

# Unilateral Familial Exudative Vitreoretinopathy



Dilan Yildiz<sup>1</sup>, Gamze Maden<sup>1\*</sup>, Serkan Erdenöz<sup>1</sup>, Mehmet Egemen Karataş<sup>2</sup> and Mustafa Nuri Elçioğlu<sup>1</sup>

<sup>1</sup>Department of Ophthalmology, Okmeydani Training and Research Hospital, Istanbul, Turkey

<sup>2</sup>Department of Ophthalmology, Sisli Hamidiye Etfal Training and Research Hospital, Istanbul, Turkey

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\*Corresponding author: Gamze Maden, Department of Ophthalmology, Okmeydani Training and Research Hospital, Istanbul, Turkey

## Abstract

**Aim:** We aimed to present the patient because being unilateral and symptomatic, who is diagnosed as familial exudative vitreoretinopathy (FEVR)

**Case:** In an 11-year-old male patient who presented with low vision and misalignment in the left eye, the best corrected visual acuity (BCVA) was found to be 0,9-1,0 for the right eye and 0,1 for the left eye. In the primary position, approximately 20 ° of esotropia was presented in the left eye. The globe movements were normal in all directions, and fixation was maintained with the cover test. The biomicroscopic examination was normal. On the fundus examination, the right eye was naturally observed, but on the left eye; a fibrovascular band extending from the optic disc to temporal (Figure 1) and the flattened temporal veins were observed. The differential diagnosis was premature retinopathy and FEVR, the patient didn't have a premature birth history. The patient was directed to our retina and strabismus units. In the fundus fluorescein angiography of the retina of the patient, the right eye was naturally observed; in the left eye, fibrovascular band was extending from the optic disc to the upper temporal lobe, parallelism in the temporal veins, diffuse ischemic areas in the periphery, telangiectatic vessels and leaks were observed (Figure 2) The patient was referred for follow-up by considering the diagnosis of FEVR, and family members were called to the examination but the patient and relatives could not be reached again.

**Conclusion:** FEVR is a bilateral, asymptomatic, asymmetric, inherited peripheral retinal vascular disease that commonly seen in individuals without premature birth history. However, it should not be forgotten that most of the cases emerged as new mutations. In small children, deterioration of following the object, pandemic nystagmus, strabismus, leukorrhoea may be found. Visual loss usually develops in the first decade. It must be kept in mind for this reason; As seen in our patient, being unilateral of the findings supporting the diagnosis does not exclude the FEVR.

**Abbreviations:** FEVR: Familial Exudative Vitreoretinopathy; RD: Retinal Detachment; BCVA: Best Corrected Visual Acuity; VEGF: Intravitreal Antivascular Endothelial Growth Factor

## Introduction

Familial exudative vitreoretinopathy (FEVR) is a rare vitreoretinal dystrophy that can be seen in childhood but can be diagnosed at any age [1]. Genetically heterogeneous FEVR may be autosomal dominant, recessive or X-linked [2-3] However, it should be kept in mind that only 37% of cases of FEVR have family history

and most of the cases are new mutations [4,5]. The disease is often bilateral and asymmetric. Systemic findings and preterm delivery are usually absent [2]. Symptoms vary by age and most cases are asymptomatic. (2.5) In young children, deterioration following the object, pandemic nystagmus, strabismus, leukorrhoea may occur. Visual loss usually develops in the first decade. Visual loss in the second and third decades is rare and is often associated with retinal detachment (RD) [2]. Cataract, glaucoma, band keratopathy, vitreous hemorrhage, giant retinal tears and retinal detachment are common complications of the disease [6]. Optic atrophy and

phthisis bulbi can also be seen in advanced stages of the disease. In this study, we aimed to present the patient for the reason that being unilateral and symptomatic, who is diagnosed as familial exudative vitreoretinopathy (FEVR)

## Case Report

In an 11-year-old male patient who presented with low vision and misalignment in the left eye, the best corrected visual acuity (BCVA) was found to be 0,9-1,0 for the right eye and 0,1 for the left eye. In the primary position, approximately 20 ° of esotropia was presented in the left eye. The globe movements were normal in all directions, and fixation was maintained with the cover test. The biomicroscopic examination was normal. On the fundus examination, the right eye was naturally observed, but on the left eye; a fibrovascular band extending from the optic disc to temporal (Figure 1) and the flattened temporal veins were observed. The

differential diagnosis was premature retinopathy and FEVR, the patient didn't have a premature birth history. The patient was directed to our retina and strabismus units.

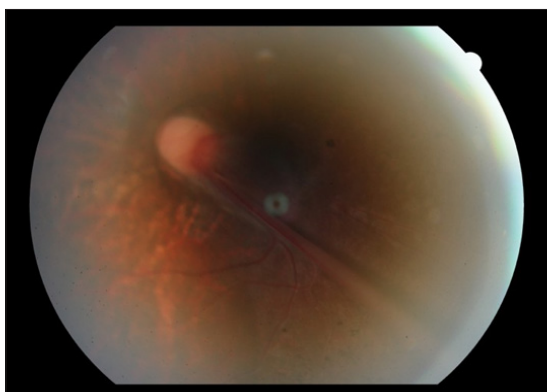


Figure 1: Fibrovascular band.

## Results

In the fundus fluorescein angiography of the retina of the patient, the right eye was naturally observed; in the left eye, fibrovascular band was extending from the optic disc to the upper temporal lobe, parallelism in the temporal veins, diffuse ischemic areas in the periphery, telangiectatic vessels and leaks were observed (Figure 2). The patient was referred for follow-up by considering the diagnosis of FEVR, and family members were called to the examination, but the patient and relatives could not be reached again.

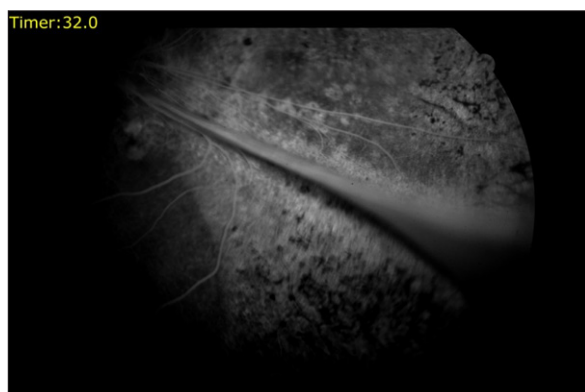


Figure 2: Fundus fluorescein angiography.

## Discussion

Familial exudative vitreoretinopathy is a rare hereditary disease due to developmental failure in vascularization of the peripheral retina. Inability to complete vasculogenesis and angiogenesis leads to the appearance of vitreoretinal disorders [7]. Mutations in genes responsible for normal retinal vasculature cause peripheral capillaries to cause sudden termination of development. These endings lead to the development of new vessels and complications due to compensatory mechanism. The resulting gene mutations also contribute to increased vascular endothelial growth factor release and contribute to the chronic course of the disease [8].

Prematurity retinopathy, Norrie disease, incontinentia pigmenti, persistent fetal vasculature, toxocariasis and Coats' should be considered in the differential diagnosis. Although they are less likely to interfere retinal capillary diseases such as hemangioblastomas, Eales disease, retinoschisis, sickle cell anemia and retinoblastomas, should be considered in the differential diagnosis too (2.5) Although it is clinically similar to retinopathy of prematurity, there is no preterm delivery in patients with FEVR, and in our study we did not have a preterm birth history [9].

Familial exudative vitreoretinopathy shows bilateral asymmetric involvement, especially in the adult age group [6]. As age increases, changes in FEVR findings and asymmetric clinical findings occur. Unilateral involvement was seen in our patients diagnosed at an early age. Clinical findings of the disease may vary widely. Mild peripheral avascular changes that occur incidentally in FA can be seen, as well as exudative and tractional findings that can lead to rapid retinal detachment. The vitreoretinal tight adhesions in the peripheral avascular retina, iatrogenic retinal tears and posterior hyaloid contraction in the posterior cause the tractional retinal detachment to occur easily [11,12]. FEVR is usually asymptomatic. Our present case complained of low vision.

Although various classifications have been made to staging familial exudative vitreoretinopathy, the most commonly used Pendergast and Trese-related disease is classified as similar to staging in premature retinopathy, which distinguishes five stages (Table 1) [13]. According to this classification, our case is consistent with Stage 2a. Treatment options include laser photocoagulation, cryotherapy, intravitreal anti-vascular endothelial growth factor (VEGF) injections, or surgical treatment options for the treatment of patients with appropriate disease [14].

**Table 1:** Clinical classification of familial exudative vitreoretinopathy in Pendergast and Trese.

Stage 1: Avascular areas in peripheral retina without retinal vascularization
Stage 2: Avascular areas in peripheral retina with non-retinal vascularization
2a: No exudation
2b: with exudation
Stage 3: subtotal retinal detachment not involving the fovea
3a: No exudation
3b: with exudation
Stage 4: subtotal retinal detachment involving the fovea
4a: No exudation
4b: with exudation
Stage 5: Total retinal detachment

## Conclusion

FEVR is a bilateral, asymptomatic, asymmetric, inherited peripheral retinal vascular disease that commonly seen in individuals without premature birth history. However, it should not be forgotten that most of the cases emerged as new mutations. In small children, deterioration of following the object, pandemic

nystagmus, strabismus, leukorrhoea may be found. Visual loss usually develops in the first decade. It must be kept in mind for this reason; As seen in our patient, being unilateral of the findings supporting the diagnosis does not exclude the FEVR.

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Gamze Maden. Biomed J Sci & Tech Res



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