

Congenital Simple Hamartoma of The Retinal Pigment Epithelium: Case Report

Retina Pigment Epitelinin Konjenital Basit Hamartomu: Olgu Sunumu

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ABSTRACT

Congenital simple hamartoma of the retina pigment epithelium (CSHRPE) is a rare benign tumor with dark black color located in macula especially in parafoveal region. The CSHRPE is frequently asymptomatic. We reported a 6 years old child who presented with a strabismus and a unilateral retinal lesion through clinical examination.

Key Words: Hamartoma, RPE, Strabismus.

ÖZ

Retina pigment epitelinin konjenital basit hamartomu (CSHRPE), makülada, sıklıkla parafoveal bölgede yerleşim gösteren, koyu siyah renkte, nadir görülen iyi huylu bir tümördür. Tümör genellikle asemptomatiktir. Bu yazıda; şaşılık nedeniyle kliniğimize başvuran 6 yaşında CSHRPE li bir çocuk olguyu sunmayı amaçladık.

Anahtar Kelimeler: Hamartom, RPE, Şaşılık.

INTRODUCTION

Congenital simple hamartoma of the retina pigment epithelium (CSHRPE) was first recognized by Laqua in 1981 and later characterized and termed by Gass in 1989.^{1,2} The CSHRPE is a rare benign tumor with dark black color which is located in macula especially in parafoveal region. Fovea is rarely affected. CSHRPE is frequently asymptomatic and diagnosis is made incidentally.³⁻⁴ We reported a 6 years old child who presented with a strabismus and a unilateral retinal lesion through clinical examination.

CASE REPORT

The patient had strabismus over one year. His visual acuity in the right eye and left eyes were 1.0 and 0.1 respectively. Ophthalmic examination showed that 8 prism diopters esotropia prominent in the left eye.

Biomicroscopic examination and intraocular pressure was normal. Retinal examination showed that there was a circumscribed black tumor in the center of fovea through posterior pole of the left eye (Figure 1). Optical coherence tomography (OCT) showed an abruptly elevated hyper-reflective mass with deep optical shadowing (Figure 2). Fundus autofluorescence demonstrated hypo autofluorescence.³ Fluorescein angiography showed blockage of background fluorescence due to the pigmented lesion (Figure 4-5). The diagnosis was compatible with CSHRPE, and the patient was followed by further observation.

DISCUSSION

Congenital simple hamartoma of the retina pigment epithelium is a rare benign tumor with dark black color which located in macula especially in parafoveal region.

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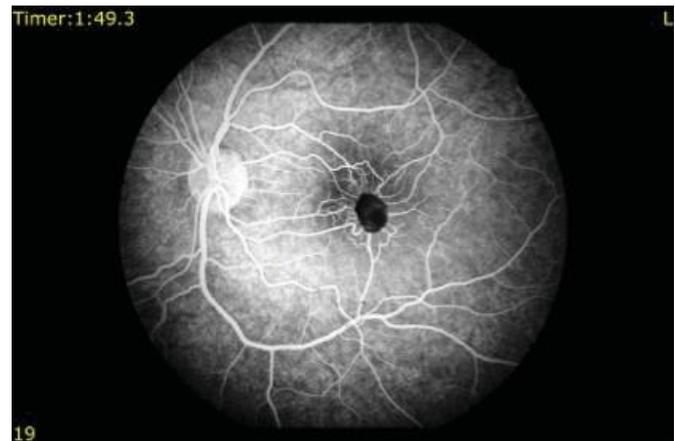
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Figure 1.



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Figure 4.

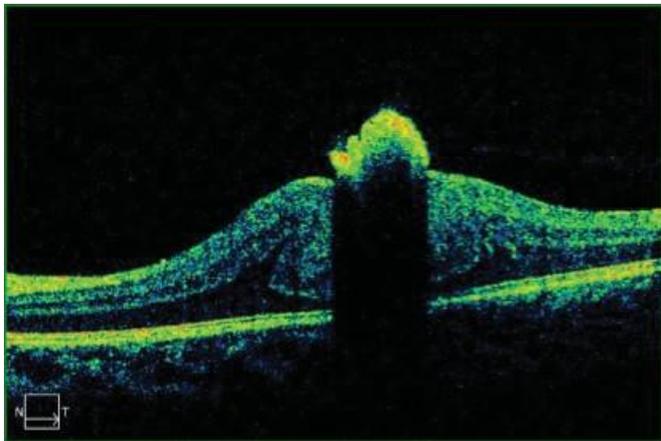
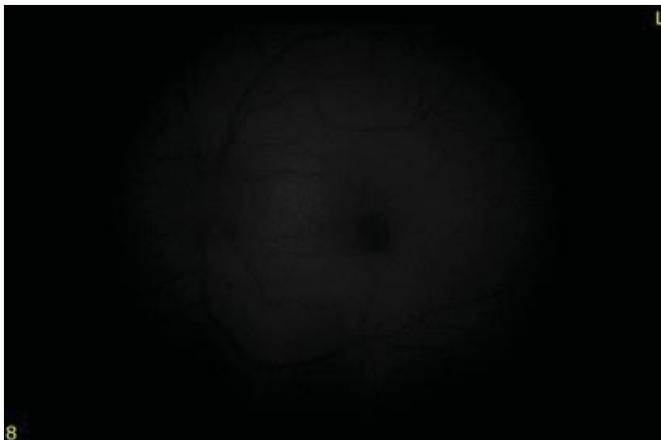


Figure 2.



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Figure 5.

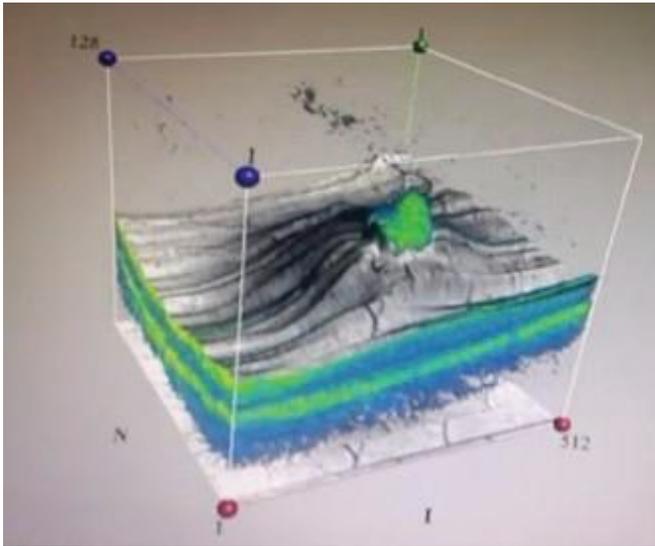


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Figure 3.

CSHRPE is thought to be congenital and non-progressive.⁵ The fact that pediatric cases have been reported in the literature supports this idea. However CSHRPE is frequently asymptomatic and diagnosis is made incidentally at advanced age. Differential diagnoses include the following: combined hamartoma of the retina and RPE, congenital hypertrophy of the RPE, adenoma or adenocarcinoma of the

RPE, RPE hyperplasia, intraretinal foreign body, and retinal invasion from an underlying choroidal nevus (melanocytoma) or choroidal melanoma.⁶ Although FA findings were unremarkable in the present case, hyperfluorescence is occasionally observed in the late phases.¹⁻² OCT typically shows a complete blockage of optical transmission.⁷ In our case optical coherence tomography showed that an abruptly elevated hyper-reflective mass with deep optical shadowing. Fluorescein angiography showed blockage of background fluorescence due to the pigmented lesion. Fundus autofluorescence demonstrated hypoautofluorescence. OCT is most important tool for differential diagnosis. We diagnosed the vitreous protrusion and tumor in our patient with 3D imaging in addition to SD-OCT (Figure 6).

Since the tumor is asymptomatic and non-progressive, no treatment is necessary in general. However, few cases which have been treated are reported in the literature. Barns et al diagnosed CSHRPE at 66-years old patient with vitreomacular traction and treated him with surgery. They demonstrated the tumor with histopathological examination.⁸ The lesion showed a nodular proliferation



Picture 6.

of hyper- plastic RPE cells with attached gliotic retina and internal limiting membrane. Bach et al. diagnosed CSHRPE at 14 years old patient with macular edema and treated him with intravitreal bevacizumab. We did not observed any findings of vitreomacular traction, macular edema or epiretinal membrane.⁹ In a case reported by Arumi et al. excision was performed to exclude retinal metastasis of uveal melanoma and treated amblyopia. They demonstrated the tumor with histopathological examination. No significant increase in visual acuity was observed during patient's clinical follow- ups.¹⁰ Retina and retina pigment epithelium invasion was demonstrated with histopathological examination since the study by Arumi et al. did not use OCT. However, we easily detected this distinction using OCT.

Currently, treatment of this rarely found lesion is a controversial issue. According to the literature, cases with asymptomatic and parafoveal tumor does not need to be treated but they need to be closely followed. Visual loss secondary to epiretinal membrane, vitreomacular traction, macular edema are only treated. Solely, Arumi et al diagnosed and treated a tumor surgically. Our patient has newly been diagnosed and there was no finding for secondary visual loss due to reasons like macular edema or macular traction. Visual loss was due to mass effect of the tumor. We decided to observe the case for progression of the tumor, amblyopia and strabismus. More cases are needed for the development of treatment modalities.

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