

Combined Hamartoma of the Retina and Retinal Pigment Epithelium with Poor Visual Acuity

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Combined hamartoma of the retina and retinal pigment epithelium (RPE) is an uncommon benign tumor that can resemble choroidal melanoma or retinoblastoma. This tumor can present as an isolated finding or manifest as part of the disease spectrum of neurofibromatosis (NF) type 2.¹ Visual impairment can result from parafoveal tumors.² We hereby report a case of combined hamartoma of the retina and RPE with profound poor visual acuity.

CASE REPORT

A 5-year-old white male, noted by the school nurse to have poor visual acuity in the right eye, was referred for evaluation. The patient's mother noticed right esotropia beginning at the age of 1 year; however, eye examination at that time was reportedly normal. Systemic workup revealed no features or history of NF.

On examination, visual acuity was counting fingers at 3 feet OD and 20/20 OS. Slit-lamp examination was normal for both eyes, and there were no Lisch nodules of the iris.

Fundus examination OS was unremarkable. The fundus OD showed an ill-defined pigmented retinal lesion in the papillomacular region estimated at 12 x 10 mm in basal dimensions. The overlying retinal vessels displayed slight traction (Figure 1A and B). Ultrasonography demonstrated a solid plateau-shaped retinal lesion measuring 2.7 mm in thickness with a dense epiretinal membrane (Figure 1C). Optical coherence tomography (OCT) showed prominent, folded and disorganized retinal thickening measuring 413 μ m with an overlying epiretinal membrane (Figure 1D).

The patient was diagnosed with combined hamartoma of the retina and RPE of the right eye. The amblyopia was treated with patching. At 1-year follow-up examination, the visual acuity was 20/400 OD. Slit-lamp examination of both eyes remained normal. Fundus examination of the right eye showed a stable hamartoma of the retina and RPE. Systemic workup for NF was negative. At 3 years' follow-up, the combined hamartoma was more prominent with increased retinal vascular traction and evidence of further thickening and undulation of the involved retina on OCT (Figure 2).



Figure 1. At presentation, age 5 years, A-D. The pigmented lesion in the papillomacular region (A, B) demonstrated slight retinal vascular traction. B-scan ultrasonography showed a solid plateau lesion 2.7 mm in thickness with a suggestion of epiretinal density (C). Optical coherence tomography revealed folded, distorted retinal thickening with epiretinal membrane (D).

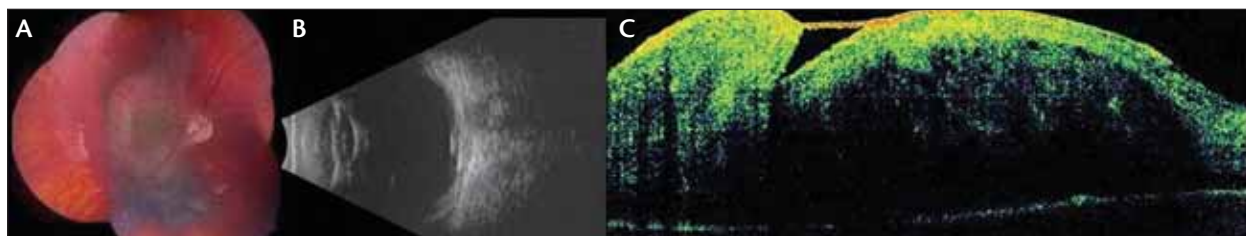


Figure 2. At 3 years' follow up, age 8 years, A-C. The lesion has progressed with more prominent retinal vascular traction (A), stable thickness (B), and more prominent retinal distortion (C).

DISCUSSION

The clinical features of combined hamartoma of the retina and RPE were initially described by Gass³ as: (1) a mildly raised, black or dark grey lesion affecting the retina, RPE and surrounding vitreous; (2) expanding toward the periphery; (3) fusing indiscriminately with neighboring RPE; (4) encapsulated by dense grey-white retinal and preretinal tissue; (5) demonstrating compression of the inner surface; (6) lacking peripheral RPE and choroidal atrophy; with (7) absence of retinal detachment, hemorrhage, exudation or vitreous inflammation. Painless vision loss and strabismus are the most frequent presenting symptoms,^{2,4} as in our patient.

The diagnosis of combined hamartoma is established through recognition of the classic clinical signs in conjunction with diagnostic features demonstrating a noncalcified plateau-shaped mass on ultrasonography and undulating, folded, thickened, and disorganized retina with photoreceptor attenuation on OCT.⁵ In a study of the OCT features of this tumor by Shields and associates,⁵ preretinal membrane was found in 10 of the 11 patients diagnosed with combined hamartoma of the retina and RPE.

Similarly, our patient presented with an epiretinal membrane and a thickened, disorganized retina. This observation concurs with previous reports in which epiretinal membrane was a prominent clinical feature and this feature was predictive of poor visual outcome.^{4,5}

The main ocular concern with combined hamartoma of the retina and RPE is its effect on visual acuity, particularly because this tumor occurs in children and can lead to blindness. In a retrospective analysis of visual prognosis in 79 eyes with combined hamartoma, it was found that macular tumor location was directly correlated with poor visual acuity.² In this comparatively large group of patients, the most common presenting symptoms were decreased vision ($n=32$, 40%) and strabismus ($n=22$, 28%). The mean tumor base size was 6.6 mm (median 6 mm, range 1.5-20 mm) for macular tumors and 8.5 mm (median 7 mm, range 3-37.5 mm) for extramacular tumors. The mean visual acuity by logarithm of the minimal angle of resolution (Snellen) for macular tumors ($n=29$, 51%) was 1.2 (20/320) on pres-

entation, compared with 0.61 (20/80) in patients with extramacular tumors ($n=28$, 49%). At 4 years' follow-up, the mean visual acuity was 1.72 (20/800) for patients with macular tumors versus 0.79 (20/125) for extramacular tumors. Vision loss of 3 Snellen lines or more occurred in 60% of patients with macular tumors compared with 13% of patients with peripheral tumors. The authors found that macular tumor location was strongly predictive of poor visual acuity.²

In conclusion, patients with combined hamartoma of the retina and RPE involving the macular region are at high risk for poor visual acuity. Detection of this condition and treatment of amblyopia, as in our case, might allow improved vision. ■

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