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BILATERAL MYELINATED RETINAL NERVE FIBER LAYER: A RARE CASE

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ABSTRACT
Background: Myelinated retinal nerve fiber layer (MRNFL) is a rare congenital anomaly and mostly benign, which the anterior retinal nerve fibers of the lamina cribrosa have a myelin sheath. To report a rare case of bilateral myelinated retinal nerve fiber layer.
Case Presentation: A 14-year-old boy with chief complain of blurred vision in both eyes when looking at a distance, according to the patient's parents, the patient's left eye occasionally seemed to be misaligned since the age of 2 years but had never been taken for treatment, the patient's father has same complaint. Ophthalmology examination revealed 15-degree exotropia on the left eye, the other physical examination within normal limit.
Conclusion: Bilateral myelinated retinal nerve fiber layer is a benign familial condition. Common associated findings that can lead to amblyopia in children with myelinated retinal nerve fiber layer include high refractive error, strabismus, and anisometropia. Myelinated retinal nerve fiber layer usually has a good prognosis if the associated ocular findings are treated promptly, as described in this case.
Keywords: myelinated retinal nerve fiber layer, MRNFL

BACKGROUND

Myelinated retinal nerve fiber layer (MRNFL) is a rare and mostly benign congenital anomaly in which the anterior retinal nerve fibers of the lamina cribrosa have a myelin sheath.[1] Normally, myelination of the optic nerve does not extend beyond the lamina cribrosa and into the retina. Although the direct cause is unknown, MRNFL occurs when myelination extends past this point and can be detected on fundus examination, clouding the underlying retinal blood vessels.[1,2]

This anomaly can occur in up to 1% of the population, and approximately 7% of affected patients will have bilateral involvement. MRNFL is usually present at birth and involves static lesions. Although rare, acquired or progressive MRNFL conditions have been reported. MRNFL was first described in 1856 by the German pathologist, Rudolf Virchow, who described the retina as "white, very thick and wrinkled".[3] Clinically, the fibers appear as gray to white patches with well-defined borders on the anterior surface of the retina. Most cases are diagnosed incidentally in healthy individuals by ophthalmoscopy.[4]

Although most children are asymptomatic, few may present with ocular findings of high refractive error, strabismus, and amblyopia or failed vision screening because myelin blocks the transmission of light to the underlying retinal cells, there is a high probability of vision loss or an enlarged blind spot in affected patients.[5]

CASE PRESENTATION

A 14-year-old boy Malay descent was referred from a general hospital in Palembang City to the Eye Polyclinic of the Neuro-ophthalmology Subdivision of Mohammad Hoesin Hospital Palembang on November 18, 2021.

The patient complains of blurred vision in both eyes when looking at far object, especially when reading the blackboard at school, double vision is denied. According to the patient's parents, the patient's left eye occasionally seemed to be misaligned since the age of 2 years but had never been taken for treatment, then the patient's parents decided to go to an There is no pathological past medical history, no previous history of using glasses, no history of previous eye surgery or trauma, no history of seizures but there is a family history of the same disease, the patient's father.

All general physical examination within normal limit, on ophthalmological examination visual acuity on the right eye was 6/21 *ph* 6/7.5 with crowding phenomenon 6/6 and on the left eye was 6/15 *ph* (-) with crowding phenomenon 6/12, the IOP of both eyes within normal limit, the Hirschberg test was exotropia (XT) 15º, and eye movement was good to all gaze.

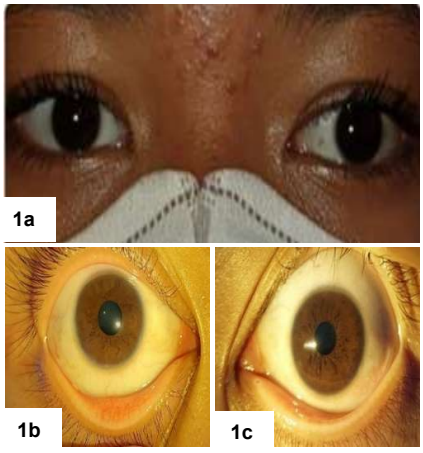

Figure 1a. Both eyes of patient, 1b. Right eye, 1c. Left eye. There are no abnormalities found on Ishihara test and Contrast Sensitivity

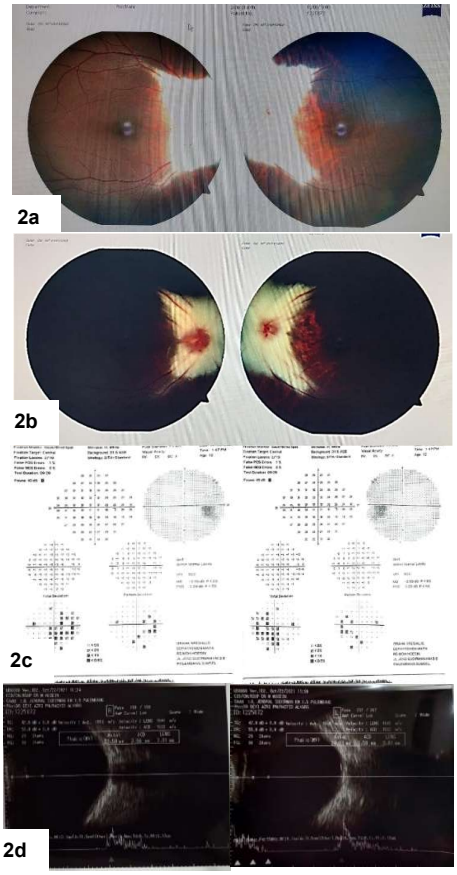

Figure 2a. The original photofundus from patient eyes, 2b. The photofundus of patient with decrease of contrast settings, 2c. Perimeter humprey of both eyes, 2d. Ultrasonography of both eyes.

Anterior segment examination all within normal limit, relative afferent pupillary defect (RAPD) on both eyes was negative, from posterior segment examination on the both eyes shows papil was round, borders are hard to identify due to covered by myelin sheath, papil were red color, and the cup-disc ratio was difficult to identify, arteries:vein was 2:3, from macula shows normal foveal reflex on both eyes, and retina on both eyes are good blood vessel contour, no exudate, no bleeding, but there was myelination sheath seen arise from RNFL papillary direction on the both eyes, and also on both eyes there was tigroid appearance.

Ultrasonography from both eyes shows no abnormality but axial length was 23.58 mm on right eye and 24.03 mm on left eye, and from automated visual field examination there was enlargement blind-spot and relative scotoma.

There was no other physical abnormalities and/or any syndrome found when we consult this patient to pediatric department except from pedigree there was same conditions that occurs on patient father and grandfather also has same eye complaint.

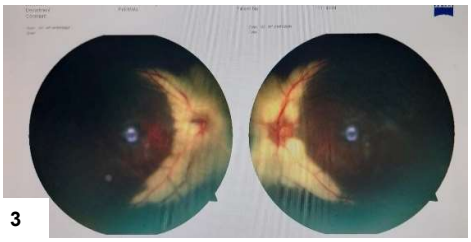

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Figure 3. The photofundus of patient father eyes

DISCUSSION

It was reported a 14-year-old boy of Malay descent with complaints of blurred vision when looking far away. The patient's had basic visual acuity in the right eye 6/21 with pinhole 6/7.5 and crowding phenomenon 6/6, on the left eye 6/15 with pinhole negative and crowding phenomenon 6/12, we can conclude this condition as amblyopia. Amblyopia develops when the brain fails to process input from one eye due to a refractive error, strabismus, or structural anomaly.[1,2]

Posterior segment examination obtained in both eyes revealed flat white patches on the retina in a distribution consistent with the retinal nerve fiber layer. MRNFL is caused by a developmental error involving the presence of ectopic oligodendrocyte-like cells in the retina, leading to myelination of retinal ganglion cell fibers.[6] This disorder occurs when the lamina cribrosa, a protective barrier, is disrupted or underdeveloped, allowing oligodendrocytes to penetrate forward into the retina. The impact that MRNFL has on visual function is highly variable and may also depend on the extent of the myelination patches or if the macula is involved although this is rare.[7] On this patient macula shows no anomaly and normal foveal reflex seen on both eyes.

The patient seems to have myopia because there was tigroid appearance on both retina and from ultrasonography transpalpebral examination there was axial length 23.58mm on right eye and 24.03mm on left eye. So the patient were consult to refractive and low vision subdivision and diagnosed as High Myopia with Anisometropia. These refractive anomalies cause blurred vision and contribute to visual acuity and strabismus.[1,2,11] Refractive errors in this patient are corrected with prescribing glasses.

Visual field examination was performed, the results showed defect on total deviation and also on pattern deviation which suggesting enlargement of blind spot and relative scotoma due to MRNFL, theoretically myelinated nerve fibers cause scotoma in the affected area and also the patients with MRNFL usually have a deep physiological cup. This is happened because occasionally, a retinal vascular inversus site or a tilted optic disc may occur on MRNFL.[1,2,11]

The position of the left eyeball was XT 15º and good eye movement in all gaze, the patient was consulted to pediatric and strabismus division, strabismic amblyopia is also treated with eye alignment, either with glasses and/or eye muscle surgery.[8] The patient presented in this case were diagnosed Exotropia Divergent Excess Alternans and requires strabismus surgery as part of the treatment of significant exotropia, which will ultimately help his vision improve further.[1,2,11]

MRNFL is characterized by non-increased whitening of the affected retina, following a pattern of distribution of nerve fibers in the retina. The affected area may

become hyperreflective on optical coherence tomography (OCT) results. MRNFL is formed due to errors during the myelination process of retinal ganglion cell fibers. For this process is the responsibility of oligodendrocytes.[9,10]

Physiologically, it begins around the 5th month of fetal life from the lateral geniculate body, follows the visual pathway and ends in the lamina cribrosa at delivery or shortly thereafter. Lamina cribrosa is a protective barrier against the penetration of myelin fibers in the retinal area. When the disorder affects the integrity and underdevelopment, oligodendrocytes penetrate between the fibers and ganglion cells produce myelin, the more, the lower the density of these fibers. And that is why myelinated nerve fibers are so rare in the macula and in the retina of the nose. [1,2,11]

The development of myelinated nerve fibers associated with the optic disc depends on the degree of disc hypoplasia and that is why fibers of this type are often accompanied by amblyopia, myopia and different presentations of strabismus.[11] As seen in the case of this patient, MRNFL can be associated with several ocular findings, and it is important to recognize and treat each one accordingly. The patient was therefore diagnosed with strabismus (exotropia divergent excess alternans), high myopia, and anisometropia, as well as bilateral MRNFL.

The patient also has a history of a similar disease in his family member, the patient father and grandfather. Genetic control of myelination of mature retinal ganglion cells is largely unknown and still an active area of research, but some cases of familial isolated MRNF have been reported, including a family with two generations of 10 cases and in a mother and daughter with bilateral MRNF.[12,13]

Most children with MRNFL have good visual potential in the affected eye because the macula is generally spared due to the high concentration of ganglion cells and low nerve fiber density. In several clinical studies, patients frequently report visual field defects. This is a relative scotoma and an enlargement of the blind spot, which corresponds to the area covered by the fibers. Retinal myelinated nerve fiber layer is usually observed as an asymptomatic isolated finding after routine examination and it is usually considered as non-progressive.[1,2,11]

CONCLUSION

Bilateral MRNFL is rare benign inherited anomaly which usually has a good prognosis if the associated ocular findings are treated promptly.

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