



A 6-Year-Old Girl with Abnormal Fundus Findings in Her Left Eye

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Introduction:

A 6-year-old girl was referred to clinic for abnormal fundus findings. She had come to medical attention after she failed a pediatric vision screening examination. According to parental report, the patient's left eye was usually turned in. The patient reported blurry vision in her left eye but was unsure how long this had been present. She had no known relevant past medical history or family history of significant ocular disease.



Figure 1: External photograph of patient demonstrating left esotropia.

Exam:

Best corrected visual acuity was 20/25 in the right eye with cycloplegic refraction of +1.00 sphere and count fingers vision in the left eye with cycloplegic refraction of -7.00 - 3.00 x 180. In addition to this profound anisometropia, the patient was found to have a comitant left esotropia of 20 prism diopters. On dilated funduscopic examination, the right eye was normal, while the left eye was notable for profound whitening, thickening, and opacification of the nerve fiber layer across much of the posterior pole, including the macula. There was a wedge of normal nerve fiber layer emanating from the inferotemporal aspect of the optic disc across the inferior macula. The left fovea was not visible beneath the opaque nerve fiber layer. Both discs appeared normal otherwise.

Diagnosis:

The patient's profound vision loss in significant anisometropia raises high concern for left-sided amblyopia. Overall, the patient's constellation of findings is consistent with a syndrome of unilateral myelinated retinal nerve fiber layer, axial myopia, and amblyopia. The differential diagnosis

for myelinated nerve fiber layer includes cotton-wool spots, branch retinal artery occlusion, peripapillary epiretinal membrane, retinal infiltrate, and retinoblastoma.

Further testing which would be abnormal but unnecessary for diagnosis include visual field, which would most commonly show a nerve fiber layer-type pattern of unilateral field loss or generalized depression, fundus autofluorescence, which would appear dark in the myelinated areas, and optical coherence tomography, which would show a thickened nerve fiber layer.^{1,2}

Discussion:

Myelinated retinal nerve fibers appear as white, sharply demarcated peripapillary patches with feathery borders. They occur in up to 1% of all eyes, and several pathogenic mechanisms have been proposed. One early

theory was that a structural defect in the lamina cribrosa, through which the fibers comprising the optic nerve travel, allows myelin to be deposited beyond the orbital optic nerve and in the retina. Autopsy studies have not demonstrated such defects, however. In recent years, several studies have proposed retinal glial cells or heterotopic oligodendrocytes as the cause of the abnormal myelination.³ The presence of myelination can be static or dynamic, sometimes congenital and sometimes acquired, and has been demonstrated in some cases to regress after ocular insult.^{2,4-6}

The association of unilateral retinal myelination with ipsilateral high axial myopia, profound amblyopia, and strabismus, or Straatsma syndrome, has been described in the literature since the 1970s.^{7,8} Treatment involves prompt treatment with full cycloplegic refraction and deprivation therapy for amblyopia (e.g., patching or cycloplegic drops of the better-seeing eye).⁹ Due to multifactorial etiologies of vision loss, i.e., anisometropic amblyopia as well as

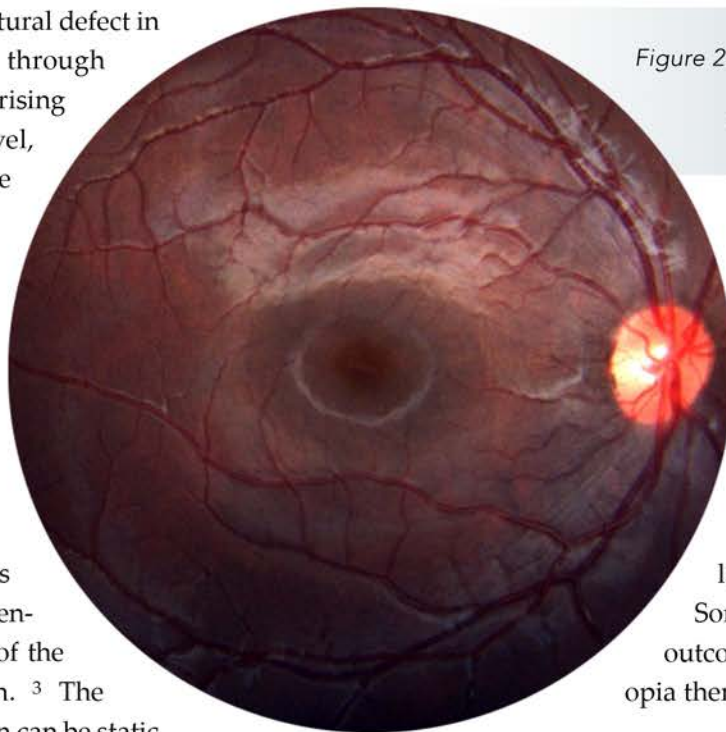


Figure 2: Color fundus photo of the right eye centered on the macula, demonstrating a normal fundus.

organic component, this treatment has often been reported to result in a partial improvement in vision. The organic vision loss has been suggested to be due to myelination in the macula impeding transmission of light through the retina or signals from the retina to the lateral geniculate nucleus.¹⁰ Some authors report poor visual outcomes even with rigorous amblyopia therapy.^{9,11}

Other associations with myelinated retinal nerve fiber layer include systemic disease such as neurofibromatosis type I and epilepsy, anterior segment abnormalities, congenital cataracts, persistent fetal vasculature, colobomas of the optic disc and uvea, and congenital cataracts.^{10,12,13}

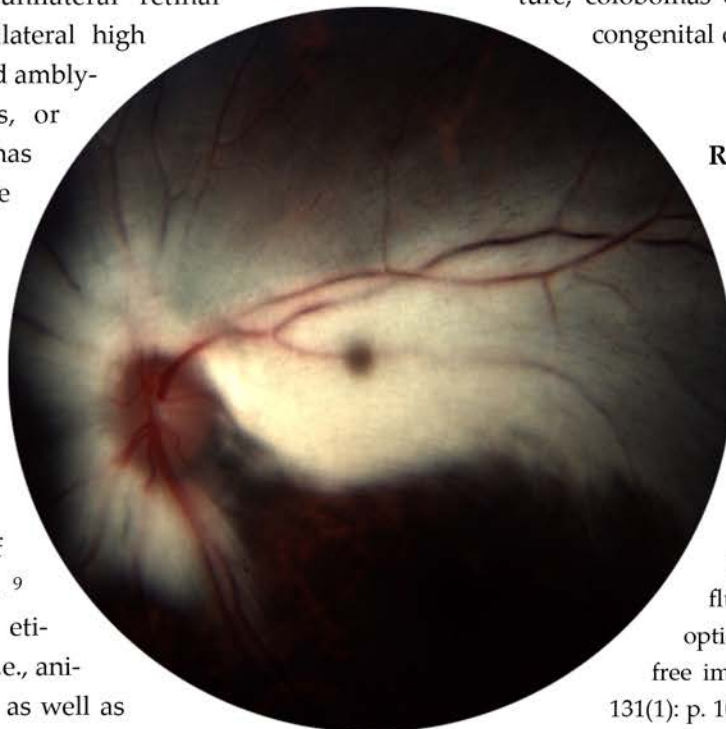


Figure 3: Color fundus photo of the left eye centered on the macula, demonstrating marked whitening and opacification of the nerve fiber layer throughout much of the macula and posterior pole. Note the asymmetry between the two eyes. The dark portions represent normal coloration, which appears darker as a result of color balancing of the digital camera.

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