Stage 1 combined hamartoma of the retina and retina pigment epithelium: case report

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ABSTRACT

Combined hamartoma of the retina and retinal pigment epithelium (RPE), which was first described in 1973, is a unilateral and extremely rare malformation in which the retina, RPE and adjacent vitreous are effected. In this report, we present a 9-year-old case of Stage 1 combined hamartoma, who had previously been operated on due to strabismus and was referred to us with the preliminary diagnosis of choroidal nevus. The aim here is to highlight the early imaging findings of retina and RPE combined hamartoma, which can cause serious vision loss in childhood and can be confused with intraocular tumors, and to facilitate early diagnosis of the lesion.

Keywords: Combined hamartoma of the retina and retinal pigment epithelium, Stage 1, Optical coherence tomography**.**

INTRODUCTION

Hamartomas are benign, tumor-like lesions that occur when mature tissue elements in the area where they normally should be grow in a way that does not comply with the tissue integrity. Combined hamartoma of the retina and retinal pigment epithelium (RPE) is a unilateral and extremely rare malformation in which the retina, RPE and adjacent vitreous are effected. Although it was first described by Gass in 1973, the characteristics of the lesion could be described with the introduction of optical coherence tomography (OCT).[1] Combined hamartomas can show quite different clinical findings and can also be confused with malignant tumors such as melanoma and retinoblastoma.[1-6] Although combined hamartomas can cause unilateral vision loss and strabismus in childhood, diagnosis in this age group may be delayed due to the early stage of the lesion and imaging difficulties. In this report, we present a 9-year-old combined hamartoma case with imaging methods, who had previously been operated twice due to strabismus and was referred to us with the preliminary diagnosis of choroidal nevus. The aim of this report is to emphasize the early stage findings of retina and RPE combined hamartoma, which, although very rare, should be considered in the differential diagnosis of strabismus and amblyopia in childhood, and to facilitate early diagnosis of the lesion.

Case report

A 9-year-old male patient was referred to our clinic with poor vision and a preliminary diagnosis of choroidal nevus in the left eye. From the patient's history, it was learned that he had undergone surgery twice in total, the last of which was 1 year ago, due to strabismus on his left eye, and that he had no known systemic disease. Best-corrected visual acuity was 1.0 with +4.00 +1.00 *85 refraction in the right eye and 0.1 with +4.00 +0.50 *85 refraction in the left eye. In the cover/uncover test, exotropia of 15 prism diopters was observed in the left eye. Intraocular pressures were normal and anterior segment examination was normal in both eyes. On fundus examination, while the right eye appeared normal, a single, relatively well-circumscribed, oval, yellowish-white lesion extending from the fovea to the lower temporal arcuate was observed in the left eye (Figure 1A). In addition, it was observed that the retinal vessels near the lesion were displaced towards the center of the lesion. While no pathological staining was observed

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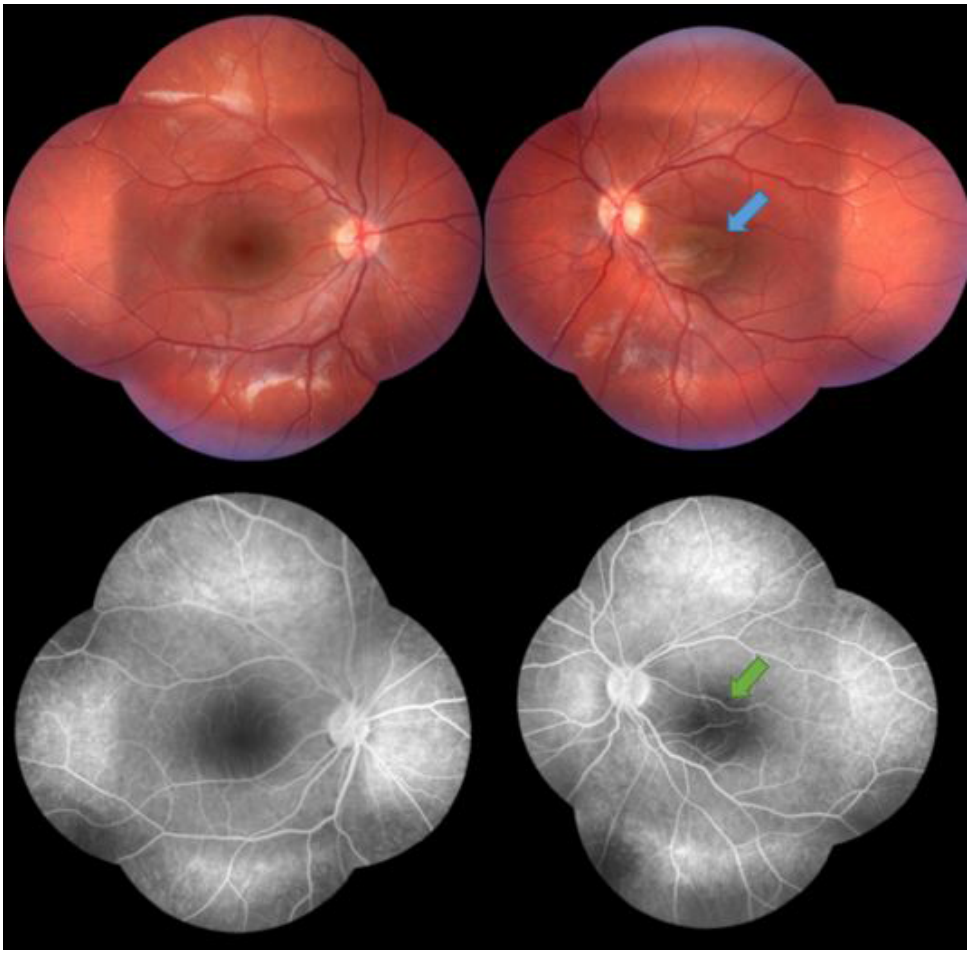

Figure 1: *A) Color fundus photographs. While the right eye looks normal, a relatively well-circumscribed, oval, yellowish-white lesion (blue arrow) extending from the fovea to the lower temporal arcuate is observed in the left eye. B) Fundus fluorescein angiography images. While the right eye looks normal, the retinal vessels near the lesion in the left eye are seen to be displaced towards the center of the lesion (green arrow).*

in the fundus fluorescein angiography, displaced retinal vessels were clearly observed (Figure 1B). In the OCT image passing through the lesion, focal vitreous adhesion, epiretinal membrane (ERM), increased hyperreflectivity in the inner retinal layers, increased thickness and irregularity in the inner retina, including the outer plexiform layer, were observed. The ellipsoid zone, external limiting membrane and RPE appeared unaffected (Figure 2). No

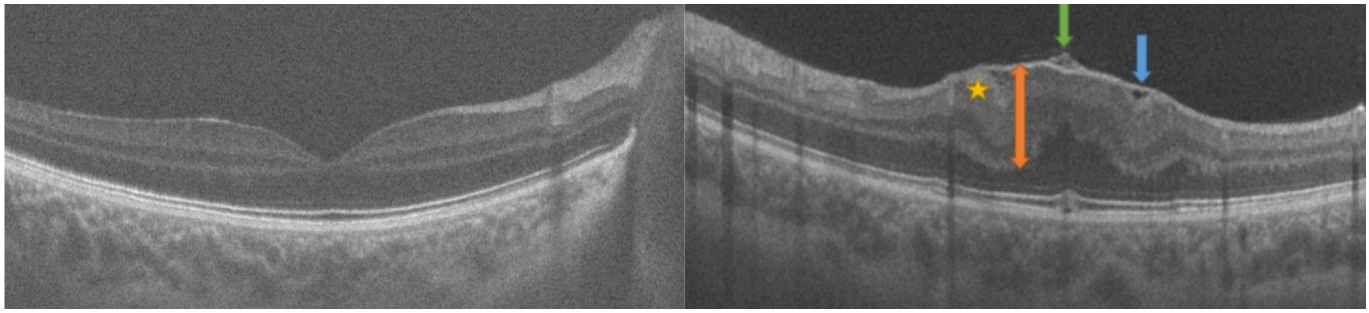

Figure 2: *While the right eye looks normal in optical coherence tomography images, in the section passing through the lesion in the left eye, focal vitreous adhesion (green arrow), epiretinal membrane formation (blue arrow), increased thickness and irregularity in the inner retina, including the outer plexiform layer (orange arrow), and increased hyperreflectivity in the inner retinal layers (star) are observed.*

choroidal nevus was observed in the examination and imaging findings. In light of all these findings, the patient was diagnosed with Stage 1B combined hamartoma of the retina and RPE according to the classification system described by Dedania et al.[7] The follow-up of the patient continues.

DISCUSSION

Although combined hamartoma of the retina and RPE is a benign lesion, both the lesion itself and its complications can cause serious vision loss. Although combined hamartomas are mostly located in the peripapillary and macula, they can also occur in the periphery.[2] Combined hamartomas, which are usually observed as a single lesion in one eye, can be bilateral in phacomotoses (neurofibromatosis).[3-6]

When the combined hamartoma of the retina and RPE was first described, it was described with common fundus findings that would help distinguish it from intraocular tumors.[1] However, with the widespread use of OCT in ophthalmological imaging, differences in the lesion have emerged. On fundus examination, the lesion is usually raised from the surface and shows varying degrees of pigmentation. As a result of contraction on the inner surface, distortion in the retinal vessels, fibrosis and ERM on the retinal surface can be observed. Vitreoretinal interface changes, ERM, disorganization of retinal layers, subretinal fluid, retinoschisis and choroidal neovascular membrane can be observed in OCT. RPE changes occur mostly in cases with optic nerve head involvement.

OCT-based staging systems have been proposed to guide the follow-up and treatment of combined hamartoma, which varies in both localization and clinical findings.[7,8] Dedania et al.'s staging system, which we used as a reference when staging the clinical and OCT findings of our case, classified the lesions both in their localization and clinical and OCT findings.[7] According to this system, lesion localizations are divided into 3 zones: (Zone 1: macular and peripapillary area; Zone 2: mid periphery; and Zone 3: far periphery), clinical findings (Stage 1: no retinal traction; Stage 2: retinal traction or retinoschisis; Stage 3: retinal detachment) and OCT findings (A: epiretinal component; B: partial retinal involvement; and C: complete retinal and RPE involvement) are also described. According to the results of this study, our case can be described as Zone 1, Stage 1B and follow-up is recommended at 2-4 month intervals.

The occurrence of combined hamartoma of the retina and RPE in childhood causes the development of severe amblyopia. Despite repeated strabismus surgeries, as in our case, functional and visual success is limited. Schachat et al. emphasized that vision improved with amblyopia treatment in only 10% of patients with combined hamartoma.[9] Surgical treatment for ERM accompanying combined hamartoma is still a matter of debate, as visual acuity does not improve despite peeling of the membrane. Additionally, it may not be possible to remove the entire ERM without damaging the retina. However, in a newly published multicenter study by Ozdek et al., it was reported that vitreoretinal surgery is effective in reducing retinal distortion and increasing visual acuity in Zone 1 patients.[10] Vitreoretinal surgery is unavoidable in case of complications such as retinal detachment and vitreous hemorrhage.

In conclusion, although it is rare, earlier diagnosis and follow-up of retina and RPE combined hamartoma, which can cause serious vision loss in childhood and can be confused with intraocular tumors, is possible thanks to increasing case reports and imaging methods.

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