Bilateral combined hamartoma of retinal pigment epithelium and retina

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DESCRIPTION

An adolescent girl presented with poor vision in both eyes since childhood. She was evaluated elsewhere 13 years ago and was prescribed low-vision aids. Best-corrected visual acuity was 6/24, N10 in the right eye (refraction: −2 diptres sphere) and 6/12, N8 in the left eye (refraction: −7.5 diptres sphere). The anterior segment was normal in both eyes. Both eyes showed macular thickening with preretinal membrane (figure 1A,B).

Optical coherence tomography (OCT) showed dysplastic inner retinal layers and adjacent normal-looking retina (figure 2A,B). This retinal morphology remained unchanged over 13 years of follow-up (figure 2C, D). A diagnosis of combined hamartoma of retinal pigment epithelium and retina (CRPE-RH) was made. A detailed systemic evaluation revealed that the patient had bilateral acoustic neuromas of the cerebellopontine angle consistent with neurofibromatosis type 2 (NF 2). As the vision remained unchanged for the last 13 years, no further intervention was done.

CRPE-RH is rare benign tumours of RPE and retina predominantly involving optic nerve head or juxta papillary retina. They are usually unilateral and discovered in infants or children with strabismus or subnormal vision. Clinically, they appear as slightly elevated, partly pigmented lesions with fine capillaries within the tumour and a preretinal membrane. Unlike the tumour involving optic disc, macular lesions show no evidence of RPE proliferation or capillary angiomaticus tissue at their inner surface. In young patients, OCT of these lesions shows only inner layer involvement with preserved outer nuclear layer and RPE. Other common causes of epiretinal membrane in young must be ruled out including peripheral retinal breaks, vitreous inflammation and trauma.

Bilateral presentation is rare and known to occur in NF 2.3 4 They may be mistaken for postinflammatory scar or idiopathic epiretinal membranes as OCT findings are very subtle in the young. CRPE-RH lesions may be the presenting feature of NF 2 and secondary tumours may develop later in life. Hence, a regular follow-up is warranted, especially in patients with bilateral CRPE-RH.

Learning points

► Epiretinal membranes in the young are very rare with diverse aetiologies.
► Congenital benign tumours of retinal pigment epithelium are one of the causes for epiretinal membranes.
► Bilateral epiretinal membranes may be the earliest manifestation of systemic pathologies like phacomatosis and hence they need long-term follow-up.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research.

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Figure 1 Fundus photograph of the right eye showing a greyish white epiretinal membrane with thickened macula (A). Fundus photograph of the left eye shows a subtle epiretinal membrane at the temporal edge of the macula on a tessellated background. A small focus of nevis is seen supero-temporal to the macular lesion (arrow).

Figure 2 OCT through the macula of the right (A) and left (B) eyes taken 13 years ago. Note the disorganised inner layers with preserved outer layers and a membranous proliferation at the inner surface. Current OCT scans of the right (C) and left eyes (D) showing no change in morphology over 13 years. OCT, optical coherence tomography.
Images in...

They should not be used in isolation to guide treatment choices or public health policy.

REFERENCES


