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Persistent Hyperplastic Primary Vitreous: A Case Report and Literature Review

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Abstract **Case Report**

Persistent hyperplastic primary vitreous (PHPV) is a rare ocular disorder caused by the incomplete regression of primary vitreous with the abnormal persistence of hyaloid vasculature. It is the second most common cause of leukocoria after retinoblastoma. Aim of this work is to report the case of unilateral PHPV in a 6 months old child. We will describe the main ocular, echographic and magnetic resonance findings, that are essential to conduct a right differential diagnosis.

Keywords: hyaloid vasculature, Persistent hyperplastic primary vitreous (PHPV), diagnosis, retinoblastoma.

INTRODUCTION

Persistent hyperplastic primary vitreous (PHPV) is a rare vitreoretinal disorder. The most common clinical manifestations of the condition are leukocoria, microphthalmia, and cataract [1]. It is a congenital disorder of the eye, which occurs due to abnormal persistence of fetal intraocular vessels and embryonic vitreous [2]. Primary vitreous forms around the seventh week of fetal life and starts regressing in the 20th week. It is replaced by avascular secondary vitreous by birth. Failure of regression of primary vitreous can lead to PHPV [3].

PHPV is one of the most important differential diagnoses of retinoblastoma [1]. It may present a diagnostic challenge for ophthalmologists and pediatricians. A detailed clinical examination along with radiological investigations is required for the diagnosis and efficient management of the disease.

CASE PRESENTATION

Our patient was a 6-months-old female infant of nonconsanguineous patents, who presented to our hospital with a unilateral leukocoria. The patient had been born at term by uneventful spontaneous vaginal delivery. There was no antenatal and family history related to presenting complaints. Systemic examination was normal. On ophthalmological examination, pupillary reactions were found to be normal. Bilateral corneal haziness was present. Bilateral pupillary reflexes were positive and the size of the right cornea was relatively smaller than of left the one. On slit-lamp examination, posterior sub-capsular opacification was observed along with a fibro-vascular thread-like extension from the posterior surface of the lens. At this stage, we were suspecting retinoblastoma.

The patient was referred to the department of radiology for further evaluation. The radiologist performed an ultrasound B scan for the identification of the pathology. Ultrasound showed echogenic masses in the posterior segment that were extending from the posterior surface of the lens to the optic disc without any intra-lesion calcification. Doppler study showed the presence of hyaloid vasculature (Figure 1).

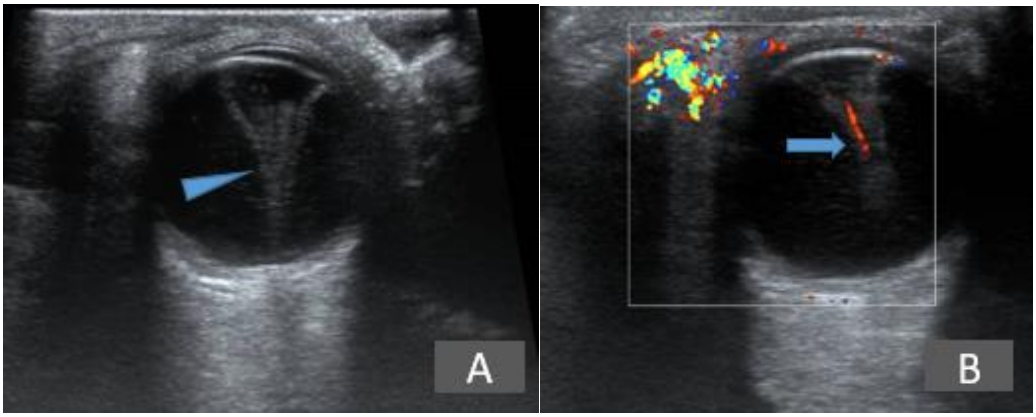

Figure 1: A: B-mode ultrasound showed an echogenic mass in the posterior segment extending from the posterior lens surface to the optic disc without intra-lesional calcification (arrowhead). B: Doppler ultrasound showed the presence of hyaloid vasculature (arrow)

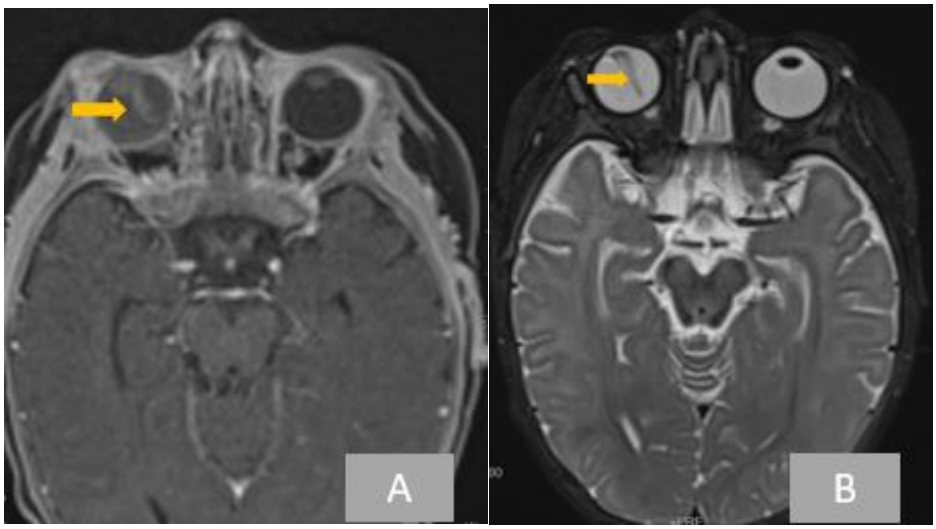

Figure 2: Figure 1: Axial T1 contrast material–enhanced (A) abd T2-weighted (B) 1.5-T MR image; A funnel-shaped extending from posterior segment extending from the posterior lens surface to the optic disc (arrow). The lens has abnormal morphology compared with the normal left lens. Contrast enhancement to indicate an intraocular tumour is evident

MR imaging of the brain and orbits revealed the right eye typically is normally sized with an abnormal funnel-shaped structure extending from the retrolental area to the region of the optic disc (Figs 2).

After correlating the clinical presentation and imaging findings, we made a diagnosis of unilateral PHPV.

DISCUSSION

As embryonic development progresses, the primary vitreous and hyaloid vasculature are replaced by avascular secondary vitreous. This progressive regression starts in the 20th week of gestation and completes at birth [3]. Rarely, this regression fails and leads to the persistence of primary vitreous and hyaloid vasculature. PHPV is usually unilateral. Bilateral cases are rare and are mostly associated with certain other congenital disorders like Norrie disease, trisomy 13, 15, and 18 [2]. A study performed by Pollard revealed that the bilateral subtype of PHPV accounts for only 2.4% of all such cases [4].

The most common clinical signs and symptoms of PHPV include corneal haziness, leucocoria with watery discharge, microphthalmia, and decreased vision [5]. Based on the involvement of the ocular segment, PHPV is classified into three types: anterior, posterior, and mixed [1]. The posterior form is rare and accounts for only 22% of all cases whereas the anterior and the mixed form make up 36% and 42% of all cases respectively [6]. According to this classification, our case fell under the mixed type of unilateral PHPV.

Ocular ultrasound shows, as in our case, an echogenic cord extending from the posterior surface of the lens to the optic disc. It also shows the decreased axial length of the globe. Ocular ultrasound can also help to rule out calcification, which was not found in this child [7]. CT scan can aid in better visualization of

the underlying pathologies and provisional diagnosis of PHPV. Although CT scan can better demonstrate all the findings of ultrasonography, it also carries the risk of radiation exposure [8].

MR imaging may show microphthalmos, shallow anterior chamber, enhancing retrolental tissue, persistent Cloquet's canal and retinal detachment [9]. The combination, as in our case, of persistent Cloquet's canal and retrolental soft tissue may result in a characteristic funnel-shaped mass occupying the globe. Fluid-fluid levels may be seen in the vitreous, consistent with the development of blood products, although in this case they are not observed [9]. There may also be enlargement of the anterior chamber, thought to be related to elongation of the ciliary processes, possibly through the mechanism of leaky vessels [10].

The differential diagnosis (radiographic and clinical) of PHPV includes retinoblastoma, Coats' disease, retinopathy of prematurity, optic nerve drusen and toxocariasis, all of which may be associated with leukocoria at presentation [11]. Retinoblastoma is a malignant tumour and is the most common intraocular tumour in children. Retinoblastoma may be familial or sporadic. Hereditary retinoblastoma is bilateral in up to 85% of cases [10, 11]. The presence of calcifications, together with the nodular appearance of the vast majority of cases, helps to differentiate retinoblastoma from other entities. Retinoblastoma typically follows grey matter signal characteristics and is highly enhancing on gadolinium chelation [11]. In contrast to PHPV, which usually presents with microphthalmia, the eye in patients with retinoblastoma is typically normal in size or enlarged [9].

Coats disease is a congenital, non-hereditary, unilateral vascular malformation of the retina that is commonly associated with a normal sized globe and subretinal exudates. These subretinal exudates can lead to massive exudative retinal detachments [2]. Coats disease usually presents in boys between the ages of 6-8 years, which helps to distinguish Coats disease from PHPV, which is present at birth; however, the age of presentation can range from 4 months to 7 years in patients with Coats disease. The disease is unilateral in 80-90% of patients [7]. Imaging features in the early stages of Coats disease may not be apparent on CT or MR imaging; however, thickening of the retina may be seen in the later stages of the disease. This can lead to retinal detachment, which appears as homogeneous hyperattenuation on CT images and high T1 and T2 signal intensity on MR images. Notably, calcifications are rare and there is no enhancement of the subretinal exudates, which contain high levels of cholesterol and protein from the immature telangiectatic retinal vessels [11]. The absence of microphthalmos may help to differentiate Coats disease from PHPV [2].

Retinopathy of prematurity is caused by premature arrest of vasculogenesis, which can lead to abnormal fibrovascular proliferation at the junction of the vascular and avascular retina. While ROP is associated with microphthalmia, it is usually bilateral, although it may affect both eyes asymmetrically. This process usually develops in premature infants with a history of supplemental oxygen exposure, although this is not reported in this case. Imaging findings typically show bilateral microphthalmos. There may be increased signal intensity on T1 and proton density weighted images due to associated retinal detachment. Calcifications are rare but may be seen in very advanced cases [11].

The management of PHPV depends on the age of the patient at the time of diagnosis and extension of the pathology. Mild cases can be managed conservatively due to their benign nature. Moderate and severe cases that are diagnosed late require surgical interventions. These interventions include anterior and posterior capsulotomy along with anterior vitrectomy [8].

CONCLUSIONS

PHPV is the second most common cause of leukocoria after retinoblastoma. A detailed ophthalmic examination together with imaging can help in the early diagnosis of the disease. Early recognition, together with surgical intervention in severe cases, is key to a successful outcome.

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