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Case Report / Olgu Sunumu

# Demonstrationof CicatricialStagewith Optical CoherenceTomography in A Child with Best VitelliformMacularDystrophy

## Best Vitelliform Makular Distrofili Bir Çocukta Optik Koherans Tomografi İle Sikatrisyel Evrenin Gösterilmesi

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**Abstract:**Best vitelliform macular dystrophy (BVMD) is a rare hereditary macular dystrophy. Various staging systems are performed in the clinical evaluation of BVMD. In this case report, it is aimed to demonstrate the presence of cicatricial stage in a 9-year-old boy. A 9-year-old boy was applied to our Eskisehir YunusEmre State Hospital OphthalmologyClinic with decreased visual acuity in his right eye. When the family history of the patient was questioned, it was learned that the other family members were also followed in another center due to loss of vision. BMVD diagnosis was confirmed by using electrophysiological tests and additional clinical studies. In optical coherence tomography, a hyporeflective lesion consistent with subretinal fluid was observed for both eyes. Additionally, a hump-shaped subretinalhyporeflective lesion was found in the right eye. Visual acuity is generally stable in the early stages of the disease for patients with BVMD, but the presence of a visual impairment is usually thought as a sign of the development of a choroidal neovascular membrane. Although rare, however, cicatricial stage, which is observed in the advanced stages of the disease, can be seen in small children. Distinguishing choroidal neovascular membrane from the cicatricial stage is necessary especially in children due to the risk of amblyopia and to make an accurate treatment planning.
**Keywords:** Best vitelliform macular dystrophy, optic coherence tomography, cicatricial stage

**Özet:**Best VitellifomMakulaDistrofisi (BVMD), nadir görülen kalıtsal bir makuladistrofisidir. BVMD'nin klinik değerlendirmesinde çeşitli evreleme sistemleri uygulanmaktadır. Bu olgu sunumunda 9 yaşında bir erkek çocukta sikatrisyel evrenin varlığının gösterilmesi amaçlanmaktadır. 9 yaşında erkek çocuk, sağ gözünde görme keskinliği azalması şikayetiyle Eskişehir Yunus Emre Devlet Hastanesi Göz Hastalıkları kliniğine başvurdu. Hastanın soy geçmişi sorgulandığında, ailedeki diğer bireylerin de görme kaybı nedeniyle başka bir merkezde takip edildiği öğrenildi. BMVD tanısı elektrofizyolojik testler ve ek klinik çalışmalar kullanılarak doğrulandı. Optik koherenstomografide her iki gözde subretinal sıvı ile uyumlu hiporeflektif lezyon izlendi. Ayrıca sağ gözde tümsek şeklinde subretinalhiporeflektif lezyon tespit edildi. BVMD'li olgularında hastalığın erken evrelerinde görme keskinliği genellikle stabildir. Ancak görme bozukluğunun varlığı genellikle koroidalneovaskülermembran gelişiminin bir işareti olarak düşünülür. Nadir de olsa hastalığın ileri evrelerinde görülen sikatrisyel evre küçük çocuklarda da görülebilmektedir. Özellikle çocuklarda ambliyopi riski nedeniyle koroidalneovaskülermembranınsikatrisyel evreden ayırt edilmesi ve doğru tedavi planlaması yapılması gerekmektedir.
**Anahtar Kelimeler:**Bestvitelliformmakuladistrofisi, Optik koherenstomografi, sikatrisyel evre

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## 1. Introduction

Best vitelliform macular dystrophy (BVMD) is one of the most frequently encountered autosomal dominant (AD) retinal dystrophies and it predominantly affects the macula [1]. BMVD is typically bilateral although asymmetric disease is not uncommon [2]. BVMD is a hereditary retinal disease characterized by the bilateral accumulation of large egg yolk-like lesions in the sub-retinal and sub-retinal pigment epithelium spaces. Macular degeneration in BVMD can begin in childhood or adulthood. The variation in the age of onset is not clearly understood [3]. In our case report, a 9-year-old child with a typical BVMD pattern was presented and the presence of a rare cicatricial stage in other eye was demonstrated with fundus fluorescein angiography and optical coherence tomography.

## 2. Case Presentation

A nine-year-old boy was applied to our clinic with reduced visual acuity in his right eye. The family history of the patient was also questioned. The other family members were followed in another center related with visual impairment. The best corrected visual acuity (BCVA) was 20/30 in the right eye and 20/100 in the left eye. Biomicroscopic examination, intraocular pressure, anterior segment evaluation and pupillary reflex were normal. EOG was depressed with Arden ratio of 1.25 and 1.34 in the right and left eyes, respectively. An Arden ratio <1.5 along with clinical features is considered diagnostic of BVMD. There was a positive family history, but examination of family members or genetic testing could not be done. Fundus examination revealed a subretinal yellowish lesion in each eye, which was more characteristic in the left eye, demonstrating a typical vitelliform pattern in the central macula. Asymmetric pigmentary alterations were determined in the center of the yellowish lesion in the right eye, (figure 1, 2). Fundus fluorescein angiography performed at the same time (figure 3, 4).

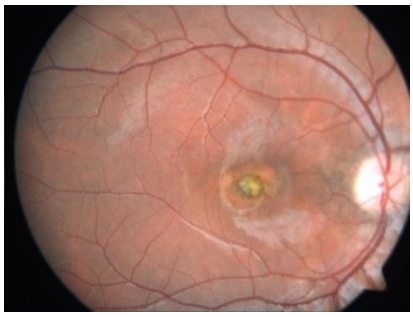

**Figure 1.** The right eye had central pigmentary alterations surrounded by a ring of yellowish deposits.

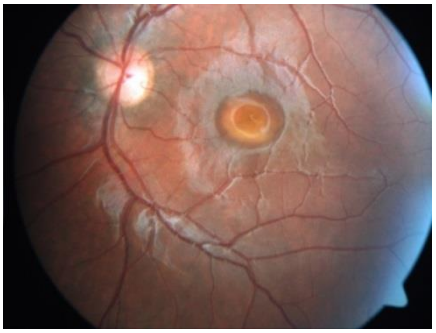

**Figure 2.** The left eye had macular pigmentary alterations surrounded by yellow deposits in a ring-like manner.

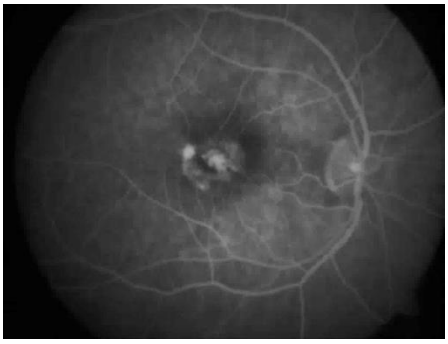

**Figure 3.**Right eye, fundus fluorescein angiography demonstrating unshaped hyperfluorescence fields and focal hypofluorescence at the center of the lesion.

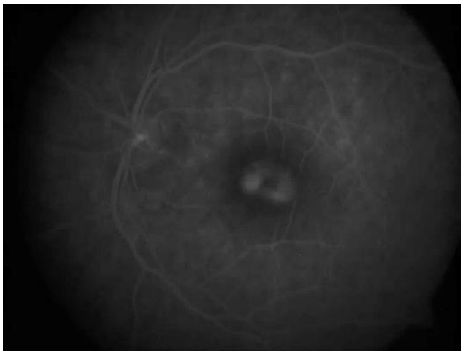

**Figure 4.**Left eye, fundus fluorescein angiography demonstrating regular oval shape hyperfluorescence zone compatible with the yellowship lesion at the center of macula.The fluorescein accumulation within the lesion in the late phase.

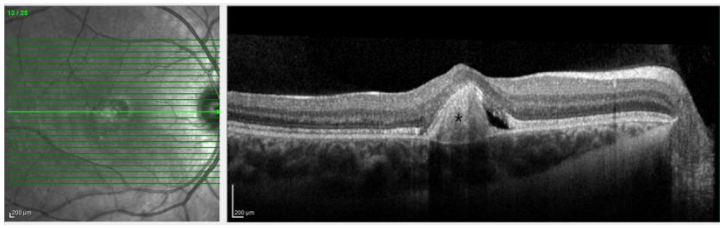

**Figure 5.** Right eye had a hyperreflective mound in the hyporeflectivesubretinal space under the fovea region (showing with black star -*). The elevation of the macula with a highly reflective subfoveal mass, corresponding to the scar [1]. All retinal layers and RPE appear to be relatively preserved over the entire area. The thickened photoreceptor layer at the left borders of the serous retinal detachment corresponds to an outer ring of increased hyperfluorescence. An area of partial photoreceptor loss around the central mass co-locates to reducedperifovealhypofluorescence zone. The prominent scar under the fovea seems to contain an irregular RPE cell layer.

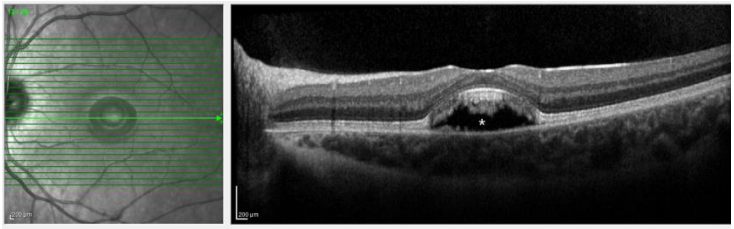

**Figure 6.** Left eye, hypo reflective field compatible with subretinal fluid (showing with white star-*). Pictures demonstrating thickened ellipsoid zone which is separated from the retinal pigment epithelium/Bruch complex by an optically clear space typical of vitelliform lesion.

## 3. Discussion and Conclusion

BVMD is an autosomal dominant inheritance maculopathy with different expressiveness and penetration, associated with mutations of the bestrophin gene located in chromosome 11 (11q131). The product of said gene is bestrophin-1, a transmembrane protein the expression of which is related with chlorine channels in the EPR cells [4].

Several classifications of BVMD have been suggested, according to the aspect of the lesions on ophthalmoscopy. The first stage is generally considered as the carrier or previtelliform stage among these classifications, in which the fovea is normal or demonstrates discrete RPE changes, in combination with an abnormal EOG. This stage may be followed by the vitelliform stage, in which a slightly increased macular lesion is observed, that is completely filled with yellowish material, resembling an egg yolk. In the next stage, the previously confluent vitelliform material breaks up, resulting in the vitelliruptive or ‘‘scrambled-egg’’ stage. This may then be followed by the pseudo hypopyon stage in which a horizontal level of the yellowish vitelliform material is observed in the inferior part of the lesion. Above this level of vitelliform material, the lesion consists of relatively transparent fluid. By the time, chorioretinal atrophy ensues, in the so-called atrophic stage. When subsequent scarring appears, which is sometimes related with choroidal neovascularization, this is thought to be the final cicatricial and/or neovascular stage by most classifications [1]. Many patients demonstrate a different stage in each eye [5]. Even eyes with lesions in the cicatricial stage often retain a remarkably good visual acuity, despite impressive central scarring [6].

Best's disease is usually diagnosed in childhood. However, vision loss or scar formation occurs at later ages Visual acuity was initially normal or slightly decreased. Vision is preserved in most patients until old age. However, some patients (even young people) may experience reduced vision down to the level of counting fingers [7]. The decrease in vision in patients is often temporary. Visual acuity is generally stable for long periods of time in patients with BVMD, but if visual impairment occurs, usually the development of a choroidal neovascular membrane is considered [8]. Reportsindicatethatsuddenvisionlossduringthe naturalcourse of BVMD is causedbysecondaryneovascularization (NV) and is a relativelycommonoccurrence[9]. A leakage from the lesion in fundus fluorescein angiography signifies the development of the neovascular membrane. In optic coherence tomography, subretinal or intraretinal fluids in addition to hyperreflectivity are observed due to neovascularization [10]. We did not observe these findings in our case.

Using OCTA, Batıoğlu et al. detected NV networkswithassociatedpolypoidaldilations at thechoriocapillarislevel in botheyes of a pregnant BVMD patientwithcomplaints of reducedvision, anddescribedthecase as pachychoroidneovasculopathy[11].

BVMD is a rare retinal disease and has a highly variable phenotypic expression [12]. In BVMD macular degeneration can start in childhood or adulthood. The variation in the age of onset is not well understood [3]. The last stage of BMVD is generally observed in an advanced age. The cicatricial stage is rarely observed among small children. Distinguishing choroidal neovascular membrane from the cicatricial stage is necessary especially for children because of the risk of the amblyopia and to make an accurate treatment planning. The cicatricial stage should be kept in mind when subretinal hemorrhage and exudates are not observed but a typical hump-like hyperreflective lesion is observed in optical coherence tomography.

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### Ethics

**Informed Consent:** The authors declared that informed consent form was signed by the patient.
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**Peer-review:** Internally peer-reviewed.
**Authorship Contributions:** Surgical and Medical KD Practices:KD Concept:KD Design: KD Data Collection or Processing: KD Analysis or Interpretation: KD Literature Search:KD Writing: KD.
**Conflict of Interest:** No conflict of interest was declared by the authors.
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