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OPHTHALMOLOGY
Case Report

Congenital Hypertrophy of the Retinal Pigment Epithelium in Two Different Presentations

Kader Kasar[1], Sabiha Güngör Kobat[2] and Burak Turgut[3]*

[1]*Bayrampaşa State Hospital, Ophthalmology Clinic, Turkey*

[2]*Elazig Education and Research Hospital, Ophthalmology Clinic, Turkey*

[3]*Department of Ophthalmology, School of medicine, Firat University, Turkey*

***Corresponding Author:** Burak Turgut, Department of Ophthalmology, School of medicine, Firat University, 23119, Elazig, Turkey.

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Abstract

We purpose to report two cases of congenital hypertrophy of the retinal pigment epithelium in two different presentations in this report. The patients included a woman, who is 38 years old and a man, who is 42 years old. Both patients were admitted to our outpatient clinic for correction of their refractive errors and their best corrected visual acuities are 20/20 in both eyes. Intraocular pressures of both patients were normal range in both eyes. Slit-lamp biomicroscopy revealed no pathological anterior segment sign. In both patients, congenital hypertrophy of the retinal pigment epithelium was unilateral. Fundus examination of Case 1 revealed flat, grouped, well-demarcated, oval and black spots at the superotemporal macula and the next of temporal macula in the left eye. In the fundus examination of Case 2, it was observed a single well-demarcated, oval, hyperpigmented lesion with three optic disk diameters at the lower nasal midperipheral retina in the right eye. As both patients have no multiple and bilateral lesions, they were not needed to subject to gastroenterology clinic for the presence of familial adenomatous polyposis or any gastrointestinal malignancy. Solitary, unilateral lesions and grouped CHRPE are restricted to the RPE with no other ocular or extracellular findings. Multiple or bilateral CHRPE may be associated with autosomal dominant FAP. However, in all lesions, but especially the atypical lesions of CHRPE should be monitored with retinal photographic documentation at regular follow-ups.

Keywords: *Congenital hypertrophy of the retinal pigment epithelium; solitary; bear-track; familial adenomatous polyposis*

Abbreviations: CHRPE: Congenital hypertrophy of the retinal pigment epithelium; RPE: retinal pigment epithelium; FAP: familial adenomatous polyposis; FFA: fundus florescein angiography

Introduction

Congenital hypertrophy of the retinal pigment epithelium (CHRPE) is a congenital, well-demarcated, hyperpigmented lesion surrounded hypo pigmented ring at the level of RPE [1-4]. CHRPE occurs in three different forms: either as solitary or grouped or multiple pigmented fundus lesions. The patients are usually asymptomatic and the lesions are observed during routine ophthalmoscopy [1]. Solitary CHRPE is typically a flat, round, hyperpigmented lesion with smooth or scalloped margins that is well-demarcated from normal-appearing retinal pigment epithelium (RPE). When several lesions of varying size are arranged in a cluster, resembling the footprint of an animal ('bear tracks'), they are named "grouped CHRPE". Grouped lesions are flat, well-demarcated round oval or geographic black spots with increasing size towards the fundus periphery. Multiple CHRPE lesions in familial adenomatous polyposis (FAP) are generally smaller (50-100 µm in diameter) compared with solitary CHRPE. They are black, brown, or light gray. Larger lesions may be surrounded by a depigmented halo, mottled RPE; window-defect- type changes adjacent to retinal vessels, may contain depigmented lacunae, and can be accompanied by small pigmented satellite lesions [2, 3]. Solitary, unilateral lesions and grouped CHRPE are restricted to the RPE with no other ocular or extracellular findings. Multiple or bilateral CHRPE may be associated with autosomal dominant FAP [4-7].

We aimed to report two cases of congenital hypertrophy of the retinal pigment epithelium in two different presentations.

Case Report

Case 1

A female patient, who is 38 years old, was admitted to our outpatient clinic for routine eye examination and correction of her refractive errors. She had no history remarkable of systemic or ocular disease. Ocular history of her family was also unremarkable. Her best corrected visual acuity is 20/20 in both eyes. Intraocular pressures of both eyes were 14 mmHg. Slit-lamp biomicroscopy revealed no pathological anterior segment sign. Fundus examination was normal on the right eye. Fundus examination of the left eye revealed flat, grouped, well-demarcated, oval, hyperpigmented lesions (bear tracks) which are located at the superotemporal macula and the next of temporal macula (Figure 1). In early and late stages of the fundus florescein angiography (FFA) (Zeiss SS 450 plus IR), multiphocal hypo florescence in the regions of lesion due to the blockade of florescein was observed (Figure 2). As the patient has no an atypical and bilateral lesion, she was not needed to subject to gastroenterology clinic for the presence of any FAP or any gastrointestinal malignancy.

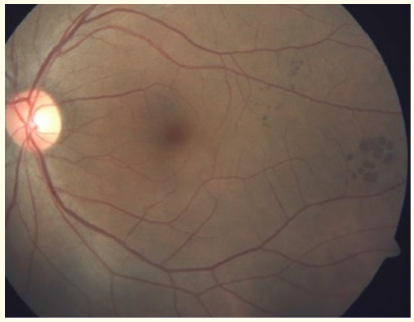

Figure 1: *Colour image showing bear tracks at the next of superotemporal and temporal macula in Case 1.*

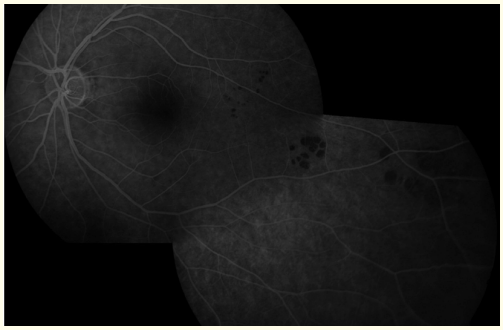

Figure 2: *The fundus fluoresce in angiography image showing multifocal hypo fluorescence due to lesions in Case 1.*

Case 2

A male patient, who is 42 years old, was admitted to our outpatient clinic for routine eye examination. He had no history remarkable of systemic or ocular disease. His family ocular history was also unremarkable. His best corrected visual acuity is 20/20 in both eyes. Intraocular pressures of both eyes were 16 mmHg. Slit-lamp biomicroscopy revealed no pathological anterior segment sign. Fundus examination of the right eye revealed a single well-demarcated, oval, hyperpigmented lesion with three optic disk diameters at the lower nasal midperipheral retina (Figure 3). Fundus examination was normal on the left eye. FFA was recommended for the patient; however,

he did not accept it. As the patient has no an atypical and bilateral lesion, he was not needed to subject to gastroenterology clinic for the presence of any FAP or gastrointestinal malignancy.

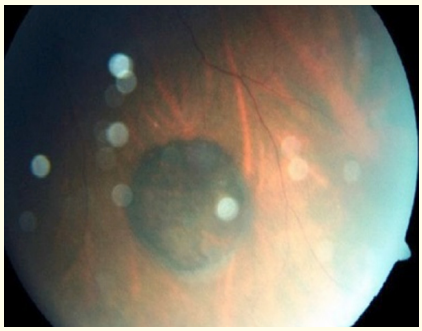

Figure 3: *Colour image showing shows solitary hyper pigmented lesion with depigmented halo in Case 2.*

Discussion

Congenital hypertrophy of the retinal pigment epithelium was described firstly Blair and Trempe in 1980 as congenital [5], well-demarcated, hyperpigmented lesion surrounded hypo pigmented ring at the level of the RPE. CHRPE usually presents before 30 years of age. CHRPE lesions are often incidentally discovered on dilated routine ophthalmoscopic examination because they are usually asymptomatic and, most commonly located at mid-peripheral or peripheral retina. However, it may rarely involve the posterior pole and the macula [3,5,6]. CHRPE occurs in three variant forms: either as solitary or grouped or multiple pigmented fundus lesions. Multiple CHRPE may be associated with FAP, an autosomal dominant disease with numerous adenomatous polyps of the colon and rectum. The most common form of disease is solitary CHRPE. It often seems as a unilateral, single, dark -gray/black or brown in colour, flat, pigmented, round or oval lesion with well-demarcated borders usually located near the equator. The presence of depigmented lacunae in lesion is a common finding. Juxtapapillary or juxtamacular lesion is not common. Solitary CHRPE is usually more commonly seen in the temporal rather than nasal retina. Grouped CHRPE consist multiple, small, brown-black in colour lesions organized in a pattern simulating animal footprints (bear-tracks). Bear tracks are typically larger in the periphery and decrease in size towards the optic disc. If these lesions are depigmented, they are called as ''polar bear tracks'' [3-7].

The features of multiple lesions include, especially, bilaterality, multiplicity (number than three, largeness (diameter with more 0.5 of the optic disk diameter), oval or fusiform/spindle-shaped lesions of variable size associated with hypo pigmentation at one margin and widely separated localization. Multiple lesions have systemic associations, including some familial colonic adenomatous polyposis syndromes such as Gardner's and Turcott Syndrome [5-12]. FAP is an autosomal inherited disease characterized by multiple gastrointestinal polyps. When FAP was diagnosed, 75% of the cases have symptomatic colon carcinoma. CHRPE is the most common extra-colonic manifestation of FAP, occurring in 70-75% of patients. CHRPE lesions in FAP are atypical in that they are multiple, bilateral, and familial (multiple CHRPE) [7-12].

CHRPE lesions have no drusen and pigmentary mottling. This provides to distinguish from choroidal melanoma or nevus [1-4]. It was considered that CHRPE lesions were stationary with no potential to grow or undergo malignant transformation. However, it is known that in 75-80% of lesions, diameter slowly grows with time. The number and size of lacunae also typically increase over time [1-12].

Both patients have no multiplier and bilateral lesions and the association with FAP of these lesions is low, we did not need a consultation of gastroenterologist for the presence of any FAP or any gastrointestinal malignity.

In cases of CHRPE, fluorescein angiography typically demonstrates hypo fluorescence due to blockage of fluorescence or hyper fluorescence in the areas of RPE atrophy. Ultrasonography shows a minimal elevation and a normal or slightly increased acoustic reflection due to CHRPE [1-12]. Differential diagnosis of CHRPE should be made from the choroidal melanoma, choroidal nevus, RPE hamartoma, RPE adenocarcinoma and acquired retinal pigment epithelial hypertrophy using fundus angiography and ocular ultrasonography [1-12]. It was reported that the RPE hamartomas might be associated with FAP". In fundus examination, congenital simple hamartoma of the RPE seems as localized, elevate, black and small lesion usually located in the fovea. Unlike CHRPE, RPE hamartoma is distinctly elevated lesion and usually remains stationary [13].

Conclusion

The features of atypical lesions include the bilaterality, multiplicity (number than three, largeness (diameter with more 0.5 of the optic disk diameter), and oval or fusiform/spindle-shaped lesions of variable size associated with hypo pigmentation at one margin and widely separated localization. In each patient with atypical lesion, even if visual acuity of the patient is 20/20, a complete ophthalmological examination also included ophthalmoscopy with wide angle lenses should be performed. In all lesions, but especially the atypical lesions, regular follow-up and photographic documentation is recommended for future comparison. Although CHRPE is a benign lesion, especially, the patients with atypical and multiple lesion should be directed to a gastroenterologist and they were strictly recommended continuing the frequency of ocular screening examinations.

Conflict of Interest

The authors certify have NO affiliations with or involvement in any organization or entity with any financial interest (such as honoraria; educational grants; participation in speakers' bureaus; membership, employment, consultancies, stock ownership, or other equity interest; and expert testimony or patent-licensing arrangements), or non-financial interest (such as personal or professional relationships, affiliations, knowledge or beliefs).

Patient Consent

The patient has consented to the submission of the paper to the journal.

Bibliography

1. Kurz GH., *et al*. "Vagaries of the retinal pigment epithelium". *International Ophthalmology Clinics* 2.2 (1962): 441-464.
2. Traboulsi EI., *et al*. "Prevalence and importance of pigmented ocular fundus lesions in Gardner's syndrome". *New England Journal of Medicine* 316.11 (1987): 661-667.
3. Chamot L., *et al*. "Fundus changes associated with congenital hypertrophy of the retinal pigment epithelium". *American Journal of Ophthalmology* 115.2 (1993): 154-161.
4. Shields JA., *et al*. "Lack of association among typical congenital hypertrophy of the retinal pigment epithelium, adenomatous polyposis, and Gardner syndrome". *Ophthalmology* 99.11 (1992): 1709-1713.
5. Blair NP., *et al*. "Hypertrophy of the retinal pigment epithelium associated with Gardner's syndrome". *American Journal of Ophthalmology* 90.5 (1980): 661-667.
6. Coleman P., *et al*. "Congenital hypertrophy of the retinal pigment epithelium: prevalence and ocular features in the optometric population". *Ophthalmic and Physiological Optics* 27.6 (2007): 547-555.
7. Meyer CH., *et al*. "Congenital Hypertrophy of the Retinal Pigment Epithelium". In: Ryan SJ., *et al. Retina*, 5th Edn Saunders (2013): 2209-2213.
8. Tiret A., *et al*. "Fundus lesions of adenomatous polyposis". *Current Opinion in Ophthalmology* 10.3 (1999): 168-172.
9. Nusliha A., *et al*. "Congenital hypertrophy of retinal pigment epithelium (CHRPE) in patients with familial adenomatous polyposis (FAP); a polyposis registry experience". *Biomed Central Research Notes* 7 (2014): 734.
10. Chen CS., *et al*. "Congenital hypertrophy of the retinal pigment epithelium (CHRPE) in familial colorectal cancer". *Familial Cancer* 5.4 (2006): 397-404.

11. Iwama T., *et al*. "Association of congenital hypertrophy of the retinal pigment epithelium with familial adenomatous polyposis". *The British Journal of Surgery* 77.3 (2003): 273-276.
12. Tourino R., *et al*. "Value of the congenital hypertrophy of the retinal pigment epithelium in the diagnosis of familial adenomatous polyposis". *International Ophthalmology* 25.2 (2004): 101-112.
13. Shields CL., *et al*. "Congenital simple hamartoma of the retinal pigment epithelium. A study of five cases". *Ophthalmology* 110.5 (2003): 1005-1011.

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