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CASE REPORT

Combined hamartoma of the retina and retinal pigment epithelium associated with optic coloboma

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Abstract: The authors relate an uncommon case of combined hamartoma of the retina and retinal pigment epithelium associated with optic coloboma.

Keywords: combined hamartoma of the retina and retinal pigment epithelium, benign tumor, visual loss, optic nerve coloboma

Introduction

Combined hamartoma of the retina and retinal pigment epithelium is a benign tumor that may involve pigment epithelium, and vascular and glial tissue of the retina to varying degrees.[1] It may cause significant visual loss and simulate choroidal melanoma and other intraocular tumors.[2]

Case report

The purpose of this report is to describe a rare case of combined hamartoma associated with optic coloboma in the fellow eye. A 24-year-old man presented with visual acuity of 20/20 in the right eye and 20/400 in the left eye as per the Snellen chart. In the fundus of the right eye, there were two nonpigmented nodular lesions, one placed close to the optic disc and the other in the temporal retina (Figure 1 above). In the left eye, there was a large excavation placed inside the optic disc tissue (Figure 1 below).

Fluorescein angiography showed hyperfluorescence in the right eye's lesions (Figure 1 above). The same evaluation in the left eye demonstrated optic disc coloboma (Figure 1 below). In the ocular ultrasound, the lesion close to the optic disc in the right eye was elevated and dense (Figure 2).

Discussion

Combined hamartoma of the retina and retinal pigment epithelium has been occasionally associated with systemic disease such as neurofibromatosis and tuberous sclerosis. There were various ocular abnormalities associated with combined hamartoma such as choroidal neovascularization, secondary epiretinal membrane with macular hole formation, X-linked juvenile retinoschisis, optic nerve head pits, and optic nerve head drusen.[1] Combined hamartoma are almost always a solitary, unilateral tumor, and located close to the optic disc in 76%, in the macula in 17%, and in the peripheral retina in only 7%.[3] Visual acuity varies with the location of the lesions, secondary macular distortion (epiretinal membrane, macular hole, choroidal neovascularization), and vitreous hemorrhage. The nodular lesions of posterior pole

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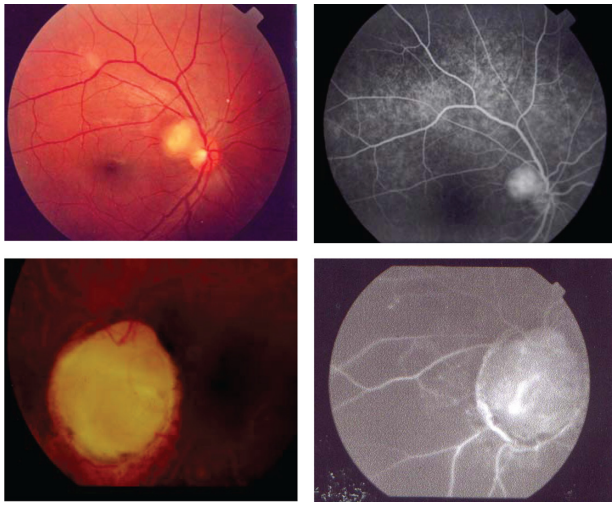

Figure 1 Color retinography – right and left eye and fluorescein angiography – late phase – right and left eye.

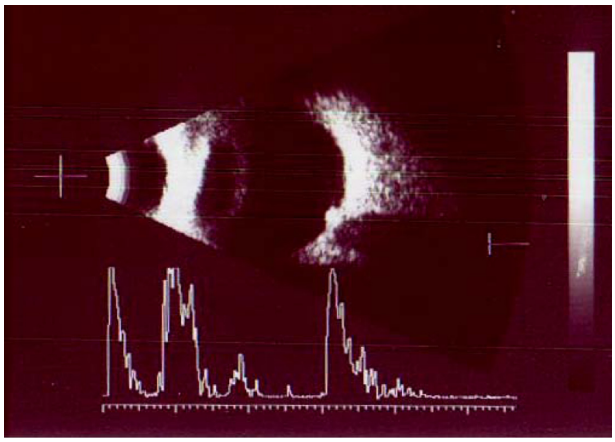

Figure 2 B ultrasound – nodular lesion – right eye.

could vary different amounts of retinal pigment, vascular tissue, and glial components. In this case, the combined hamartoma had a low pigmentation and was associated with an optic coloboma. The main reason for this association is considered incidental and makes this an uncommon case. The literature reports little manifestations involving the optic disc.[4] As recommended by the literature, the patient was observed through periodical follow-up.[5]

Disclosure

No conflicts of interest were declared in relation to this paper.

References

1. Elliot D, Schachat AP. Combined hamartoma of the retina and retinal pigment epithelium. In Ryan SR, editor. *Retina*. St. Louis, MO: Mosby Ed. 2001:640–646.
2. Schachat AP, Shields JA, Fine SL, et al. Combined hamartomas of the retina and retinal epithelium. *Ophthalmology*. 1984;91(12):1609–1615.
3. Font RL, Moura RA, Shetlar DJ, Martinez JA, Mcpherson AR. Combined hamartoma of sensory retina and retinal pigment epithelium. *Retina*. 1989;9(4):302–311.
4. Harper CA, Gole GA. Combined hamartoma of the retina and RPE: an unusual cause of the dragged disc appearance. *Aust NZ J Ophthalmol*. 1986;14(3):235–238.
5. Avitabile T, Franco L, Reibaldi M, Pulvirenti M. Combined pigment epithelial and retinal hamartomas: long-term follow-up of three cases. *Can J Ophthalmol*. 2007;42:318–320.