Successful surgical management of bilateral epiretinal membrane in a child with only café-au-lait spots

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A 6-year-old boy diagnosed as anisometropic amblyopia, with only café-au-lait spots and a family history of neurofibromatosis, presented with decrease in vision in the both eyes. Dilated fundus examination showed epiretinal membrane in both eyes over the macula. He underwent successful surgical management of the epiretinal membrane.

Key words: Epiretinal membrane, histopathology, neurofibromatosis, pediatric, surgical outcome of pediatric vitrectomy

Neurofibromatosis (NF)-2, an autosomal-inherited dominant neurocutaneous disorder,[1] is characterized by peripheral tumors (vestibular schwannoma) and central nervous system tumors (meningiomas and ependymomas).[2] The ocular manifestations of NF-2 are varied[3] and often the first sign of the NF.[2] The incidence of retinal hamartomas is reported to be between 8% and 22%.[4,5] Epiretinal membranes (ERMs), also known as macular pucker or cellophane maculopathy, are visualized on the macular surface as wrinkles or folds.[6] They are usually seen in patients above the age of 50 years and 30% show bilaterality although may have an asymmetrical presentation.[6] Macular ERMs are rare in younger patient (<40 years) except in eyes with trauma, uveitis, retinal vascular disease, or tumors.[3,6] ERMs in NF 2 are thought to be hamartomatous in origin as evidenced by histopathological examination (HPE).[7] We here describe surgical management of ERM in both eyes with successful visual outcome in a child with café-au-lait spots and a family history of vestibular schwannoma and NF 2.

Case Report

A 6-year-old boy presented to the outpatient clinic of our center with complaints of progressive decrease in vision in both eyes and inability to attend school. He had been using glasses for the past 1 year for poor vision in the left eye (OS) and was advised patching therapy for amblyopia. He had a positive family history of NF affecting his mother and maternal uncle. The child was never evaluated for NF and did not undergo any genetic testing. On examination, his best-corrected visual acuity (BCVA) was 6/36 N10 (OD) with a myopic correction of -1.00Dsph and 3/60 N36 (OS) with a myopic correction of-8.00Dsph/-2.00 × 180°. Slit-lamp examination of anterior segment of both eyes was unremarkable. Dilated fundus examination of both eyes showed a gray-flat fibrous lesion at the macula obscuring the fovea and the perifoveal region [Fig. 1a] suggestive

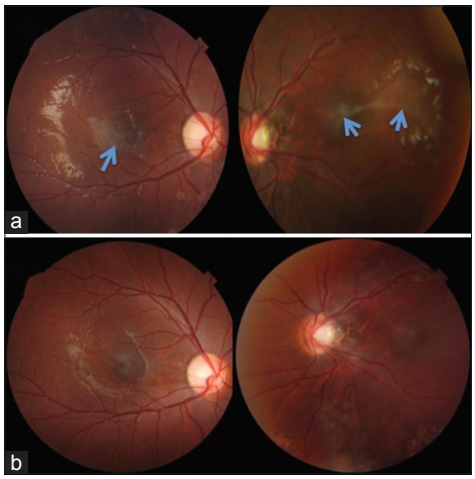

Figure 1: Color fundus photograph (a) Preoperative photograph showing a thick broad epiretinal membrane at the macula (blue arrow) in the right and left eye, (b) 5 months postoperative photograph showing the absence of recurrent epiretinal membrane

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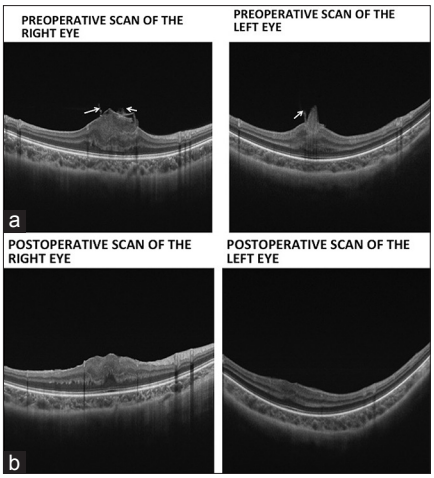

Figure 2: (a) Preoperative optical coherence tomography showing thick epiretinal membrane across the surface of the macula with curled edges stretching into the vitreoretinal interface (white arrows). (b) Postoperative optical coherence tomography showing some distortion of the fovea with edema present at the macula region

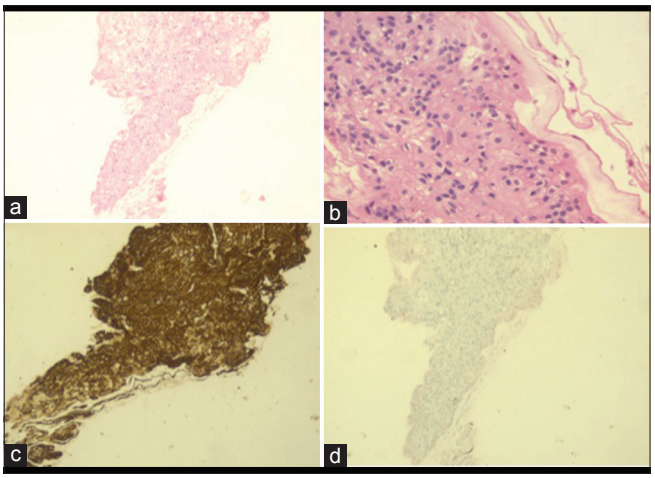

Figure 3: Photomicrograph of epiretinal membrane highlighting the round to oval cells with minimally pleomorphic nuclei with chromatin inconspicuous nucleoli and eosinophilic to vacuolated fibrillary cytoplasm. The cells are positive for glial fibrillary acidic protein and negative for CAM 5.2. (a) H and E, (×40), (b) H and E, (×400), (c) immunopositivity for glial fibrillary acidic protein (×90), (d) immunonegativity for CAM 5.2 (×90)

of a dense macular ERM. Swept-source optical coherence tomography (OCT) (Topcon, Triton plus Version 10.00) of OD showed an ERM stretching across the surface of the macula with curled edges stretching into the vitreoretinal interface with marked retinal thickening [Fig. 2a]. Due to poor vision and poor fixation in OS, a good quality OCT image could not be obtained. Fluorescein angiography of both eyes was unremarkable. The child was sent for a full pediatric and ENT evaluation as he was found to have café-au-lait spots on his trunk, upper and lower limb (3 spots were >0.5 cm and 3 spots were <0.5 cm in size). The BERA test suggested retrocochlear pathology. The MRI brain and blood investigations were within normal limits. He underwent pars plana 23-gauge vitrectomy with ERM peeling in both eyes within a time interval of 1 month. Intraoperatively, vitreous was very firmly attached to the membrane and adherent to a fibrotic tissue on the retina in two places. The ERM was peeled off from the underlying retina. The vitreomembrane junction had to be cut and endolaser was given to the areas surrounding the fibrotic tissue as there was bleeding and the surgeon (TK) was unsure if there was an underlying tear behind the bleed. The membrane was sent for HPE including light microscopy which showed scanty fragments composed of cords and strands of round to oval cells with minimally pleomorphic nuclei containing dispersed chromatin inconspicuous nucleoli and eosinophilic to vacuolated fibrillary cytoplasm set in an edematous myxoid stroma. On immunohistochemistry, the cells were strongly positive for glial fibrillary acidic protein and negative for CAM 5.2 suggesting a glial origin of the membrane [Fig. 3]. At 5-month follow-up visit, the BCVA improved to 6/12 N6 (OD) with −2. 50 Dsph/1.00 × 165° and 6/36 N24 (OS) with −8.00 Dsph/−1.00 × 150°. OCT done showed some distortion of the foveal area with edema still present at the macula region, but there was no membrane that could be visualized in both eyes [Fig. 2b].

Discussion

Various studies[6,8-10] have reported the common causes of ERM in children: Idiopathic (29%–65%), trauma (7%–39%), uveitis (20%–30%), familial exudative vitreoretinopathy (35%), and combined hamartoma of the retina and retinal pigment epithelium (5%–29%). In patients with NF 2, ERMs are a common clinical finding with Meyers *et al*. reporting 80% of their children with NF2 having ERM.[1,3] In children, ERM is reported as premacular fibrosis: Thick opaque membrane or macular folds or traction lines[6,11] similar to that seen in our patient. Rothman *et al*. compared the macular morphology of ERM in pediatric eyes (idiopathic, combined hamartomas, retinal vascular disease, and trauma) to that seen in adult eyes using the spectral-domain OCT. They reported that ERMs in pediatric eyes were more firmly attached to the retina, dragging the vessel, less fibrillary appearance of the inner retinal contour with taco folds, and disrupted external limiting membrane and inner segment band compared to adult eyes.[12] In NF-2, OCT features of ERM are described as unusual membrane thickness with serpentine, curled edges extending into the vitreoretinal interface.[13] This was similar to the findings in our case. Studies have reported 64%–86% success in functional visual acuity after surgical treatment for ERM due to other causes.[8,9] However, only one report in literature mentions the surgical management of ERM in a child with NF where the significance of visual improvement could not be determined.[7] In our study, the child had trouble with schoolwork due to decreasing vision caused by the macular distortion. He had only café-au-lait spots and a positive family history of NF. He underwent vitrectomy with ERM peeling. At the end of 5 months, his visual acuity improved significantly and his performance at school had improved remarkably. He was advised to continue with the patching therapy for amblyopia treatment. The HPE of the ERM in our case suggested a glial origin similar to that reported by McLaughlin *et al*.[1] The short follow-up is a limitation of this report and long-term follow-up of vision with amblyopia treatment and evaluation for recurrence of ERM is warranted.

Our report highlights the excellent functional outcome in the management of bilateral ERM in a child with café-au-lait

spots and a positive family history of NF without recurrence at 5-month follow-up [Fig. 1b]. Surgical management (pars plana vitrectomy with ERM peeling) of ERM in NF may be a better option rather than just observation as reported in literature[2,13] for an improved visual outcome.

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Conflicts of interest

There are no conflicts of interest.

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