Case Report

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Atypical Age Presentation of Familial Exudative Vitreoretinopathy Mimicking Persistent Fetal Vasculature in Saudi Arabia

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ABSTRACT

Background: *Patients affected with Familial Exudative vitreoretinopathy (FEVR) are the result of full term pregnancy. FEVR affects both eyes asymmetrically and patients usually present to the hospital at the age of 7. The mood of inheritance of FEVR can be autosomal dominant or recessive. FEVR is characterized by impairment of peripheral retina perfusion that can lead to retinal neuvascularization, subretinal exudate, and retinal detachment.*

Case Presentation: *Our patient presented to the Emergency department at the age of 9 weeks when his parents noticed red discoloration of the right eye pupil. The patient was born full term with no prenatal or postnatal complications and negative medical and surgical history. His parents reported that his siblings have a condition that affected their vision. Examination revealed a red colored mass on the pupil margin of the right eye, and total retinal detachment with mass located supra-temporally in the vitreous on fundus examination. The findings were also confirmed with B-scan. While there was flat retina in the left eye.*

Conclusion: *Our patient was finally diagnosed with FEVR based on examination, FFA and genetic testing which revealed an autosomal recessive inherited gene called LRP5.*

Keywords

Familial Exudative Vitreoretinopathy (FEVR), Persistent Fetal Vasculature (PFV), Retinal Disorders, Inherited Retinal Disease, Vitreoretinal Abnormalities, Ophthalmic Genetics.

Introduction

Familial Exudative vitreoretinopathy (FEVR) is first described by Criswick and Schepens in 1969 [1]. It's a rare inherited disorder of retinal vascular development in non pre-mature infants. Its typically appear bilateral and asymmetrical in any age group [1,2]. The mean age of patients affected by FEVR is 7 years [3]. This disorder had wide variability of expressivity [4]. The main phenotype characterization include peripheral retina non-perfusion as mild presentation that can progress to retinal neovascularization, subretinal exudation, formation of an abnormal vitreoretinal interface and retinal detachment in severe form [2]. The most common mode of FEVR inheritance is Autosomal dominant with mutation in *FZD4*, *LRP5, and TSPAN12*. *LRP5 can also associated with Autosomal recessive FEVR,* whereas X-linked recessive forms of the disease can be caused by mutations in *NDP* [5]. One of the mimicker of FEVR is persistent fetal vasculture (PFV) which is described as idiopathic congenital malformation in which the hyaloid artery fails to regress during development, resulting in intraocular fibrovascular remnants. It usually appear as unilateral presentation. Anterior segment appear as discovery of retrolental tissue, microcornea, and microphthalmos. However,

posterior segment finding appear as retinal and optic nerve hypoplasia and macular anomalies. As the disease progress, it may associated with secondary glaucoma, intravitreal hemorrhage, retinal detachment and amblyopia [4]. We report a case of Familial exudative vitreoretinopathy in atypical age presentation that mimic persistent fetal vasculature as rare case in Saudi Arabia.

Case Presentation

A 9 weeks old Saudi baby boy presented to the emergency room on 18-11-2023, the parents noticed red color of the pupil of the right eye. The infant is a production of Full-term C-section delivery. The perinatal period was negative for any maternal infection of pregnancy complications. Also, no significant intra-natal history. The parent gave a history of his siblings having an ocular condition affecting their visual potential. There is a history of consanguinity marriage. His clinical examination revealed an intraocular pressure of 7 mmHg OU by I-care. Slit lamp examination of right eye showed clear cornea, clear lens, Anterior chamber was deep and quiet, round pupil with sluggish reaction, whitish to reddish lesion retrolental lesion at temporal side of the pupil margin (Figure 1). Left eye: showed clear cornea, clear lens. Anterior chamber was deep and quiet, round pupil with sluggish reaction. Dilated fundus exam of the right eye: showed total Retinal detachment with supertemporal mass in the vitreous. Left eye showed flat retina, abnormal tortuous blood vessels coming from the disc. B-scan of right eye confirmed the presence of total Retinal detachment with a mass.

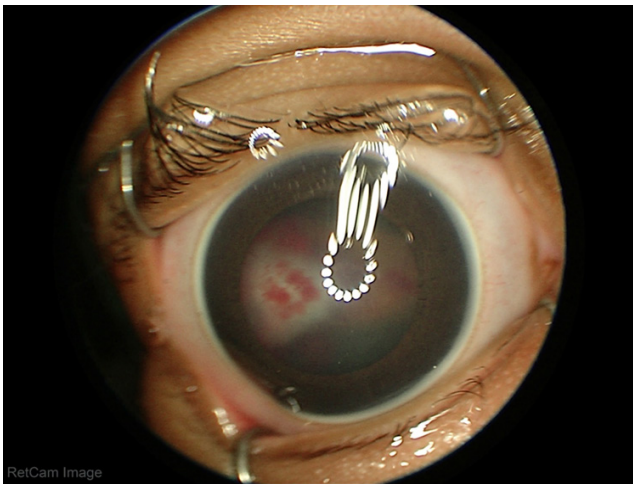

Figure 1: Photograph of the right eye whitish to reddish retrolental lesion at temporal side of the pupil margin.

The patient underwent examination under anesthesia for full examination with possible intervention. RetCam imaging was ordered for imaging. Finding of the right eye revealed whitish mass with scattered hemorrhages reaching the back of the lens temporally, tractional retinal detachment involving the posterior pole with a gliotic mass over the optic disc, a retinal and vitreous hemorrhage involving mainly the temporal retina, avascular retinal periphery with demarcation line separating the vascularized posterior pole from the avascular peripheral retina (Figure 2). left eye showed a whitish gliotic mass over the optic disc, abnormal straightening blood vessels, avascular retinal periphery with demarcation line separating the vascularized posterior pole from the avascular peripheral retina associated with neovascularization and hemorrhage at demarcation line temporally and inferonasal (Figure 3a, 3b).

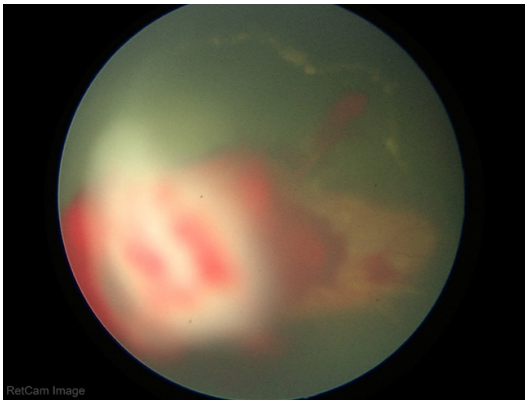

Figure 2: Tractional retinal detachment involving the posterior pole with a gliotic mass over the optic disc, a retinal and vitreous hemorrhage involving mainly the temporal retina, avascular retinal periphery with demarcation line separating the vascularized posterior pole from the avascular peripheral retina.

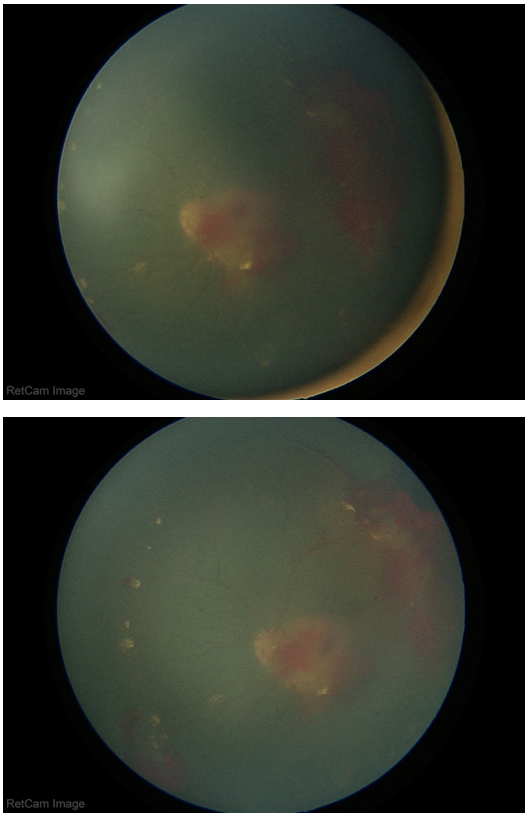

Figure 3a,3b: Fundus photo of left eye showing whitish gliotic mass over the optic disc, abnormal straightening blood vessels (3a), avascular retinal periphery with demarcation line separating the vascularized posterior pole from the avascular peripheral retina associated with neovascularization and hemorrhage at demarcation line temporally and inferonasally (3b).

Fluoresceine angiography done for both eyes which revealed leakage of the optic disc, and at areas of vascular-avascular junctions, also FFA showed an anomalous vascularization/ supernumerary vascular branching in areas of vascular-avascular junctions, nonperfused peripheral retina (Figure 4).

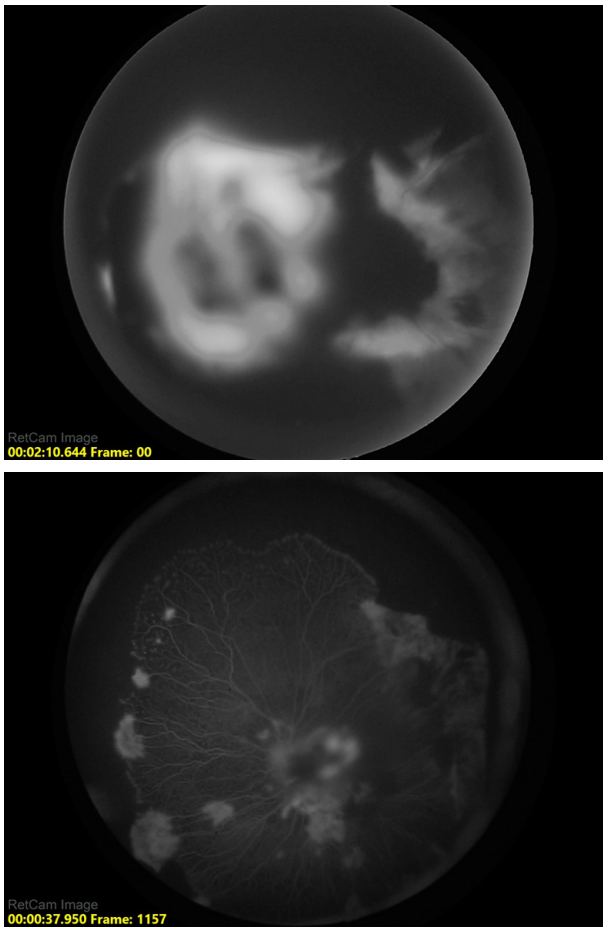

Figure 4a,4b: FFA of right eye showing sever extensive leakage of the optic disc, and leakage temporal to the macula (4a), FFA of left eye showed milder leakage of the optic disc, and leakage temporally and nasally at areas of vascular-avascular junctions, an anomalous vascularization/ supernumerary vascular branching in areas of vascular-avascular junctions, nonperfused peripheral retina (4b).

Clinical Impression was Familial Exudative Vitreoretinopathy (FEVR), laser photocoagulation applied to the avascular retina in both eyes in two different sessions with 2 weeks apart (Figure 5a, 5b). Follow up given in the clinic after 6 weeks from last treatment. Centovision genetic testing (Next generation sequencing based) was done that reveal a homozygous likely pathogenic variants identified in the LRP5 gene in autosomal recessive mode of inherence. Variant named c.2827+1G>A p.?.

Discussion

Vitreoretinal dystrophy caused by familial exudative vitreoretinopathy is a rare genetically heterogenous form of retinopathy it usually presents in full term babies with progressive nature of the disease. It typically affects the peripheral retinal vasculature bilaterally in a variable way between both eyes. Most affected individuals present at 7 years of age. However, in our case the patient who is a full-term baby presented in his 9th week of life which is atypical for this condition. patient might present with nystagmus, decrease in vision, or leukocoria in advanced cases [1]. However, mild or early FEVR is mostly asymptomatic. As in our case, the patient was asymptomatic apart from red discoloration of the pupil that was noticed by his parents. Since FEVR is an inherited disease the presence of affected family members strengthens the diagnosis of the condition. However, there is a high possibility that family members have an early-stage disease with minor or no manifestations due to this fact the absence of family history does not rule out the diagnosis. The mode of inheritance, manifestations, and severity can vary significantly between members of the same family [3]. In our case, the patient parent reported vision impairment in patient's siblings with no identified etiology.

Fundus examination of such patients shows avascular peripheral retina associated vascular leakage and exudate that is similar to ROP findings. Other manifestations include neovascularizations with increased diameter of the new vasculature that causes displacement of the macula due to dragging of retinal vasculature and traction, optic disc hypoplasia, and retinal folds. Given the progressive nature of FEVR it might lead secondary glaucoma, intravitreal hemorrhage, retinal detachment, and amblyopia [6]. On examination of our patient, we found whitish to reddish retrolental lesion on the temporal side of right eye pupil, and total RD with superiotemporal vitreal mass that was confirmed with B-scan. One important differential diagnosis that mimics FEVR is PFV which can be confused with FEVR due to the presence of retinal folds that may appear similar to PFV stalk. The presence of cataract, microphthalmia, microcornea that is unilateral and sporadic help to differentiate it from FEVR that is bilateral and inherited with picture that is similar to ROP with the patient being full term. FEVR is most commonly autosomal dominant in inheritance with mutations in FZD4, LRP5, and TSPAN12. However, LRP5 can be autosomal recessive in inheritance [5,6]. After genetic testing of our patient, we found an LRP5 mutation that is autosomal recessive which is rare in FEVR.

Conclusion

We presented a case of familiar Exudative vitreoretinopathy (FEVR) in atypical age presentation which was 9 weeks instead of mean age that presented in literature. Also, the patient was presented with Finding of the right eye revealed whitish mass with scattered hemorrhages reaching the back of the lens temporally, tractional retinal detachment involving the posterior pole with a gliotic mass over the optic disc, a retinal and vitreous hemorrhage involving mainly the temporal retina, avascular retinal periphery, which is mimic persistent fetal vasculature (PFV) in the right eye. However, left eye shows striation of blood vessel with ischemic retina in periphery that associated with hemorrhage and neovascularization. The diagnosis was confirmed clinically

by examination and FFA finding with genetic testing. Genetic testing reveals LRP5 gene in autosomal recessive mode of inherence. Variant named c.2827+1G>A p.?. With this paper we wish to encourage further research about atypical presentation of FEVR. Because this will help the physicians to detect the accurate diagnosis and treat it probably. Furthermore, with this paper we wish to encourage further studies about different FEVR presentations in Saudi Arabia. Because until this day there have been no studies regarding this subject in our country yet.

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