LACK OF RESPONSE TO INTRAVITREAL RANIBIZUMAB TREATMENT IN ADULT ONSET FOVEOMACULAR VITELLIFORM DYSTROPHY COMPLICATED WITH CHOROIDAL NEOVASCULARIZATION: A CASE REPORT

Berhuni Mustafa[1], Tıskaoğlu Nesime Setge[1], Ozturkmen Cem[2]

[1]Ersin Arslan Research and Education Hospital, Gaziantep, Turkey
[2]Goznuru Eye Hospital, Gaziantep, Turkey

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Mustafa Berhuni, MD
Batıkent mah 9005 no street 4/3
Gaziantep
Turkey
E-mail: mustafaberhuni@gmail.com

SUMMARY

Adult-onset foveomacular vitelliform dystrophy (AOFVD) is a rare disease characterized by accumulation of yellowish deposits in the macula. Rarely, it may be complicated by choroidal neovascularization (CNV). Cases with CNV may be confused with occult CNV in age-related macular degeneration. In our case, we will present the visual and anatomical results of a patient with AOVF-related CNV, in which we administered 3 doses of intravitreal ranibizumab (IVR). A 59-year-old female patient, who attended our clinic with the complaint of decreased vision in both eyes, was diagnosed with AOVF-related CNV in both eyes and was treated with 3 doses of IVR for 3 months. Despite the improvement in visual and anatomical functions 1 month after the first dose, vision decreased, and anatomical functions regressed to the pre-injection state in continued injections. IVR therapy is not an appropriate treatment option in the treatment of AOVF-associated CNV.

Keywords: adult onset vitelliform dystrophy, anti-VEGF, retinal dystrophy, subretinal deposits, optic coherence tomography

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INTRODUCTION

Adult-onset foveomacular vitelliform dystrophy (AOFVD) is a rare macular dystrophy characterized by subretinal yellowish material deposition similar to Best vitelliform dystrophy, first described by Gass et al. in 1974 [1]. It causes visual disturbances, due to slowly progressive macular atrophy [2]. While some cases may be asymptomatic, there may be complaints of metamorphopsia, hazy vision and scotoma in the 4th and 7th decades of life [2]. Although there is a different genetic inheritance in the etiology of the disease, most cases are sporadic [3]. The most important disease in the differential diagnosis is age-related macular degeneration. AOFVD, although rare, may be complicated by choroidal neovascularization (CNV). Although there is no definitive treatment, recent studies have shown that intravitreal anti-VEGF treatments are possibly beneficial to visual acuity and anatomical functions [4,5]. In our case, we will present the visual and anatomical results of a patient with AOFVD-associated CNV, to whom we applied intravitreal ranibizumab (IVR) for 3 months.

CASE PRESENTATION

A 59-year-old female patient attended our department with the complaint of decreased vision in her right and left eyes. The patient had no history of any other systemic disease. On ophthalmological examination of the pa-

tient, the best corrected visual acuity (BCVA) measured with the Snellen chart was 0.7 in the right eye and 0.3 in the left eye. Right and left slit-lamp examinations were normal. Intraocular pressures measured by Goldman applanation tonometry were 13 mmHg in the right eye and 14 mmHg in the left eye. Fundus examination of the patient with a 90-diopter lens revealed bilateral yellowish lesions (vitelliform material) in the macula in both eyes. With these findings, fundus fluorescein angiography (FFA) and optical coherence tomography (OCT) were requested for the patient. The color fundus photograph showed yellowish deposits in the macula (Figure 1 A, B). Increased hyperfluorescence in the early and late phases on FFA was consistent with typical classical CNV (Figure1: C, D, E, F). OCT imaging showed dome-shaped accumulation of vitelliform material and subretinal fluid in the macula, between the retinal pigment epithelium and the inner segment/outer segment (IS/OS) band (Figure 2 A, B). With the current findings, the patient was scheduled to be treated with 3 doses of IVR (Lucentis; Genentech, South San Francisco, California, USA) injection once a month. IVR (0.05 ml/0.5 mg) was applied to the right and left eyes under sterile operating room conditions. At the patient's follow-up 1 month later, the BCVA of the right eye was 0.8, and the BCVA of the left eye was 0.6, and it was observed that the subretinal vitelliform material and subretinal fluid had regressed and the central macular thickness (CRT) had decreased on OCT (Figure 2 C, D). One month after the second IVR dose, there was no change in BCVAs and OCT findings of both eyes (Figure 2 E, F). One month after the third injection, BCVAs had decreased in both eyes, the BCVA of the right eye was 0.6 and left eye BCVA was 0.4. OCT showed an increase in CRT and an increase in subretinal fluid (Figure 2 G, H). It was concluded that the patient was unresponsive to IVR treatment with her current findings, as there was no change in BCVAs and OCT of both eyes at the follow-up 2 months later (6th Month) (Figure 2 I, J).

DISCUSSION

Adult-onset foveomacular vitelliform dystrophy is macular dystrophy characterized by fragmentation and reabsorption of subretinal yellowish vitelliform material, with macular atrophy developing in the advanced stage of the disease [2]. Choroidal neovascularization is a rare complication of the disease. In our presented case, although there was an increase in BCVA and a decrease in subretinal fluid in OCT at the 1st month after IVR applied to both eyes, due to CNV developed in a patient with AOFVD, a decrease in BCVA and an increase in subretinal fluid in OCT were observed after the 3rd injections. On the contrary, in some case series and case reports, it has been reported that IVR treatment increases BCVA in AOFVD complicated with CNV [4–6]. Mimoun et al. in their case series of AOFVD complicated by CNV, argued that 3-dose IVR treatment was successful in stabilizing BCVA at 1 year and IVR was a reasonable treatment option [4]. Gallego-Pinazo et al. reported that after 3 months of IVR injection they administered to 6 female patients with AOFVD, mistakenly diagnosed with occult

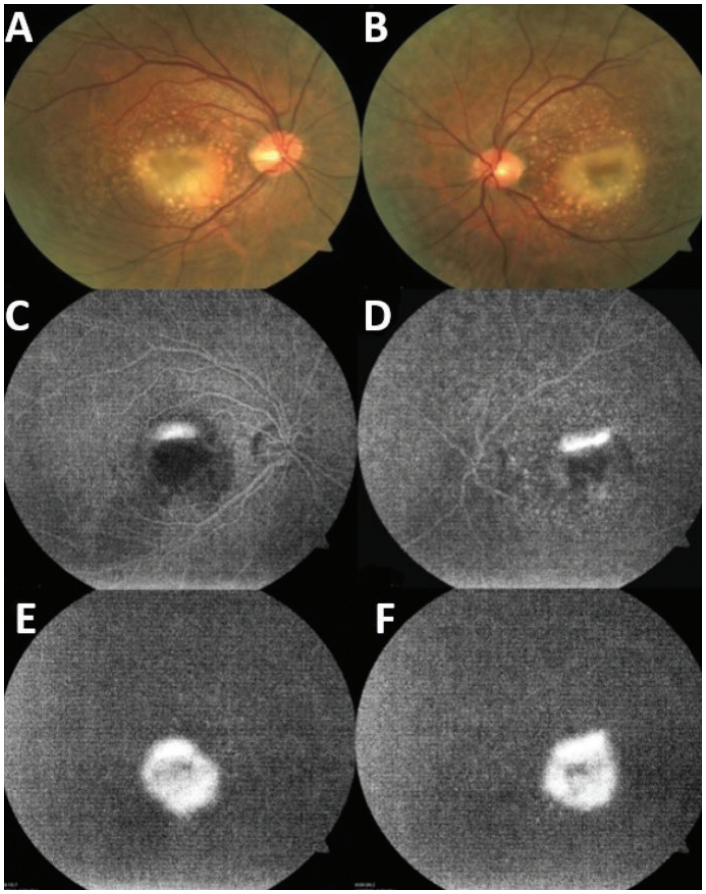

Figure 1. Preinjection, fundus imaging of right (**A**) and left (**B**) eye, fundus fluorescein angiography early phase of right (**C**) and left (**D**) eye, late phase of right (**E**) and left (**F**) eye

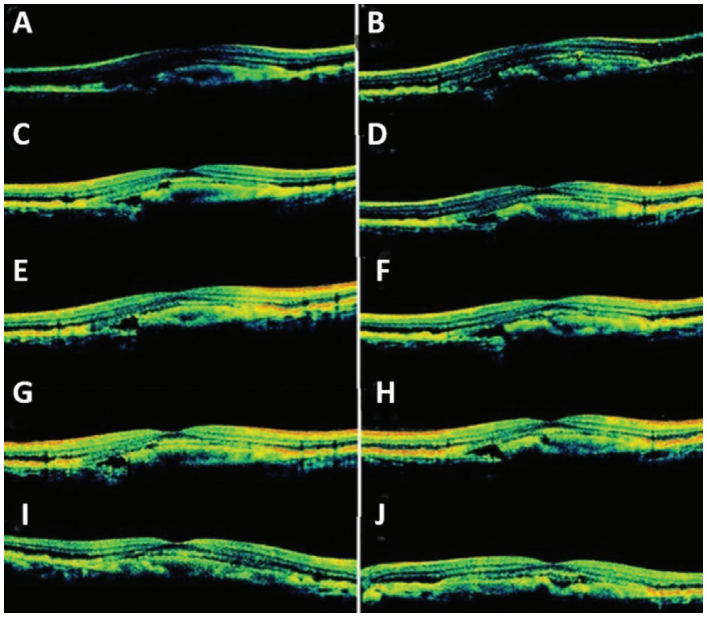

Figure 2. Optical coherence tomography imaging, preinjection right (**A**) and left (**B**) eye, postinjection 1st month right (**C**) and left (**D**) eye, 2nd month right (**E**) and left (**F**) eye, 3rd month right (**G**) and left (**H**) eye, 6th month right (I) and left (J) eye

CNV due to age-related macular degeneration, there was a significant increase in BCVA 1 month after the last injection, compared to the pre-injection values. However, the anatomical improvement did not reflect this [5]. After short-term follow-up (3 doses of IVR) in a patient with AOFVD, complicated with Type 3 CNV, Querques et al. reported that IVR was effective in preventing the progression of CNV and was an important treatment option [6]. Tissano et al. reported that the effect of intravitreal bevacizumab (IVB) treatment on BCVA was controversial in their case series with AOFVD-related CNV, but it provided a significant improvement in anatomical results [7]. Similarly to our presented case, Montero et al. reported that although IVB treatment provided morphological improvement in a patient with CNV associated with AOFVD, it had no effect on visual outcomes [8]. In our case, an early good response was obtained after the first dose of IVR in CNV associated with AOFVD, but visual and anatomical functions returned to the pre-injection state with repeated injections.

CONCLUSIONS

Although there was an increase in BCVA and a decrease in subretinal fluid 1 month after the first IVR injection in AOFVD-related CNV, BCVA decreased and subretinal fluid increased in subsequent injections. Ranibizumab treatment is not an appropriate treatment option in CNV associated with AOFVD.

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