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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 coherens tomography (OCT), differential diagnosis

Özet

Kombine retina ve retina pigment epitel hamartomu (KRRPEH), nadir rastlanılan iyi huylu retinal lezyondur. Koroid Malign Melonomu veya retinoblastom gibi tanı alabileceği için, ayırıcı tanı çok önemlidir. Bu çalışmada, KRRPEH olan 4 olgu, klinik ve optik koherens tomografideki (OKT) karakteristik özellikleri ile sunulmuş, ve ayırıcı tanı tartışılmıştır. Tüm gözlerde, klinik olarak gri eleve retinal lezyon tespit edildi. Ayrıca, OKT'de, yoğun fibröz doku veya epiretinal membran nedeni ile yüksek reflektivite gösteren retinal kabarıklık altında uzanan düşük reflektivite gölgelenmesi görüldü. Bu OKT bulguları ile, klinik gözlemde KRRPEH tanısını destekleyebilir sonucuna varılmıştır. *(Turk J Ophthalmol 2013; 43: 353-7)*

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[1] in 1973, is a rare benign tumor characterized by the proliferation of retinal pigment epithelium (RPE) and glial tissue.[2] It generates a large papillary and retinal distortion and frequently misdiagnosed as choroidal melanoma of retinoblastoma. The typical clinical symptom of the disease is a painless vision acuity loss and epiretinal membrane or choroidal neovascular membranes (CNVM) may accompany the clinical picture.[3] 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.[4-7]

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).

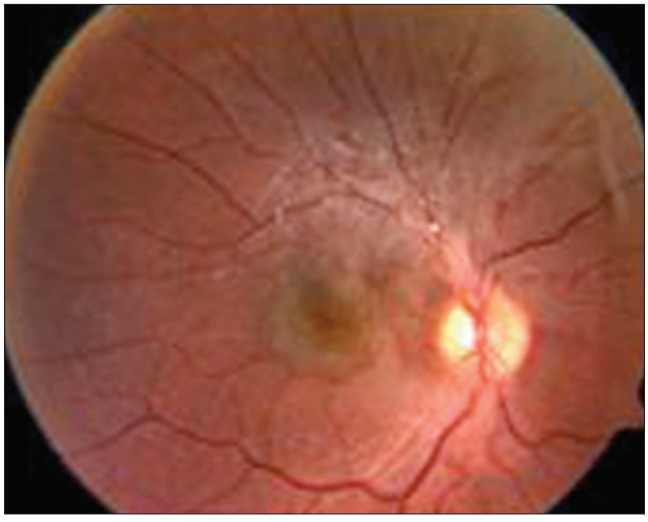

Figure 1. Color fundus photography of the right eye of the first case shows grey elevated mass which extending from temporal retina to macula, tortuous retinal vessels and ERM over the lesion

Figure 2. FA shows of the right eye of the first case increased retinal venous tortuosity in venous phase

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 which generated 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

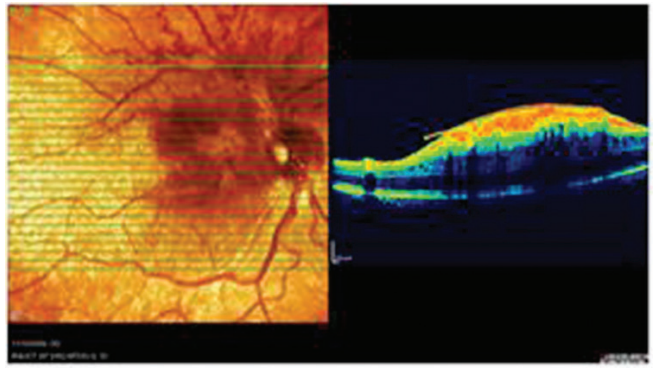

Figure 3. Horizontal section of OCT shows hyper-reflecting retinal surface with underlying hypo-reflecting shadow due to the dense fibrous tissue and a hyper-reflecting band due to ERM

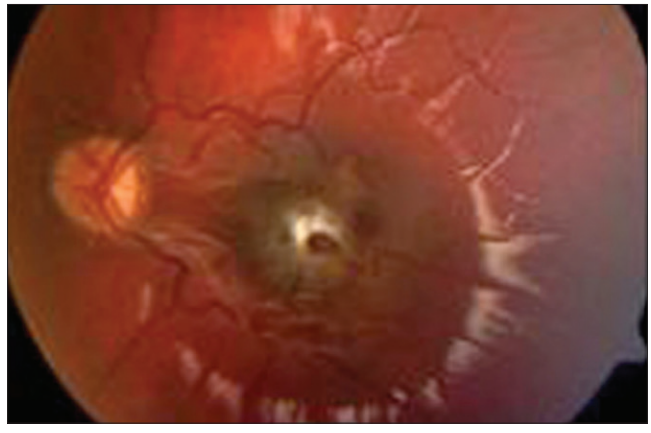

Figure 4. Color fundus photography of the left eye of the second case shows a grey, elevated retinal lesion over entire macular area with prominent retinal venous tortuosity

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).

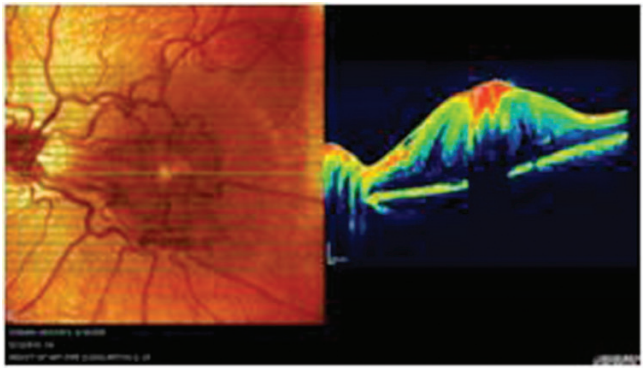

Figure 5. Macular section of OCT of the second case shows hyper-reflecting retinal surface due to the dense fibrous tissue especially in the foveal area. A hypo-reflecting shadow under the fibrous tissue gave a false impression of serous effusion

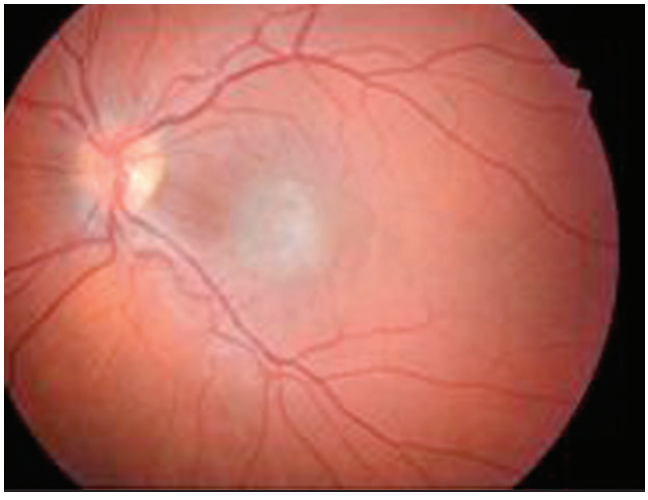

Figure 6. Color fundus photography of the left eye of the third case shows a grey, elevated macular lesion which generated macular displacement

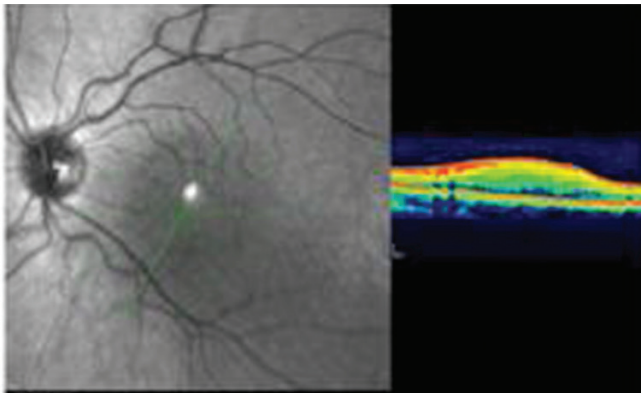

Figure 7. Oblique section of OCT of the third case over the lesion shows mild hyper-reflecting retinal surface with no serous retinal detachment

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).

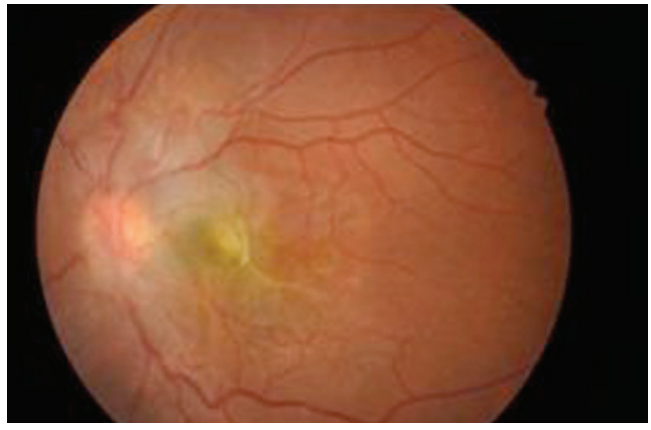

Figure 8. Color fundus photography of the left eye of the fourth case shows a grey retinal lesion over entire macular and peripapillary area which causes macular ectopia

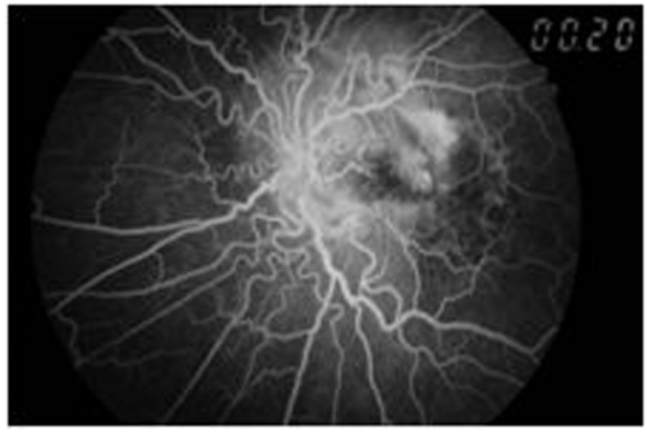

Figure 9. FA of the fourth case shows increased retinal venous tortuosity in venous phase and edema due to the traction of ERM

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.[3] 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 from the vessels inside the lesion.7 None of our cases had exudative retinal detachment and CNVM while 13% of cases had serous effusion and 6% of cases had CNVM in Shields et al.[3] study. Our first, second and fourth cases also had retinal vessel tortuosity, which was a frequent finding in CHRRPE. FA showed increased retinal venous tortuosity in venous phase in the first and second cases in addition to macular edema in the fourth case.

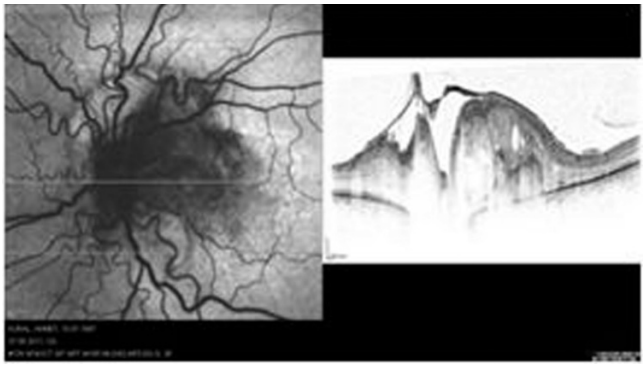

Figure 10. Horizontal section of OCT shows prominent ERM on disc and retina and a hypo-reflecting shadow due to the gliosis within and over the retina

Table 1. Characteristic properties and ocular findings of 4 cases

	Case 1	Case 2	Case 3	Case 4
Age (year)	17	9	11	24
Sex(F/M)	M	M	F	M
Time (year)	1	9	3	10
VA (R/L)	4mps/1.0	1.0/3mps	1.0 / 20/80	1.0/3mps
Biomicroscopy	Grey elevated RL,VT	Grey elevated RL, VT	Grey elevated ML, MD	Grey elevated Giant RL, ME
Localization	Temporal to macula	Macular area	Macular area	Macular area peripapillary area
ERM	+	--	--	+

F/M: Female/Male, R/L: Right/Left, RL: Retinal lesion, ML: Macular lesion, VT: Venous Tortuosity, MD : Macular Displacement, MA: Macular Area

Gass1 divided the cases with CHRRPE into 4 groups according to the location of the lesions as macular, peripheral, papillary or juxtapapillary once. Our first case had juxtapapillary, second case had macular, but third and fourth cases had almost macular lesions.

OCT is a very important for accurate diagnosis especially in the differential diagnosis. Shields et al.[4] examined 11 consecutive patients who had CHRRPE by using OCT. OCT showed retinal mass with hyper reflective surface and deep shadowing, retinal disorganization, ERM with secondary retinal folds and striae, retinal traction and/or macular edema. Huot et al.[5] presented a 17-year-old girl with CHRRPE. OCT showed hyper reflective ERM with traction and retinal folds with distortion of the foveal architecture. In all our cases, OCT showed hyper reflectivity due to ERM and/or fibrotic tissue and deep hypo reflective shadowing. We observed that the adjacent retina was normal in architecture. Ting et al.[6] also observed that, the adjacent retina had appeared to be normal and separate from the mass in their cases. Although our first, second and fourth cases had distinct hyper reflectivity; it wasn't prominent in the third case. This result was thought to be related with the contents of the lesion.

Fundoscopy generally reveals similar findings in CHRRPE and choroidal melanoma. Some cases were reported who had undergone enucleation because of prediagnosis of choroidal melanoma but had pathological diagnosis of CHRRPE later.[8-9] Only 3% of cases CHRRPE were reported to get an accurate diagnosis in the first visit, by their first ophthalmologists.[10] OCT is very important to distinguish these two diseases. OCT mostly shows normal retinal structure in melanoma while hyper reflectivity in inner retinal layers and hypo reflective shadowing in outer retinal layers are observed in CHRRPE. The locations of CHRRPE and choroidal melanoma are thought to cause these different OCT findings.[7] In fact, there are some clinical hints to differentiate these two entities. Retinal detachment, subretinal hemorrhage, vitreous hemorrhage and glaucoma are frequently associated with choroidal melanoma but almost never seen in CHRRPE. Furthermore retinal vessel tortuosity is one of the most important sign in CHRRPE but is not seen in choroidal melanoma.[7] Also some other diseases like choroidal nevus, optic disc melanocytoma, RPE adenoma and RPE hypertrophy should be kept in mind in the differential diagnosis.

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

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.

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