Case Report

Rare case of right eye persistent fetal vasculature with left eye optic nerve hypoplasia

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Abstract

Persistent Fetal Vasculature (PFV), previously known as Persistent hyperplastic primary vitreous (PHPV), is a rare congenital developmental malformation of the eye, caused by the failure of structures of primary vitreous to regress. Most common presentation is unilateral. It can occur in isolation, or in association with other ocular disorders and rarely as a part of systemic disorder. Most cases of PFV are sporadic, but it can be inherited as an autosomal dominant or recessive trait. Characteristic features include a persistent hyaloid artery along with certain anterior and posterior segment findings depending on the type of PFV. Radiological investigations (B-Scan ultrasound, Computerised Tomography, Magnetic Resonance Imaging) aid in the diagnosis and differentiation from other causes of leucocoria like retinoblastoma, Coat's disease, etc Optic nerve hypoplasia (ONH) is a congenital anomaly of optic disc that results from underdevelopment of optic nerve. Most commonly, the occurrence is sporadic though few rare cases have been reported as having autosomal recessive inheritance. Optic disc is characteristically small. This condition may be associated with various neurological and endocrine pathologies. Maternal associations such as premature births, gestational diabetes mellitus may be seen. Thorough ophthalmological investigations as well as radiological investigations (Computerised Tomography, Magnetic Resonance Imaging) along with Hormonal assays, aid in the diagnosis of ONH. This case report discusses 5 days old male child with Right eye Persistent Fetal Vasculature and Left eye Optic Nerve Hypoplasia,

Keywords: Persistent Fetal Vasculature, Optic nerve hypoplasia, leukocoria, microphthalmos.

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INTRODUCTION

PFV is a congenital anomaly of the eye that results following failure of the embryological, primary vitreous, and the hyaloid vasculature to regress.[1,2] Morton F. Goldberg, MD, in the 1997 Edward Jackson Memorial Lecture, introduced the term "persistent fetal vasculature" to replace "persistent hyperplastic primary vitreous," which he felt was a misnomer because of its failure to include all of the fetal intraocular vasculature, rather than just the post-lental vessels[3] Primary vitreous forms around 7th week of intrauterine life and starts involuting around 20th week and nearly always disappears at the time of birth. Failure of regression of primary vitreous results in many of the abnormalities seen in PHPV.[4] Pupillary strands and a Mittendorf's dot represent the mildest manifestations and leukokoria with a dense retrolenticular membrane and/or retinal detachment the most severe. Depending on which intraocular structures are involved it is divided into anterior, posterior or a combination of anterior and posterior.[4] The anterior type of PHPV includes a shallow or collapsed anterior chamber, a retrolental vascular membrane, elongated ciliary processes, microphthalmia cataract, intralenticular hemorrhage, lens swelling with secondary glaucoma and total lens absorption in rare cases.[5] Posterior PHPV consists of vitreous membrane and fibrovascular stalk that emanates from the optic nerve and courses anteriorly, retinal folds, traction retinal detachment, hypoplastic optic

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nerve and macula, microphthalmia. A combination of anterior and posterior forms also commonly occurs. Without surgery, most eyes with PHPV will develop severe glaucoma, retinal detachment, intraocular hemorrhage, and/or phthisis early in life. If left untreated, many eyes have been enucleated early in life.[6,7] PFV can occur with other disorders such as Norrie's disease trisomy 13 (Patau syndrome) Walker-Warburg syndrome Axenfeld Rieger syndrome, Peter's anomaly, Morning glory syndrome,, neurofibromatosis, Aicardi syndrome. Differential diagnosis of PHPV includes retinoblastoma, retinopathy of prematurity, Coat's disease, toxocariasis, Norrie's disease, familial exudative vitreoretinopathy, congenital cataract, uveitis, and incontinentia pigmenti.[5] The congenital malformation optic nerve hypoplasia (ONH) is a unilateral or bilateral non-progressive underdevelopment of the optic nerve, accounting for about 15%-25% of infants with serious vision loss.[8] ONH may occur as an isolated defect or in association with other ocular abnormalities (microphthalmos, aniridia, coloboma, nystagmus, and strabismus), cranial abnormalities (agenesis of septum pellucidum, anenephaly, midline abnormalities of brain) or facial anomalies. It may occur as a component of the syndrome of septo-optic dysplasia (de Morsier's syndrome), which includes midline brain malformations and hypopituitarism.[9] ONH occurs due to diminished number of axons in the involved nerve with normal development of supporting tissues and the retinal vascular system. ONH may result from an insult suffered by a normally developing system occurring any time between the sixth week and the fourth month of gestation[10] It may be associated with one of the following two causative factors: (1) a genetic alteration and (2) alteration of the intrauterine environment by maternal metabolic or toxic stress. Gestational risk factors include: Preterm birth, Low birth weight, Intrauterine growth restriction, Young maternal age, Primiparity, Viral infection (CMV) On examination, visual acuity ranges in a wide spectrum from no light perception to near normal. In unilateral or asymmetrical cases, the Marcus-Gunn pupil can be elicited[11] Diagnosis of ONH is primarily clinical and is made by ophthalmic confirmation of a small optic disc with the vasculature appearing very large relative to the disc. The retinal nerve fiber layer is variably thinned, and the disc itself may appear grayish or even white in colour.[12]

CASE REPORT

Ophthalmology consultation of 5 days old male child admitted in Neonatal Intensive Care Unit, was sent for white reflex in pupillary area of right eye. The informant was the mother. She complained of oscillatory movements of baby's eyes along with a white reflex in the right pupillary area since birth. 1.530 kgs male child was born out of non-consanguinous marriage, by Normal Vaginal Delivery at 37 weeks of gestation to a 23 years old Primi. The baby's general condition was poor and was adviced admission to Neonatal Intensive Care Unit, for low birth weight.

On ocular examination:

Leukocoria was noted in right eye. Pupillary reaction of left eye showed Relative Afferent Pupillary Defect. No other positive findings were noted on anterior segment examination. Dilated examination showed a white mass occupying the entire pupillary area of right eye.

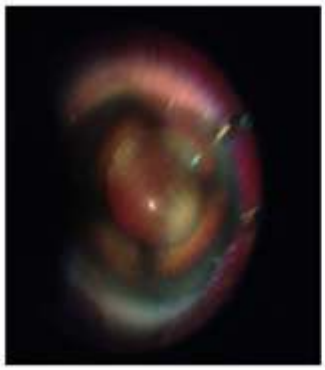

Figure 1: Right eye

Figure 2: Left eye

Dilated fundus examination of right eye was not possible due to leukocoria. Dilated fundus examination of left eye showed a small optic disc with prominent blood vessels.

Figure 3

B-scan of both the eyes was carried out. Impression: An echogenic band is seen in the posterior aspect of right globe extendingfrom optic nerve head to posterior aspect of lens with an artery running through it (s/o Hyaloid Artery) On colour and spectral Doppler, an artery is seen running through this band, suggestive of hyaloid artery. No calcification seen.

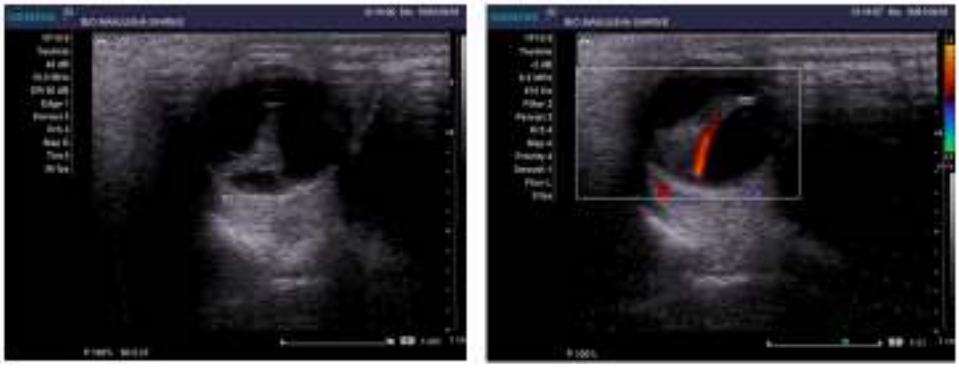

Figure 4: Right eye

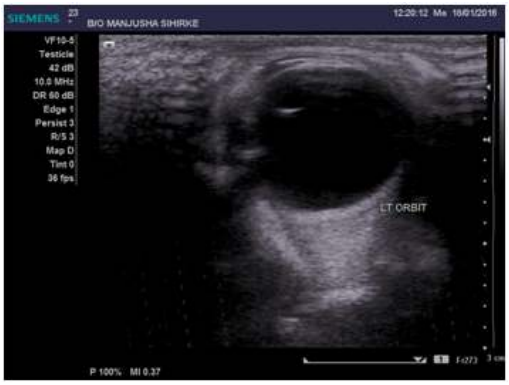

Figure 5

Impression: Right eye: PERSISTENT PRIMARY HYPERPLASTIC VITREOUS**,** Left eye: Normal**,** TORCH screen of mother was carried out which was positive for Rubella IgG (17.28) and CMV IgG (19.95). All the above features were suggestive of diagnosis of Right eye: Persistent Fetal Vasculature; Left eye: Optic nerve hypoplasia.

CONCLUSION

Mild PFV can run a relatively benign natural course without surgery. Surgery may be avoided if the visual axis is clear, anatomical anomalies are not progressive, and the anterior chamber angle is not compromised Recent advances in surgical techniques have improved the prognosis of PFV. Surgery usually consists of lensectomy, 3-port vitrectomy, or both, through a limbal

or pars plicata/pars plana approach. Surgery for severe posterior PFV is rarely undertaken. Although the fibrovascular tissue can be dissected and the accompanying retinal detachment may approximate to the retinal pigment epithelium (RPE), visual outcomes are quite poor. This is a result of the associated retinal dysplasia. Bilateral disease is also typically associated with a poor outcome due to the high prevalence of posterior component. It is important to exclude retinoblastoma in all cases of leukokoria. PHPV can be differentiated from retinoblastoma by the absence of a calcified mass, artery running through Cloquet's canal, and typical signal characteristics of retinoblastoma on MRI. Differentiation from advanced retinopathy of prematurity (ROP) can be difficult on imaging alone. History of a premature, low birth weight infant undergoing prolonged supplemental oxygen therapy helps to distinguish it from bilateral PHPV.It is differentiated from vitreoretinal dysplasia by a patent hyaloid arteryat it is not a feature of vitreoretinal dysplasia. The diagnosis of congenital ONH is of great importance because of the genetic and systemic implications. The ophthalmologist has the opportunity of being the first to diagnose an underlying disease or syndrome and referring patients for genetic and pediatric assessment. Based on the high incidence of structural brain malformations, which may be associated with an increased risk of endocrinopathy and developmental delay, it is advisable that neuroimaging of the brain is advisable in all patients with ONH. Amblyopia treatment can result in improvement of vision in the worse eye. Although there is no cure or treatment *per se*, the purpose of early intervention programs and early vision stimulation programs is to minimize the impact of the vision loss on general development in patients of ONH.

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