

# Myelinated Retinal Nerve Fiber and Associated Pathologies: High Myopia, Amblyopia, Leukocoria and Strabismus

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## ABSTRACT

Myelinated retinal nerve fibers (MRNF) is a developmental anomaly that is seen as gray-white patches in the retina. They may be associated with ipsilateral high myopia and amblyopia, and also may be seen as an isolated retinal abnormality. In some cases MRNF may be confused with congenital cataracts and retinoblastoma, when it is presented with leukocoria and strabismus. In this article, we aimed to present two MRNF cases, which one of them was presented with amblyopia and the other with leukocoria and strabismus.

**Key Words:** Myelinated retinal nerve fiber, Amblyopia, High myopia, Leukocoria, Strabismus.

## INTRODUCTION

Myelinated retinal nerve fiber (MRNF) is a developmental anomaly that manifests as striped, gray-white patches compatible with normal retinal fiber distribution.<sup>1</sup> It was first described by Virchow in 1856.<sup>2</sup> It is seen in 1% of eyes in the population. It can be either congenital or acquired.<sup>3</sup> The most common localization is superior peripapillary region.<sup>4</sup> In the pathogenesis, ectopic oligodendrocyte-like cells localized at retina are implied.<sup>5</sup> In addition to association with many ocular and systemic disease, it was reported that ipsilateral high myopia (axial myopia) and amblyopia accompanied to MRNF in some cases.<sup>6</sup> Ipsilateral retinal nerve fiber myelination, myopia and amblyopia triad was termed as “Straasma Syndrome” by Straasma.<sup>4</sup>

Here, we aimed to present 2 cases diagnosed as unilateral MRNF, high myopia and amblyopia.

## CASE 1

A 4-years old girl was referred to our clinic with unexplained impairment of vision in left eye. The patient was attending

to control visit for high myopia and amblyopia in left eye in another facility and she was using eye masking (4 hours/day) in right eye. She was born at term and had no additional disease in the history. In addition, family history was negative of ophthalmological disorder. In the examination, best corrected visual acuity (BCVA) was 1.0 in right abd 0.1 in left eyes and there was no strabismus. No marked refractive error was observed in the right eye (-0.25 -0.25 axis 90) while there was high myopia in the left eye (-8.00 -0.75 axis 9). Anterior segment was normal. In the fundus examination, right eye was normal while there were stripped, white-gray lesions that follow normal retinal nerve fiber distribution, which started from peripapillary area at superior to optic disc and involved superior half of superior pole (Picture 1). It was considered that the retinal lesion was compatible with RMNF. By these findings, the patient was diagnosed as unilateral RMNF anomaly, high myopia and amblyopia syndrome. Appropriate eyeglasses were prescribed with aim of full recovery and eye masking (6 hours/day) was initiated in the right eye. The patient was consulted to pediatrics department for evaluation of additional systemic anomaly.

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Received: 30.03.2018

Accepted: 04.03.2019

Ret-Vit 2020; 29: 68-71

DOI: 10.37845/ret.vit.2020.29.12

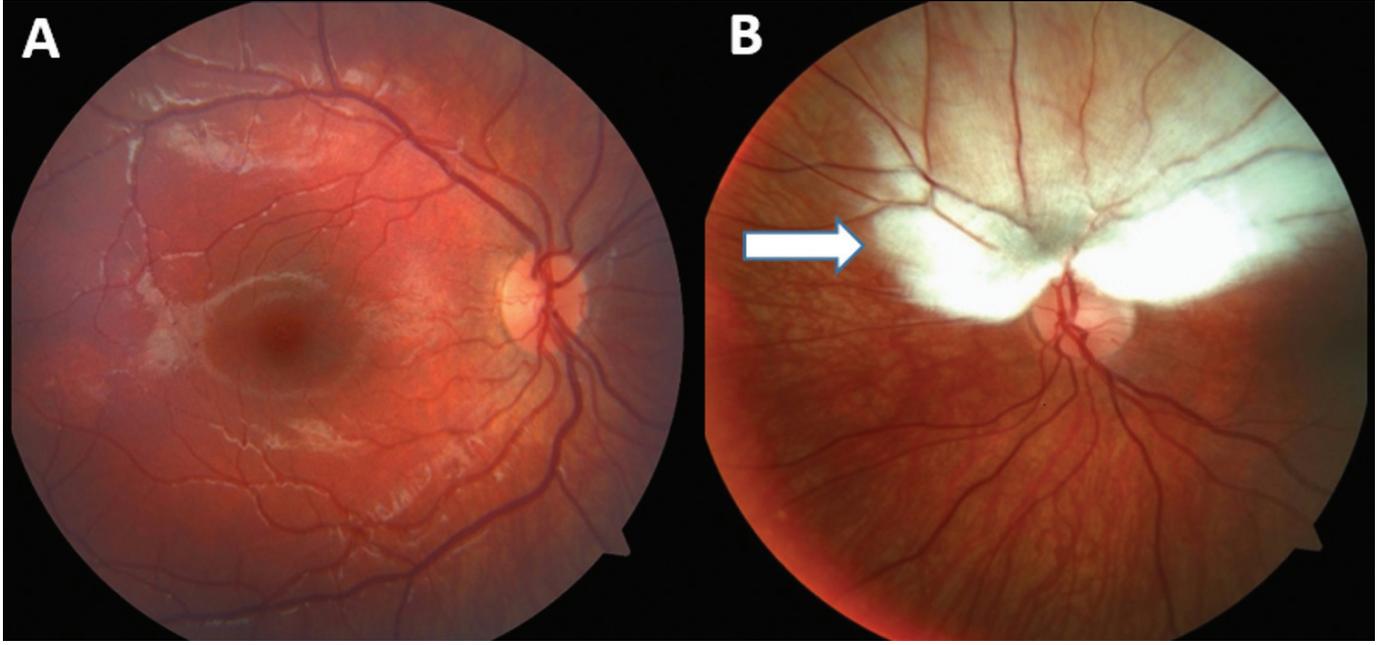
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**Picture 1.** Color fundus image of right and left eye in case 1. **A.** Retina was normal in right eye. **B.** A lesion starting from superior margin of optic disc and extending to periphery compatible to myelinated retinal nerve fiber in left eye (white arrow).

## CASE 2

A years old boy was referred to our clinic with leucocoria and strabismus in right eye. The patient was born at term and had no additional systemic disease in the history. In the examination, there was no light or object perception in the left eye but not in the left eye. There was high myopia (-8.75 -1.75 axis 75) in the right eye but no refractive error

was detected in the left eye (+0.50 -0.25 axis 100). In the anterior segment examination of right eye, leucocoria and 45 prism D exotropia were striking (Picture 2). In the fundus examination, a yellow-white lesion following normal distribution of normal retinal fibers were observed, which starts from peripapillary and involves superior and inferior temporal arcades in the right eye (Picture 2). Fundus examination was normal in the left eye. The



**Picture 2.** Color fundus image of right and left eye in case 1. **A.** Leucocoria and exotropia in re gözde lokokori ve ezotropya gorulmekte **B.** Sag göz renkli fundus fotografında beyaz ok ile gösterilen peripapiller baslayan ve arka kutupta yaygın olarak dagilim gosteren retinanin myelinize sinir lifi ile uyumlu lezyon izlenmekte.

retinal lesion in the right eye was compatible with RMNF due to its distribution pattern and clinical presentation. The patient was diagnosed as RMNF anomaly, high myopia and amblyopia syndrome together with leukocoria and strabismus. Appropriate eyeglasses were prescribed with aim of full recovery and eye masking (6 hours/day) was initiated in the left eye. The patient was consulted to pediatrics department for evaluation of additional systemic anomaly.

## DISCUSSION

Although RMNF is frequently seen as isolated, it can be accompanied by several anomalies. It can be seen together with systemic disorders such as neurofibromatosis type 1 and Gorlin syndrome.<sup>6,7</sup> The RMNF can be observed in association with ocular and cranioccephalic developmental anomalies such as coloboma, polycoria, dyscrania and keratoconus.<sup>4</sup> In addition, retinal vascular abnormalities such as neovascularization, arterial and venous occlusions and vitreous hemorrhage can be seen in relation with RMNF.<sup>8-10</sup> Other lesions that may accompany to RMNF include epiretinal membrane, vitreomacular traction and retinal rupture.<sup>11-13</sup>

Although RMNF is often asymptomatic, it can manifest as a characteristic syndrome characterized by high myopia and amblyopia. The effect on vision is highly variable. In lesions with macular involvement, there may be worse visual prognosis and severe photophobia in rare instances.<sup>14</sup> Degree of anisometropia is another factor influencing on visual prognosis. In many studies, it has been suggested that amblyopia will be more profound by increasing degree of anisometropia.<sup>15,16</sup> Similarly, marked anisometropia and profound amblyopia were observed in our cases.

Although strabismus is a common condition seen in RMNF cases, it is linked to poor prognosis.<sup>17</sup> Strabismus was observed in the second case presented.

As known, leukocoria is a concerning finding at childhood. In the literature, it was reported that the syndrome can present with leukocoria in rare cases; thus, it can be confused with retinoblastoma.<sup>18</sup> The first case presented here was also referred to our clinic with initial diagnosis of retinoblastoma due to presence of leukocoria; however, addition of other findings excluded retinoblastoma.

It has been reported that a small optic disc can be observed due to myopia and that optic disc atrophy may present in RMNF patients.<sup>3,19-21</sup> No optic disc hypoplasia or small optic disc was observed in both cases presented here.

In these patients, full myopia correction should be

performed and aggressive amblyopia therapy should be initiated at early period; however, patients are unresponsive to treatment in most cases.<sup>22</sup>

There are several approaches to occlusion therapy. In the literature, there are cases with good treatment response as well as refractory cases.<sup>23,24</sup> In patient groups with poor response to treatment, it should be kept in mind that structural and organic problems may also be present.<sup>25</sup> It seems that degree of macular involvement is a predictive factor in the assessment of response to occlusion therapy.<sup>26</sup>

In conclusion, RMNF anomaly should be kept in mind in patients presenting with high myopia, amblyopia, strabismus and leukocoria and the patient should be evaluated for RMNF. Patients with one of these ocular findings should be assessed for other potential that may be present. Although treatment response is highly variable in patients with MRNF syndrome, eye masking should be performed and detailed examination should be performed regarding additional abnormalities.

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