Clinical and Optical Coherence Tomography Characteristics of Combined Hamartoma of the Retina and Retinal Pigment Epithelium

Retina ve Retinal Pigment Epitelin Kombine Hamartomunun Klinik ve Optik Koherens Tomografideki Karakteristik Özellikleri

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Summary
Combined hamartoma of the retina and retinal pigment epithelium (CHRRPE) is a rare and benign retinal lesion. Because it may be misdiagnosed as a malignant melanoma of the choroid or retinoblastoma, the differential diagnosis is very important. In this study, clinical and optical coherence tomographic (OCT) characteristics of four cases with CHRRPE are presented and the differential diagnosis is discussed. In all of the eyes, a grey, elevated retinal lesion was detected clinically. Also OCT showed hyper-reflecting retinal elevation with underlying hypo-reflecting shadow due to the dense fibrous tissue or epiretinal membrane. It was concluded that OCT findings could support the clinical observations in the diagnosis of CHRRPE. (Turk J Ophthalmol 2013; 43: 353-7)

Key Words: Hamartoma, retina, retinal pigment epithelium, optic coherence tomography (OCT), differential diagnosis

Özet

Anahtar Kelimeler: Hamartom, retina, retina pigment epiteli, optik koherens tomografi (OKT), ayırıcı tanı

Introduction
Combined hamartoma of retina and retinal pigment epithelium (CHRRPE), which was first introduced by Gass in 1973, is a rare benign tumor characterized by the proliferation of retinal pigment epithelium (RPE) and glial tissue. It generates a large papillary and retinal distortion and frequently misdiagnosed as chorioidal melanoma of retinoblastoma. The typical clinical symptom of the disease is a painless vision acuity loss and epiretinal membrane or chorioidal neovascular membranes (CNVM) may accompany the clinical picture. Accurate diagnosis is very important because of its differential diagnosis like malignant ocular lesions and optical coherence tomography (OCT) is very useful diagnostic tool for confirming the diagnosis.

Herein, we report our clinical observations in addition to spectral-domain OCT (Spectralis, Heidelberg engineering, Germany) findings of four eyes of four cases with CHRRPE at the time of diagnosis of the disease and discuss the importance of OCT.

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Case 1

A 17-year-old male healthy case was referred to our hospital for decreased visual acuity (VA) in his right eye for 1 year. The VA was counting fingers at 4 meters in the right eye and 20/20 in the left eye. Slit-lamp examination revealed normal anterior segment findings in both eyes. Fundoscopy revealed normal findings in the left eye but a grey, elevated retinal lesion extending from temporal retina to macula, increased retinal venous tortuosity and epiretinal membrane (ERM) over the macula were seen in the right eye (Figure 1). FA showed increased retinal venous tortuosity in venous phase (Figure 2). Horizontal section of OCT showed hyper-reflecting retinal surface with underlying hypo-reflecting shadow due to the dense fibrous tissue and a hyper-reflecting band due to ERM. Serous retinal detachment wasn’t observed (Figure 3).

Case 2

A 9-year-old healthy boy was referred to our hospital for low VA in his left eye since she was born. He was misdiagnosed several times as retinoblastoma or astrocytoma according to his medical story. The VA was 20/20 in the right eye and counting fingers at 3 meters in the left eye. Slit-lamp examination revealed normal anterior segment findings in both eyes. Her right eye had normal posterior segment. Fundoscopy of the left eye revealed a grey, elevated retinal lesion over entire macular area with prominent retinal venous tortuosity (Figure 4). Horizontal section of OCT showed hyper-reflecting retinal surface with underlying hypo-reflecting shadow due to the dense fibrous tissue. Serous retinal detachment wasn’t observed but hypo-reflecting shadow under the fibrous tissue gave a false impression of serous effusion (Figure 5).

Case 3

An 11-year-old healthy girl was referred to our hospital for decreased VA in her left eye for three years. The VA was 20/20 in the right eye and 20/80 in the left eye. Slit-lamp examination revealed normal anterior segment findings in both
eyes. Fundoscopy revealed normal findings in the right eye but a grey, elevated macular lesion which generated macular displacement was seen in the left eye (Figure 6). Oblique section of OCT over the lesion showed hyper-reflecting retinal surface with no serous retinal detachment (Figure 7).

Case 4

A 24-year-old healthy male case was referred to our hospital for decreased visual acuity (VA) in his left eye for over 10 years. The VA was 20/20 in the right eye and counting fingers at 3 meters in the left eye. Slit-lamp examination revealed normal anterior segment findings in both eyes. Fundoscopy revealed normal findings in the right eye. A grey elevated giant retinal lesion retinal lesion over entire macular and peripapillary area which generated retinal venous tortuosity and macular ectopia was seen in the left eye (Figure 8). Prominent ERM was seen over the entire lesion including peripapillary and macular area. FA showed increased retinal venous tortuosity in venous phase and edema because of increased vascular permeability due to the traction of ERM (Figure 9).

Horizontal section of OCT showed hyper-reflecting retinal surface with underlying hypo-reflecting shadow due to the dense fibrous tissue and a hyper-reflecting band due to ERM. Hyper-reflecting ERM was seen to extend from the surface of the lesion to optic disc and serous retinal detachment wasn’t observed (Figure 10).

Figure 5. Macular section of OCT of the second case shows hyper-reflecting retinal surface due to the dense fibrous tissue especially in the foveal area. A hypo-reflecting shadow under the fibrous tissue gave a false impression of serous effusion.

Figure 6. Color fundus photography of the left eye of the third case shows a grey, elevated macular lesion which generated macular displacement.

Figure 7. Oblique section of OCT of the third case over the lesion shows mild hyper-reflecting retinal surface with no serous retinal detachment.

Figure 8. Color fundus photography of the left eye of the fourth case shows a grey retinal lesion over entire macular and peripapillary area which causes macular ectopia.

Figure 9. FA of the fourth case shows increased retinal venous tortuosity in venous phase and edema due to the traction of ERM.
All of the ocular findings and characteristic properties for 4 cases are shown in Table 1.

**Discussion**

OCT is a noninvasive, noncontact technique which provides high-resolution retinal images and has been widely used in the diagnosis of retinal diseases. In our study, our aim was to report and discuss the OCT findings of our cases with CHRRPE. The morphology of the lesion differs according to the type and amount of its contents like RPE, sensory retina, retinal vessels or vitreous and this may lead to difficulties in the differential diagnosis. Also our second case was misdiagnosed several times.

Shields et al. reviewed the medical records of their 77 cases with CHRRPE. Decreased VA was reported to be the most frequent presenting symptom of the disease among the others like strabismus or irritation. The main causes of decreased VA are the direct involvement of fovea, optic disc or papilla-macular bundle, macular traction by ERM and subretinal and intraretinal exudation. As a result CHRRPE is a benign tumor but its differential diagnosis has vital importance in order to avoid unnecessary enucleation or radiation therapy. Another very important point is the mood of the mostly young patients who had misdiagnosed as a malignant tumor and lost their eyes. This report emphasizes

*Figure 10. Horizontal section of OCT shows prominent ERM on disc and retina and a hypo-reflecting shadow due to the gliosis within and over the retina*
the importance of OCT in addition to careful ophthalmic examination in the diagnosis of CHRRPE. Also it can help the clinicians not only in diagnosis and follow-up but also in planning their cases treatments by detecting the onset of associated complications like ERM, CNVM or retinal detachment.

References