Bilateral Idiopathic Epiretinal Membranes Associated With Multiple Peripheral Neurofibromas In A Young Adult

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Abstract

A 28-year-old woman with multiple peripheral neurofibromas had undergone an ophthalmologic consultation. Bilateral idiopathic epiretinal membranes were detected without any other ocular changes. Though our patient did not meet the criteria set for Neurofibromatosis Type 1 and Neurofibromatosis Type 2 it is well possible that bilateral epiretinal membranes may be in the disease spectrum.

INTRODUCTION

Epiretinal membranes have been described in association with numerous ocular conditions and diseases. In children or young adults epiretinal membranes that are not the result of other ocular disorders are unusual and presumed to be congenital in origin.\(^1,2\)

Epiretinal membranes unassociated with combined hamartoma of the retina and retinal pigment epithelium (CHRPPPE) have not been reported in neurofibromatosis type 1 (NF-1). However, epiretinal membranes were described in some patients with neurofibromatosis type 2 (NF-2)\(^3,4,5\).

We hereby report a 28-year-old woman who had biopsy-proven multiple peripheral neurofibromas without any other sign of NF-1 or NF-2, and had bilateral epiretinal membranes without any other ocular sign.

CASE REPORT

A 28-year-old woman with biopsy-proven multiple peripheral neurofibromas were referred to us for ophthalmologic consultation. She developed several painless lumps in her extremities and her neck since four years of age. Histopathologic examination of an excised lump located at the right side of her neck showed findings consistent with neurofibroma. However, Horner syndrome characterized with miosis and right ptosis ensued following this surgery.

On our examination, best-corrected visual acuity was 20/80 OD and 20/25 OS. The right upper eyelid was ptotic and the pupil was miotic. There was no afferent pupillary defect. Color vision was normal bilaterally. Slit-lamp examination was unremarkable OU. Ophthalmoscopy disclosed macular epiretinal membranes in both eyes and pseudohole formation in the left eye (Figure 1a and b). Optic coherence tomography demonstrated epiretinal membranes exerting traction on macula and causing secondary foveolar detachment (Figure 1c and d).

Figure 1

Figure 1a: Right eye, Color fundus picture showing epiretinal membrane
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Physical examination yielded several neurofibromas scattered throughout the body and extremities (Figure 2a and b). There was only one cafe-au-lait spot on right axilla (Figure 2c).

Figure 2
Figure 1b: Left eye, Color fundus picture showing epiretinal membrane and pseudohole

Figure 3
Figure 1c: Right eye, Optic coherence tomogram showing the epiretinal membrane exerting traction on the macula

Figure 4
Figure 1d: Left eye, Optic coherence tomogram showing the epiretinal membrane exerting traction on the macula

Figure 5
Figure 2a: Right forearm, Neurofibroma (Arrows)
Figure 6
Figure 2b: Left palm, Neurofibroma (Arrows)

Figure 7
Figure 2c: Right axilla (Dorsal plane) Cafe-au-lait spot

Despite physical and neurologic examinations together with laboratory and radiologic investigations no other signs relevant to neurofibromatosis type I and II were detected. Our diagnosis was isolated multiple neurofibromas associated with idiopathic bilateral epiretinal membranes. The patient denied surgery for epiretinal membranes and is under follow-up.

DISCUSSION
Epiretinal membrane formation was previously described in patients with NF 2. Landau and Yasargil, evaluated prospectively fundus changes in six patients with a confirmed diagnosis of NF 2 and found that four of them had epiretinal membranes. They concluded that epiretinal membranes are possibly of congenital origin similar to those previously reported in healthy children and young adults. Meyers et al. reported that in 12 of 15 patients with NF 2 an epiretinal membrane in the macular or paramacular area was present. Also one patient had an epiretinal membrane in one eye and CHRPPE in the other eye. They underlined the fact that most of the epiretinal membranes in the macula did not affect visual acuity and speculated that in NF 2, in response to pathophysiologic factors that cause proliferation of neural crest and other cells elsewhere in the nervous system, cells of neural crest or neuroectodermal origin at the vitreoretinal juncture or in the retina proliferate or develop abnormally resulting in epiretinal membranes at an early age.

CHRPPE, and epiretinal membrane represent a spectrum of lesions from vascular malformations at one end to CHRPPE at the other, and some authors have postulated that epiretinal membranes in NF 2 patients are form fruste of CHRPPE. Vianna et al. reported a single case with bilateral CHRPPE and NF 1 and Destro et al. reported that two of their 5 patients with retinal tumors and NF 1 had CHRPPE.

Though our patient did not meet the criteria for NF 1 and NF 2 she had multiple peripheric neurofibromas. It is well possible that idiopathic epiretinal membranes may be in the disease spectrum.

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