>1</sup> in 1979 in a case series of 4 patients with unilateral myopia, amblyopia, and strabismus associated with myelinated retinal nerve fibers (MRNF). With the growing literature, the triad of MRNF, myopia, and amblyopia is now accepted as Straatsma syndrome.<sup>2</sup> However, additional findings such as strabismus, nystagmus, hypoplastic optic nerve, and heterochromia iridum have also been reported and do not preclude the diagnosis of this syndrome.<sup>2,3,4</sup> There is even a reported variation of the triad with hyperopia instead of myopia, called “reverse Straatsma syndrome.”<sup>5</sup> Although it is generally unilateral, bilateral cases of traditional and reverse Straatsma syndrome have also been reported.<sup>2,6</sup>

The challenging part of the syndrome is treating amblyopia. 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.<sup>3,7,8,9</sup>

This report presents two cases of traditional Straatsma syndrome and discusses the patients’ responses to occlusion therapy according to the literature knowledge.

## Case Reports

### Case 1

An 8-year-old girl was referred to our clinic with a 1-year history of blurred vision in the right eye (RE). Her family history was unremarkable. On examination, the patient’s best-corrected visual acuity (BCVA) was 6/120 in the RE and 6/6 in the left eye (LE). Cycloplegic refraction was -3.00 diopters (D) in the RE and -0.25 D in the LE. Pupillary reflexes were equal and symmetric with no relative afferent pupillary defect. Cover-uncover and alternate cover tests were normal for both distance and near fixation. Bilateral slit-lamp examination and Goldmann applanation tonometry were unremarkable. Dilated fundus examination of the RE revealed 5 clock hours of MRNF along the superior arcade and 4 clock hours of MRNF along the inferior arcades with the macula spared, normal foveal reflex, and normal optic disc

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(Figure 1a). Dilated fundus examination of the LE was normal (Figure 1b). Spectral-domain optical coherence tomography (SD-OCT; Heidelberg Spectralis, Heidelberg Engineering GmbH, Germany) showed hyperreflective MRNF that cast a shadow obscuring the outer retinal details in the affected areas (Figure 1c) and normal fovea in the RE (Figure 1d). Fundus autofluorescence (FAF) imaging (Heidelberg Spectralis, Heidelberg Engineering GmbH, Germany) in the RE revealed hypoautofluorescence in the corresponding areas of MRNF (Figure 1e). SD-OCT and FAF were unremarkable in the LE. Axial lengths evaluated with optical biometer (Lenstar, Haag-Streit, Koeniz, Switzerland) were 25.02 mm in the RE and 22.96 mm in the LE.

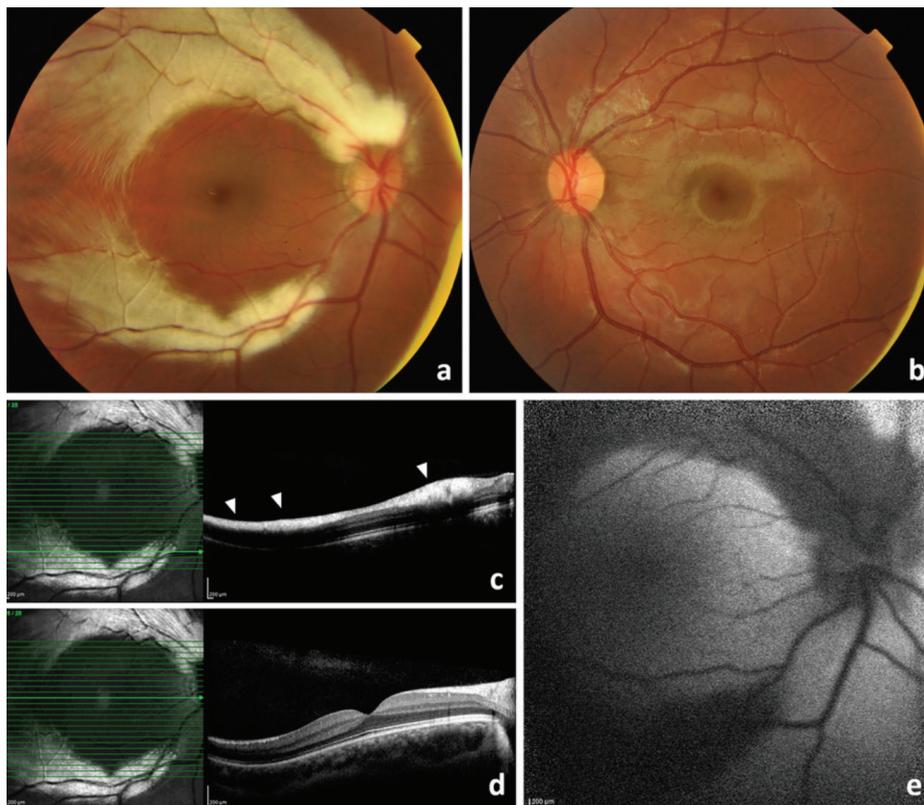
Four hours per day LE occlusion therapy with best spectacle correction was initiated for anisometropic amblyopia associated with Straatsma syndrome. However, despite patient compliance and the parents' active engagement with the occlusion regimen, 1 year following initial presentation, the patient's BCVA remained 6/120 in the RE.

#### Case 2

A 6-year-old boy presented to our clinic for routine examination. Patient history revealed that spectacle correction and occlusion therapy were recommended by another clinic 1 year ago, but the patient did not tolerate the therapy. Family history

was unremarkable. On examination, his BCVA was 6/6 in the RE and 6/30 in the LE. Cycloplegic refraction was -1.00 D and -3.25 D in the RE and LE, respectively. Pupillary reflexes were equal and symmetric with no relative afferent pupillary defect. Bilateral slit-lamp examination and Goldmann applanation tonometry were unremarkable. Cover-uncover and alternate cover tests were normal for both distance and near fixation. Dilated fundus examination of the RE was normal (Figure 2a). Dilated fundus examination of the LE revealed 6 clock hours of MRNF along the superior arcade with minimal obliteration of the optic disc superiorly, spared macula, and normal foveal reflex (Figure 2b). SD-OCT showed hyperreflective MRNF that cast a shadow obscuring the outer retinal details at the affected sites (Figure 2c) and normal fovea in the LE (Figure 2d). FAF imaging revealed hypoautofluorescence in the corresponding areas of MRNF in the LE (Figure 2e). SD-OCT and FAF were unremarkable in the RE. Axial lengths evaluated with optical biometer were 24.40 mm in the RE and 25.51 mm in the LE.

Three hours per day RE occlusion therapy with best spectacle correction was initiated for anisometropic amblyopia associated with Straatsma syndrome. With patient compliance and parents' active engagement with the occlusion regimen, the patient's BCVA improved 2 lines to 6/15 in the LE at 1 year after initial presentation.



**Figure 1.** Color fundus photographs, spectral domain optical coherence tomography (SD-OCT), and fundus autofluorescence (FAF) images of patient 1. a) Right fundus image showing myelinated retinal nerve fibers (MRNF). b) Left fundus image appears normal. c) SD-OCT scan passing through a retinal section containing hyperreflective MRNF (arrowheads) along the inferior temporal vascular arcade. d) SD-OCT scan passing through the normal fovea. e) FAF showing hypoautofluorescence in the areas corresponding to the MRNF

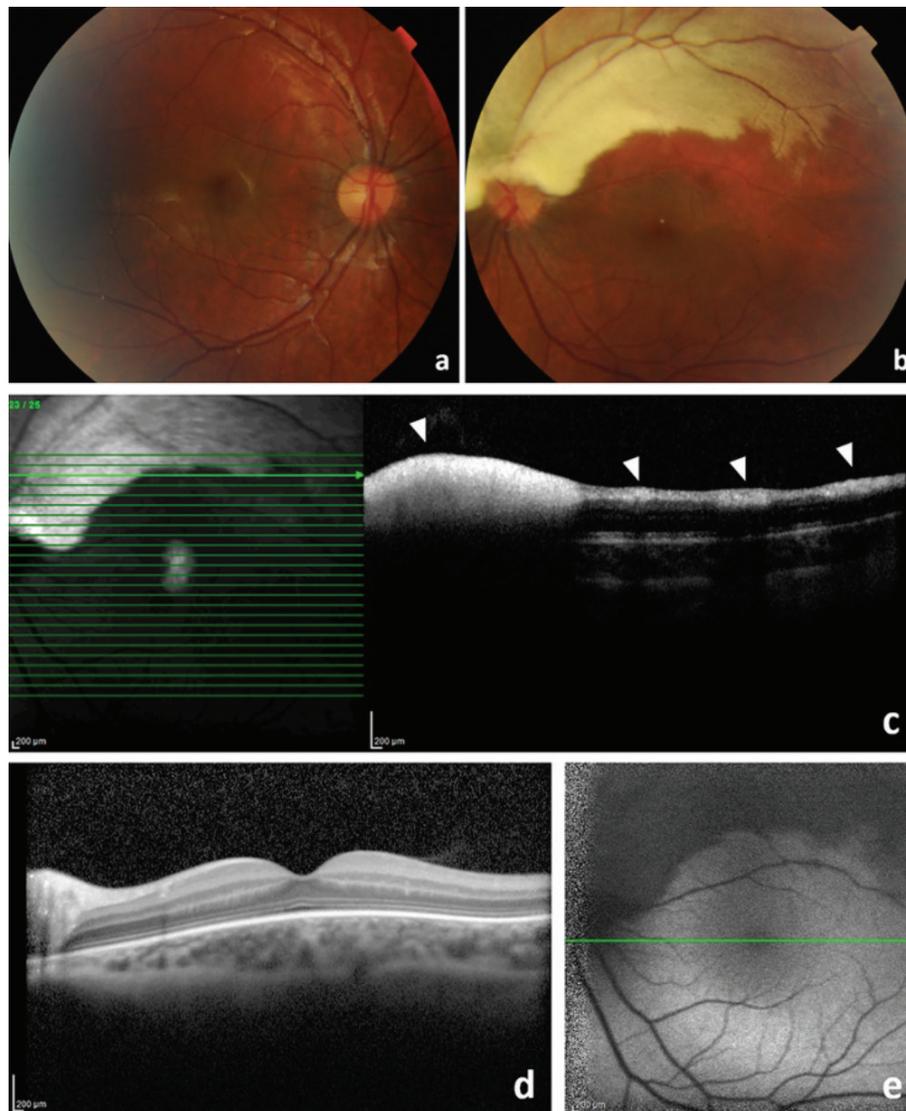
## Discussion

MRNF are rare lesions that were observed in only 0.98% of individuals and 0.54% of eyes in the large study of 3968 consecutive autopsies conducted by Straatsma et al.<sup>10</sup> They are mostly seen as white to gray-white striated patches with feathery borders following the distribution of the retinal nerve fibers. They are generally isolated findings, but can also be associated with various ocular and systemic abnormalities.<sup>3</sup> MRNF are commonly asymptomatic, and their effect on visual function is highly variable depending on the lesion's location and extent.<sup>3</sup>

Patients with MRNF often demonstrate axial myopia rather than refractive myopia.<sup>1</sup> In the literature, there is no consensus about whether the blurred image created by myelination initiates the vicious cycle of axial myopia and amblyopia

or whether the axial elongation of the globe causes late closure of the lamina cribrosa, leading to myelination and amblyopia.<sup>3,11,12,13</sup> According to animal studies and observational clinical studies, the growth of the eye is influenced by the quality of the retinal image not just at the fovea but across a wide area of the retina.<sup>14</sup> On the other hand, the theory that elongation of the globe is caused by poor retinal image quality conflicts with cases of reverse Straatsma syndrome (hyperopia with amblyopia, MRNF, and strabismus) and suggests that the etiological relationship between myelination and myopia may not be a strong association.<sup>5,6</sup>

The most critical and challenging part of Straatsma syndrome is the associated amblyopia and its treatment. There are several factors related to poor visual outcomes after occlusion therapy for amblyopia in Straatsma syndrome. An important prognostic



**Figure 2.** Color fundus photographs, spectral domain optical coherence tomography (SD-OCT), and fundus autofluorescence (FAF) images of patient 2. a) Right fundus image showing normal appearance. b) Left fundus image showing myelinated retinal nerve fibers (MRNF). c) SD-OCT scan passing through a retinal section containing hyperreflective MRNF (arrowheads) along the superior temporal vascular arcade. d) SD-OCT scan passing through the retinal section indicated by the green line in panel corresponding to the normal fovea. e) FAF showing hypoautofluorescence in the areas corresponding to the MRNF

factor according to the major series in the literature seems to be the degree of anisometropia. In a case series by Hittner and Antoszyk<sup>7</sup>, patients with higher degrees of anisometropia (an average of -13.00 D) tended to have lower post-treatment visual acuity than patients with lower degrees of anisometropia (an average of -3.75 D). Other studies also reported similar trends.<sup>3,8,9</sup> In our cases, although anisometropia was relatively low in both patients (-2.75 D in patient 1 and -2.25 D in patient 2), the higher anisometropia in patient 1 was also associated with a worse post-treatment visual outcome. However, it should be noted that the lower initial visual acuity in patient 1 may have affected this outcome.

In patients with isolated anisometropic amblyopia, occlusion therapy often produces variable results. Because of poor visual acuity outcomes despite aggressive therapy in MRNF patients, Ellis et al.<sup>11</sup> postulated that an organic etiology might also be present in these patients in addition to functional amblyopia. Abnormal macular appearance on fundus examination was also reported in several papers before the era of OCT, suggesting an organic etiology underlying poor post-treatment visual acuity.<sup>3,7,11</sup> In a recent case series of 3 patients with Straatsma syndrome, poor visual acuity was associated with loss or disturbance of the ellipsoid zone (EZ), and it was postulated that organic pathology in patients with poor prognosis might be related to that.<sup>15</sup> However, in both of our cases, the EZ was intact (Figure 1d and 2d). Therefore, we believe that while the expectation of inadequate treatment response in a patient with impaired EZ is rational, an intact EZ should not be assumed to be associated with good treatment response.

Accompanying strabismus has also been associated with poor visual outcome and a higher degree of myopia in Straatsma syndrome patients.<sup>3,9</sup> There was no strabismus in our presented cases, but as can be appreciated, a high degree of anisometropia and amblyopia can lead to strabismus in cases such as our patient 1, and strabismus alone should not be regarded as an independent factor for treatment response.<sup>9</sup>

According to their location, three types of MRNF have been described: type 1 (the most common), along the superior temporal arcade; type 2 (the least common), along both temporal arcades; and type 3, with no continuity with the optic disc.<sup>11</sup> Among them, type 2 MRNF was usually associated with a worse prognosis.<sup>10,11</sup> Although not included in this classification, rare cases of macular involvement with extensive MRNF have also been reported and associated with severe photophobia and vision loss.<sup>16</sup> According to this classification, patient 1 in our report corresponded to type 2 MRNF and, consistent with the literature, had poor visual prognosis after occlusion therapy. However, in a recent case report of a patient with BCVA of 20/400 and 30 prism diopter (PD) esotropia with type 2 MRNF in fundus examination, extensive occlusion therapy and contact lens correction was reported to be effective in improving BCVA to 20/30 and esotropia to 12 PD.<sup>4</sup>

Studies have shown that more extensive areas of MRNF may be associated with higher myopia and poor visual acuity.<sup>8,13</sup> In a study of 12 patients with MRNF, poor post-treatment visual

acuity was associated with the extent of myelination around the fovea on a clock hour scale. According to the study, patients with 5 clock hours or less of retinal involvement showed the best improvement, and patients with 9 clock hours or more of retinal involvement showed the worst results.<sup>8</sup> Similarly, in our cases, the patient with 9 clock hours involvement (patient 1) had the worst outcome. However, this classification should be approached with caution because it does not show all retinal involvement on an area basis.

In conclusion, Straatsma syndrome seems to be generally associated with poor visual outcomes, especially in patients with poor post-treatment visual acuity predictors such as deep anisometropia, strabismus, type 2 myelination, extensive myelination, and macular involvement. However, there are several reports of unexpectedly good responses in the literature, even in patients with these predictive factors.<sup>4,17</sup> Therefore, we believe that all patients with Straatsma syndrome should be approached optimistically and provided aggressive amblyopia treatment with appropriate refractive correction.

#### Ethics

**Informed Consent:** Obtained.

**Peer-review:** Externally peer reviewed.

#### Authorship Contributions

Concept: M.O.S., A.A., Ö.Ş., Design: M.O.S., A.A., Ö.Ş., Data Collection or Processing: M.O.S., N.F.K., Analysis or Interpretation: M.O.S., A.A., Ö.Ş., Literature Search: M.O.S., N.F.K., Writing: M.O.S., N.F.K.

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