Congenital Simple Hamartoma of The Retinal Pigment Epithelium: Case Report

Retina Pigment Epitelinin Konjenital Basit Hamartomu: Olgu Sunumu

Sabiha GÜNGÖR KOBAT[1], Fatih Cem GÜL[1], Burak TURGUT[2]

ABSTRACT

Congenital simple hamartoma of the retina pigment epithelium (CSHRPE) is a rare benign tumor which is dark black color and located in macula especially in parafoveal region. CSHRPE is frequently asymptomatic. We reported the case of a 6 years old child who was 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 named by Gass in 1989.[1-2] CSHRPE is a rare benign tumor which is dark black color and located in macula especially in parafoveal region. Fovea is rarely affected. CSHRPE is frequently asymptomatic and diagnosis is detected incidentally.[3-4] We reported the case of a 6 years old child who was presented with a strabismus and a unilateral retinal lesion through clinical examination.

CASE REPORT

The patient had strabismus for the last one-year period. His visual acuity in the right eye and left eye were 1.0 and 0.1 respectively. Ophthalmic examination showed that 8 prism diopters esotropia especially dominant in the left eye. Biomicroscopic examination and intraocular pressure was normal. Retinal examination showed that a circumscribed black tumor was 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 which is dark black color and located in macula especially in parafoveal region.

1- Uz. Dr., Elazığ Sağlık Bilimleri Üniversitesi, Göz Hastalıkları, Elazığ, Türkiye

2- Prof. Dr., Onsekiz Mart Üniversitesi Tıp Fakültesi, Göz Hastalıkları, Çanakkale, Türkiye

Geliş Tarihi - Received: 03.10.2018
Kabul Tarihi - Accepted: 29.11.2018
Ret-Vit 2019; 28: 410-412

Yazışma Adresi / Correspondence Adress:
Sabiha GÜNGÖR KOBAT
Elazığ Sağlık Bilimleri Üniversitesi, Göz Hastalıkları, Elazığ, Türkiye

Phone:
E-mail: drsabihag@gmail.com

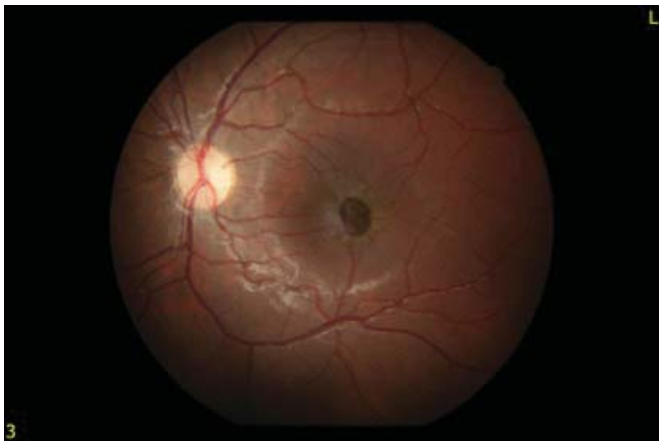


Figure 1.

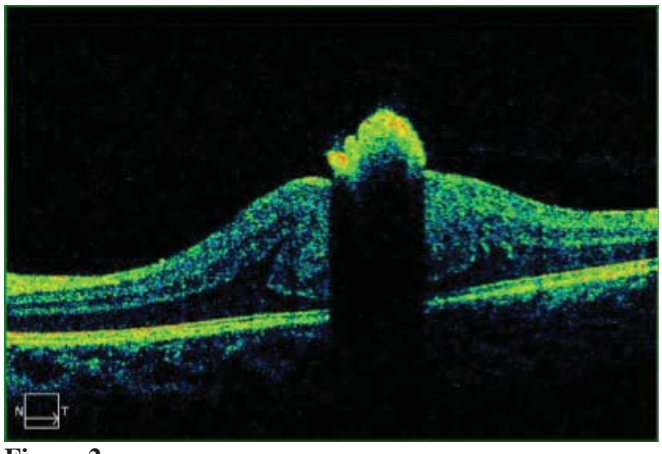


Figure 2.

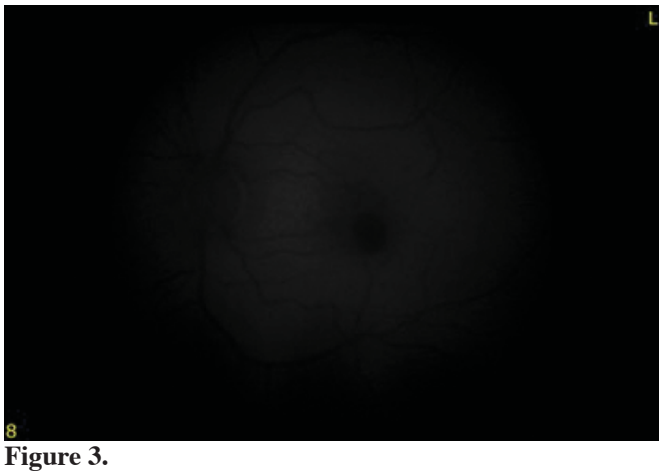


Figure 3.

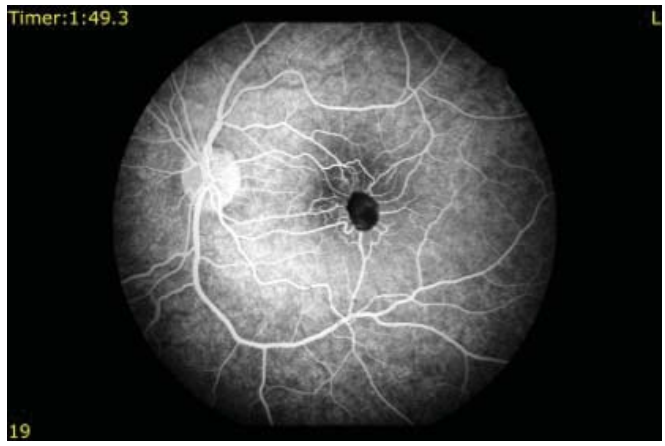


Figure 4.

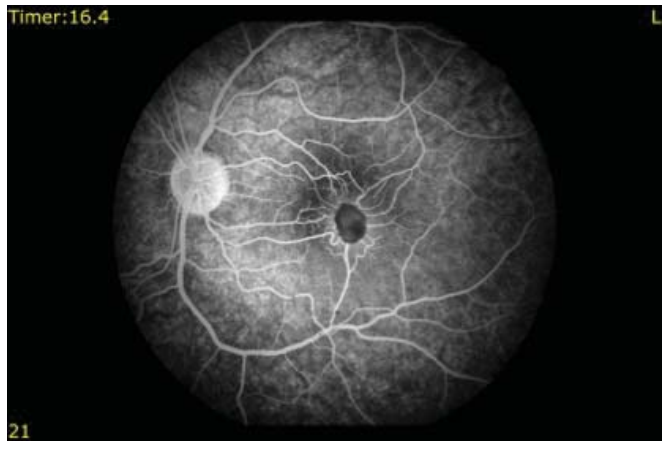
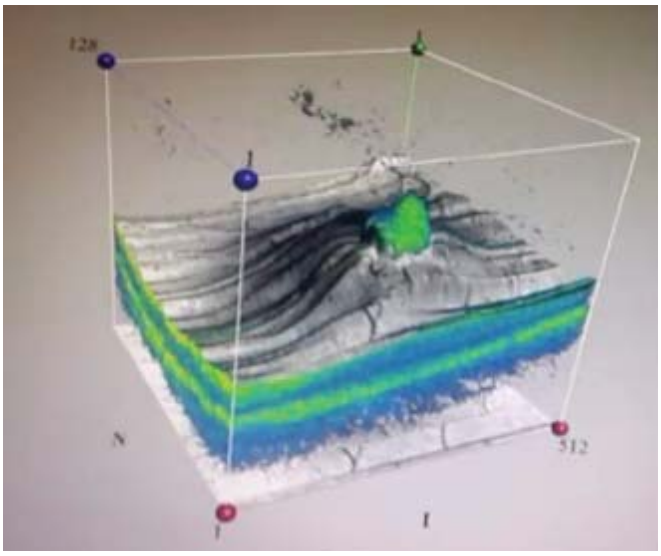


Figure 5.

CSHRPE is thought to be congenital and non-progressive.[5] The fact that child cases have been reported in the literature supports this idea. However CSHRPE is frequently asymptomatic , diagnosis is detected 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, hyperplasia of the RPE, intraretinally foreign body, and retinal invasion from an underlying choroidal nevus (melanocytoma) or choroidal melanoma.[6] Although FA findings were unremarkable in the present case, hyper fluorescence 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 hypo autofluorescence. OCT is most important examination 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 nonprogressive, treatment is usually not necessary. However, few cases which have been treated are reported in the literature. Barns at 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



Resim 6.

of hyper- plastic RPE cells with attached gliotic retina and internal limiting membrane. Bach at al diagnosed CSHRPE at 14 years old patient with macular edema and treated him with intravitreal bevacizumab. We did not have any findings about 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 Arumi et al.'s study 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 seconder 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.

REFERENCES / KAYNAKLAR

1. Laqua H. Tumors and tumor-like lesions of the retinal pigment epithelium. Ophthalmologica. 1981;183(1):34–8.
2. Gass JD. Focal congenital anomalies of the retinal pigment epithe- lium. Eye. 1989;3(1):1–18.
3. Yannuzzi LA. Congenital simple hamartoma of the retinal pigment epithelium. In: Yanuzzi LA, editor. The Retinal Atlas. Beijing, China: Saunders; 2010. pp. 650–63.
4. Shields CL, Shields JA, Marr BP, Sperber DE, Gass JD. Congenital simple hamartoma of the retinal pigment epithelium: A study of five cases. Ophthalmology. 2003;110:1005–11.
5. Souissi K, El Afrit MA, Kraiem A. J Pediatr Ophthalmol Strabis- mus. 2006 May-Jun;43(3):181-2. Congenital retinal arterial mac- rovessel and congenital hamartoma of the retinal pigment epitheli- um.
6. Shields JA, Shields CL. Intraocular tumors. In: Shields JA, Shields CL. An Atlas and Textbook. Tumors and related lesions of the pigment epithelium. Philadelphia: Lippincott Williams&Wilkins; 2008:432-9.
7. Shukla D, Ambatkar S, Jethani J, Kim R. Optical coherence tomography in presumed congenital simple hamartoma of retinal pigment epithelium. Am J Ophthalmol. 2005;139:945-7.
8. C Barnes , DR Goldman2, NV Laver ,JS Duker. Congenital simple hamartoma of the retinal pigment epithelium: clinical, optical coherence tomography, and histopathological correlation .Eye (2014) 28, 765–6.
9. Bach A1, Gold AS, Villegas VM, Wildner AC, Latiff A, Ehlies FJ, Murray TG. Simple hamartoma of the retinal pigment epithelium with macular edema. Optom Vis Sci. 2015 Apr;92(4 Suppl 1):S48- 50.
10. Garcia-Arumi J, Sararols L, Freixes S, et al.Surgical Resection of a retinal pigment epithelial hamartoma. Arch Ophthalmol. 2004;122(12):1870-2.