From an asymptomatic lesion to a vision-threatening condition: Congenital hypertrophy of the retinal pigment epithelium complicated by choroidal neovascular membrane

Raziye Dönmez Gün, Güzide Akcay, Hatice Selen Kanar, Şaban Şimşek

We reported a case of congenital hypertrophy of the retinal pigment epithelium (CHRPE) complicated by choroidal neovascular membrane (CNVM). A 41-year-old woman presented to our clinic with visual loss in the left eye. She was diagnosed as CHRPE complicated by a CNVM. The patient was treated with 2 consecutive monthly intravitreal aflibercept (IVA) injections. The best-corrected visual acuity (BCVA) improved and stabilized at 6/6. Subretinal fluid depending on CNVM resolved completely. CHRPE complicated by CNVM in the macular area is a rare condition and these cases can be treated with IVA therapy.

Key words: Aflibercept, benign melanoma, choroidal neovascular membrane, congenital hypertrophy of the retinal pigment epithelium

Congenital hypertrophy of the retinal pigment epithelium (CHRPE) is a well-demarcated, flat to minimally elevated fundus plaque that ranges from a black homogeneous lesion, usually with typical depigmented lacunae, to a completely depigmented lesion.[1,2] The prevalence of CHRPE is between 1.2% and 4.4%.[3] It is generally detected incidentally during routine fundus examination.[4] CHRPE is usually found in the mid-periphery retina, but it may also occur in more anterior locations. Rarely, it may be located in the macula (1%) or peripapillary region (1%).[2,4] It can be associated with systemic diseases, including Gardner's syndrome, or with other forms of familial polyposis.[5]

However, this benign and usually nonevolutive lesion can occasionally show a slight enlargement. It may also present with various retinal vascular changes, including capillary nonperfusion areas, capillary microaneurysms, dye leakage, and chorioretinal anastomosis. These can be detected using fluorescein angiography (FA).[6]

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Case Report

A 41-year-old female patient presented to our clinic with a four-month history of decreased visual acuity in her left eye. She did not report any known ocular or systemic diseases. Best-corrected visual acuity (BCVA) in the right eye was 6/6 and 6/15 in the left eye. Anterior segment evaluation was unremarkable in both eyes. Examination of the right fundus was normal, while in the left fundus there was a single, flat, well-demarcated, circular, black plaque that was three disc diameters in size and confined to the inferotemporal vascular arcade. The large part of the lesion was in the inferotemporal region of the macula. The plaque contained multiple small lacunae. There were lipid exudates, a small intraretinal hemorrhage near the upper margin of the lesion and a thickened retinal area corresponding to the foveal zone [Fig. 1].

Spectral domain-optical coherence tomography (SD-OCT) of the left eye revealed that irregular hyper-reflectivity at the level of the retinal pigment epithelium (RPE) and the RPE was slightly thickened in this zone. There was moderate relative shadowing of the underlying choroid. CNVM was located above the RPE, with subretinal fluid (SRF) on both sides adjacent to the CNVM lesion [Fig. 2]. The red free fundus image showed a lack of lipofuscin with a dark appearance in the region of the plaque. SRF accompanying the CNVM lesion exhibited increased autofluorescence [Fig. 3]. On FA, there was persistent hypofluorescence of the pigmented area in the left eye. There was hyperfluorescence in the early phase with an increase in intensity and size in the late phase, consistent with a classic CNVM [Fig. 4a and b]. The choroidal neovascular network can see beneath the retina in the early phase of FA.

The patient was diagnosed with CHRPE complicated by CNVM and treated with 2 mg (0.05 mL) intravitreal aflibercept injection. Following two consecutive monthly injections, BCVA improved and stabilized at 6/6. SRF resolved completely [Fig. 5]. The patient underwent regular follow-up at our retina department. No CNVM activity was observed at the three months follow-up. The patient was referred to the gastroenterology department for investigation of Gardner's syndrome or other forms of familial polyposis syndromes. There was no evidence of gastrointestinal pathology.

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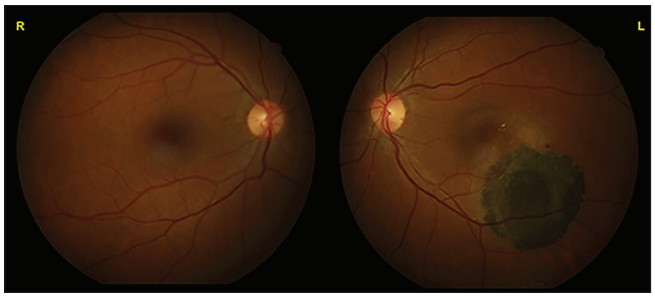

Figure 1: Color fundus photograph of the normal right eye and color fundus photograph of the left eye showing a single, flat, well demarcated, circular, black plaque (CHRPE). Lipid exudates, a small intraretinal hemorrhage near the upper margin of the lesion and a retinal thickened area corresponding to the foveal zone are seen

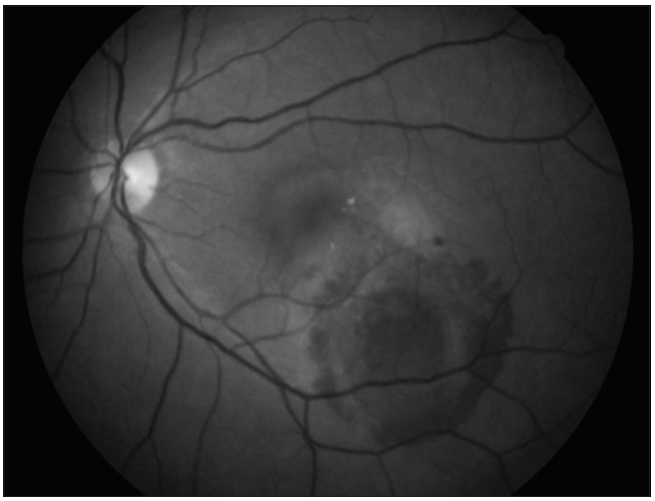

Figure 3: Red free image of the left eye showing hypoautofluorescence at the region of CHRPE lesion

Discussion

In this report, we showed the diagnosis and treatment option of a CNVM case related to CHRPE, which is a very rare condition in the literature.[6-8]

Solitary CHRPE can present similarly to choroidal melanoma.[2] In our case, we first determined that the dark plaque in our patient's left eye was CHRPE because it was important to distinguish this benign lesion from malignant melanoma to evaluate the patient's prognosis. Then, we performed the required tests and determined that the decreased vision was caused by CNVM.

Histological studies have shown that CHRPE lesions are characterized by abnormal RPE, increased cell density and increased cell height. RPE cells adjacent to CHRPE lesions may initially appear normal, but they tend to broaden and flatten out. According to the literature, these flatter RPE cells may induce pressure on the hypertrophic RPE cells present within the CHRPE lesion, and this pressure may create microtrauma at the junction between the two cells.[9] Some authors believe that microtrauma may cause breaks in the RPE associated with these areas, and RPE cells may migrate into the inner retina. This may lead to subretinal fibrovascular growth associated with neovascular membranes at the junction between CHRPE

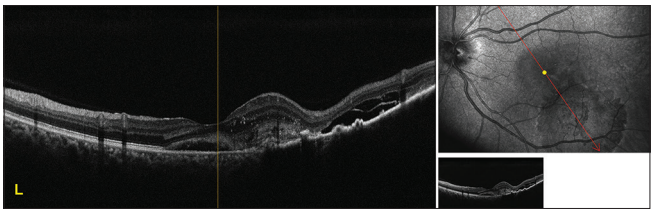

Figure 2: SD-OCT of the left eye showing flat hyper-reflective lesion with irregularly, slightly thickened RPE, moderate relative shadowing of the underlying choroid and replacement of the choroidal architecture with the CHRPE lesion. CNVM is seen above the RPE with SRF on both sides adjacent to the CNMV lesion

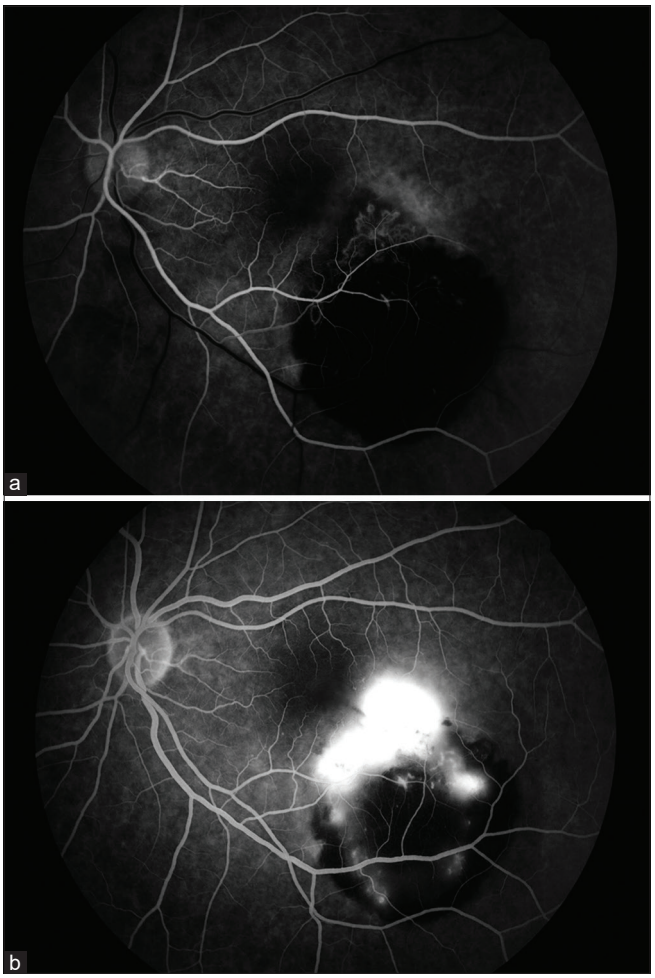

Figure 4: (a) FFA of the left eye at 5.8 seconds. The CHRPE lesion is showing hypofluorescence. Dye leakage and early hyperfluorescence adjacent to the CHRPE plaque is seen. (b) FFA of the left eye at 5 minutes, 16 seconds. Increased hyperfluorescence and late retinal leakage consistent with a CNVM

lesions and adjacent normal tissue.[7] Like Youhnovska *et al*., in our patient, it may be explained as a CNVM formation at the junction of the CHRPE lesion and adjacent normal tissue. Similar to their case, in our patient, there was fluorescein leakage on FA at the margin of the lesion, which was caused by CNVM.[7] Garoon *et al*. reported a 12-year-old female patient diagnosed with CHRPE complicated by CNVM. They treated the patient with two consecutive monthly intravitreal

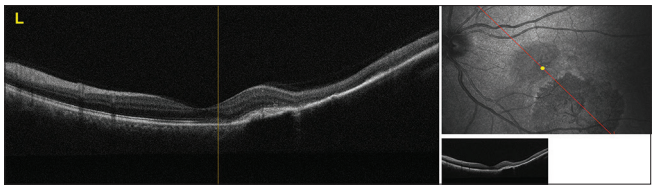

Figure 5: SD-OCT of the left eye showing completely resolved SRF on both sides adjacent to the CNMV lesion

bevacizumab injections and noted no recurrence through one year of follow-up.[8]

Conclusion

In conclusion, CHRPE is a nontumorous lesion with a generally excellent prognosis. When it is complicated by CNVM, however, it can become a vision-threatening condition. CHRPE complicated by CNVM can be treated with intravitreal aflibercept therapy. We would like to state that we presented the short-term treatment result in this report and the patient should be followed up longer for any recurrence detection. The best management of solitary CHRPE is periodic observation.[10] We want to emphasize the great importance of routine ophthalmic examination from birth for detecting asymptomatic lesions like CHRPE. An asymptomatic lesion may become a vision-threatening condition when it is complicated with other diseases as in this case.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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