OLGU SUNUMU CASE REPORT

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Choroidal Neovascularization Secondary to Vitelliform Dystrophy: Optical Coherence Tomography Angiography versus Fluorescein Angiography

Vitelliform Distrofiye Sekonder Koroidal Neovaskülarizasyon: Optik Koherens Tomografi Anjiyografi ve Fluoresein Anjiyografi Bulguları

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ABSTRACT This case series aims to interpret the findings of optical coherence tomography angiography (OCTA) and fluorescein angiography (FA) in detecting choroidal neovascularization (CNV) in juvenile Best vitelliform macular dystrophy (BVMD) and adult-onset foveomacular vitelliform dystrophy (AOFVD). Two patients with BVMD and one patient with AOFVD were evaluated, and color fundus photography, spectral-domain optical coherence tomography, FA, and OCTA were performed. In 4 of 6 eyes, OCTA revealed neovascular formations in the outer retina and choriocapillaris. It is an effective, non-invasive method for showing CNV formation using OCTA in BVMD and AOFVD, and it has aspects that are superior to FA.

Keywords: Best vitelliform macular dystrophy; adult-onset foveomacular vitelliform dystrophy; choroidal neovascularization; optical coherence tomography angiography

ÖZET Bu olgu serisi, juvenil Best vitelliform maküler distrofide (BVMD) ve erişkin başlangıçlı foveomaküler vitelliform distrofide koroidal neovaskülarizasyonun (KNV) saptanmasında, optik koherens tomografi anjiyografi (OKTA) ve fluoresein anjiyografi (FA) bulgularını yorumlamayı amaçlamaktadır. Juvenil başlangıçlı 2 hasta ve erişkin başlangıçlı vitelliform distrofili 1 hasta olmak üzere 3 hastanın 6 gözü değerlendirildi. Hastalara renkli fundus fotoğrafi, spektral domain optik koherens tomografi, FA ve OKTA yapıldı. Altı gözden 4'ünde OKTA, dış retinada ve koryokapillaristeneovasküler membranlar gösterdi. OKTA, BVMD ve erişkin başlangıçlı foveomaküler vitelliform distrofi hastalarının takipleri sırasında ortaya çıkabilen KNV'leri göstermede etkili, invaziv olmayan bir yöntem olup, FA'dan üstün olduğu durumlar bulunmaktadır.

Anahtar Kelimeler: Best vitelliform maküler distrofi; erişkin başlangıçlı foveomaküler vitelliform distrofi; koroidal neovaskülarizasyon; optik koherens tomografi anjiyografi

Best vitelliform macular dystrophy (BVMD) is a retinal dystrophy showing autosomal dominant inheritance affecting the central retina. It is characterized by the accumulation of lipofuscin-like material above, inside, and below the retinal pigment epithelium, and is associated with the *BEST1* gene, which contains 11 exons and is localized in the 11q12-13 chromosome.[1] Blurred vision, decreased central vision, and metamorphopsia are typical sypmtoms of BVMD. It begins during childhood and progresses toward adulthood.

Unlike BVMD, foveal lesions in adult-onset foveomacular vitelliform dystrophy (AOFVD) are smaller, appear later, and do not show similar mental changes. Patients are generally diagnosed in the third or fifth decade, and disease progression is slow. Unless

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atrophic changes and choroidal neovascularization (CNV), visual acuity is preserved for a long time. CNV is a rare complication in the course of BVMD and AOFVD.[2,3]

Optical coherence tomography angiography (OCTA) findings of CNV during the follow-up of 2 BVMD and 1 AOFVD cases are presented in this paper, together with fluorescein angiography (FA) images.

CASE REPORTS

CASE 1

A 13-year-old female presented with a complaint of low vision in the left eye. Visual acuity of the patient at admission was 1.0 in the right eye and 0.5 in the left eye according to the ETDRS chart. The anterior segment examination was bilaterally unremarkable. Fundus examination of the right eye was normal; however, her left eye was observed to have a yellowish vitelliform lesion with scrambled egg appearance, which is a typical finding of the vitelleruptive phase. In electrooculography (EOG), the Arden ratio was 1.2 in the right eye and 1.3 in the left eye. Although no significant change was found on the first examination of the right eye, the decrease in Arden ratio in this eye indicates that the disease was in the previtelliform stage. This reveals the bilateral but asymmetric nature of the disease.At the last visit of the patient, who was followed for 17 years, it was observed that visual acuity decreased to 0.5 in the right eye due to CNV development and to 0.1 in the left eye due to foveal atrophy. Images of the patient's right eye are presented in Figure 1.

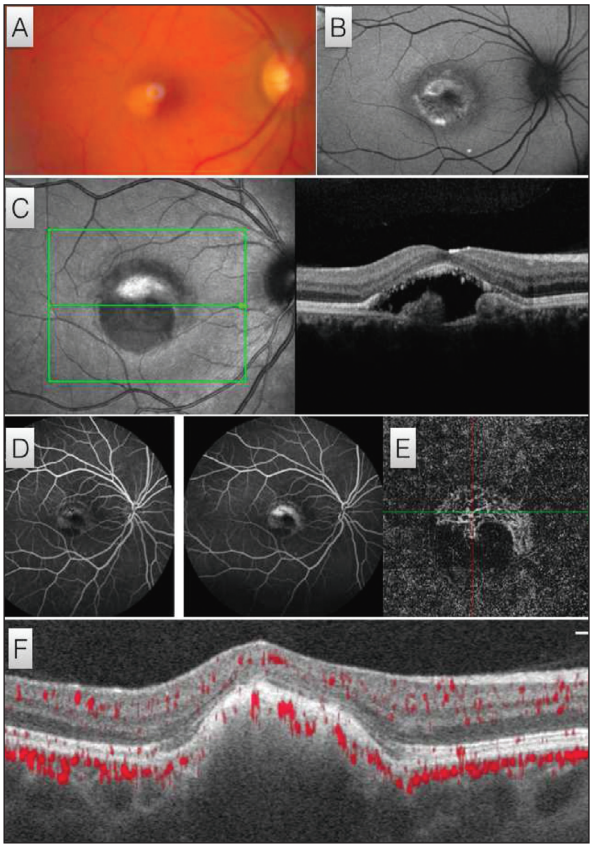

FIGURE 1: Case 1. A) Color fundus photograph shows a yellow-orange vitelliform lesion in the fovea. B) Autofluorescence shows a heterogeneous hyper-autofluorescence around the lesion. C) Spectral-domain optical coherence tomography shows resorbed vitelliform material, subretinal fluid and degenerated photoreceptor protrusions. D) Fluorescence in the middle phases of fluorescein angiography increases in the late phases and creates leakage. E) The optical coherence tomography angiography image shows the neovascular membrane in the outer retinal plexus. F) The flow signal in the flow superimposed structural optical coherence tomography image confirms the presence of choroidal neovascularization in this eye.

CASE 2

A 9-year-old female presented with blurry vision in both eyes. According to the ETDRS chart, the patient's visual acuity at the time of admission was at the 1.0 level in both eyes. A vitelliform lesion with an egg yolk appearance was observed in the fundus examination of both eyes. In EOG, the Arden ratio was found to be 1.5 in the right eye and 1.4 in the left eye. After 14 years of follow-up, the patient's visual acuity was found to be 0.1 in the right eye and 0.5 in the left eye, due to the development of CNV in both eyes of the patient at the last visit. Images of the patient's right and left eyes are presented in Figure 2 and Figure 3.

CASE 3

A 79-year-old female patient, who was under follow-up with the diagnosis of AOFVD, presented with decreased vision in the left eye. At the time of admission, visual acuity was 0.4 in the right eye and 0.1 in the left eye. Anterior segment examination showed that both eyes were pseudophakic, and a vitelleruptive lesion was observed in the fundus examination of both eyes. CNV images of the patient's left eye are shown in Figure 4.

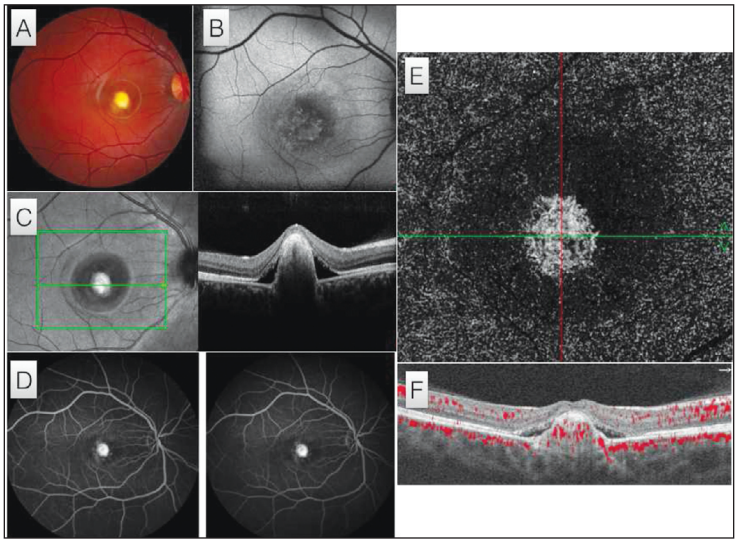

FIGURE 2: Case 2 right eye. A) Color fundus photograph shows the lesion of the vitelliruptive stage. B) Autofluorescence shows an irregular hyper-autofluorescence in the area compatible with the lesion. C) Spectral-domain optical coherence tomography shows the resorbed vitelliform material, subretinal fluid and the characteristic "circus tent" appearance. D) Hyperfluorescence in the middle phases of fluorescein angiography continues to increase in the late phases. E) The optical coherence tomography angiography image shows the neovascular membrane in the outer retinal plexus. F) The flow pattern in the flow superimposed structural optical coherence tomography image confirms the presence of choroidal neovascularization in this eye.

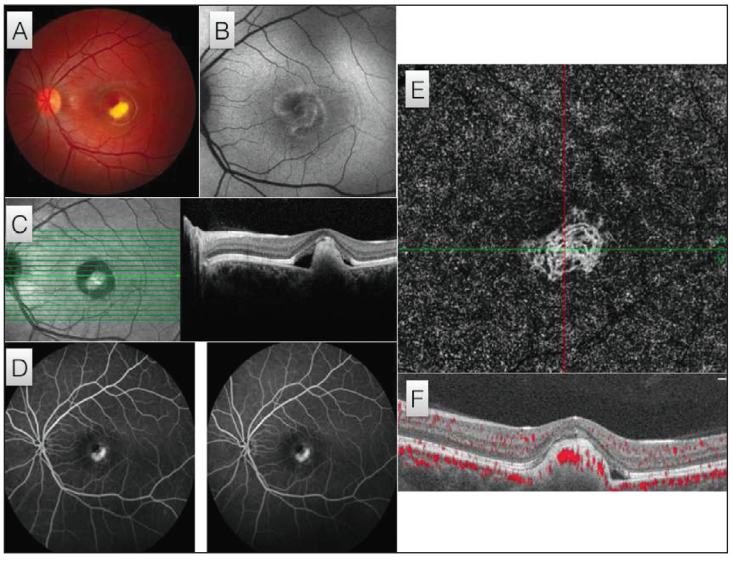

FIGURE 3: Case 2 left eye. A) Color fundus photograph shows irregular, partially resorbed vitelliform material. B) Autofluorescence shows a heterogeneous hyper-autofluorescence in the fovea. C) Spectral-domain optical coherence tomography shows hyper-reflective vitelliform material and serous macula detachment. D) Fluorescence increases in the mid and late phases of fluorescein angiography. E) The optical coherence tomography angiography image shows the neovascular membrane in the outer retinal plexus. F) The flow pattern in the flow superimposed structural optical coherence tomography image confirms the presence of choroidal neovascularization in this eye.

DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patient consent forms. The patients have given their consent to report their clinical information and images in the journal. They understand that their names and initials will not be revealed.

DISCUSSION

The development of CNV in BVMD and AOFVD is a rare condition.[2,3] An unexpected decrease in vision during the follow-up of the patients and hemorrhage around the vitelliform lesion suggest the development of CNV.

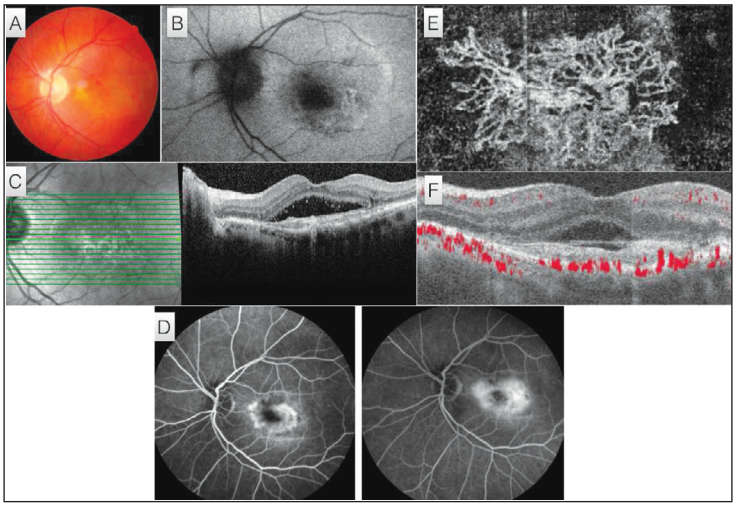

FIGURE 4: Case 3. A) Color fundus photograph shows irregular, resorbed vitelliform material in the macula. B) Autofluorescence shows hypo-autofluorescence in the area compatible with the lesion and irregular hyper-autofluorescence in its neighborhood. C) Spectral-domain optical coherence tomography shows serous macular detachment replacing resorbed vitelliform material. D) Fluorescence in the mid-phases of fluorescein angiography increases in the late phases causing leakage. E) The optical coherence tomography angiography image shows the neovascular membrane with dense anastomoses with numerous branches in the outer retinal plexus. F) The flow pattern in the flow superimposed structural optical coherence tomography image confirms the presence of choroidal neovascularization in this eye.

FA and OCTA are very useful in detecting CNV. OCTA is advantageous because it is non-invasive, does not require dye use, eliminates the risk of allergic reactions, and can be applied in kidney failure. In addition, the morphology and size of neovascular membranes due to the absence of leakage is clearly revealed by OCTA. This feature allows shrinkage and regression detection in the neovascular membrane during treatment follow-up. Furthermore, OCTA overcomes the difficulties caused by accumulation of vitelliform material in FA. The resorption of vitelliform material and its replacement with subretinal fluid makes it difficult to distinguish from the fluid originating from CNV in the later stages of the disease. At this point, although leakage in FA is in favor of CNV, vitelliform material can mask this leak, OCTA overcomes this challenge. The study of Lupidi et al. on AOFVD showed that OCTA had higher sensitivity and specificity in detecting CNV than FA After evaluating 19 eyes of 10 juvenile patients with BVMD, Guduru et al. identified different CNV patterns in 7 eyes and stated that OCTA was superior to FA.[4,5]

However, the lack of leakage in OCTA can also be encountered as a disadvantage because it limits the ability to interpret neovascular membrane activation.[6] Projection and motion artifacts, which arise with the appearance of large vessels, especially in the outer retinal layer and choriocapillaris layer, make it difficult to define the vascular network.[7]

In conclusion, this article contributes to the diversity of neovascular membranes in BVMD and AOFVD reported in different series in the literature. OCTA is helpful in detecting CNV in cases where FA cannot, and following-up on patients during the treatment process by taking into account the segmentation errors and artifacts. However, no sufficient data is available to confirm that OCTA can be used as a primary diagnostic tool. Further data will help determine the role of OCTA in clinical use.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Ali Hakan Durukan, Alper Can Yılmaz;* ***Design:*** *Ali Hakan Durukan, Alper Can Yılmaz;* ***Control/Supervision:*** *Alper Can Yılmaz, Umut Karaca;* ***Data Collection and/or Processing:*** *Alper Can Yılmaz, Taryel Rustemov;* ***Analysis and/or Interpretation:*** *Alper Can Yılmaz, Taryel Rustemov;* ***Literature Review:*** *Alper Can Yılmaz, Umut Karaca;* ***Writing the Article:*** *Alper Can Yılmaz, Umut Karaca;* ***Critical Review:*** *Ali Hakan Durukan, Umut Karaca;* ***References and Fundings:*** *Ali Hakan Durukan.*

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