CASE REPORT

Bilateral persistent fetal vasculature: mimicker of retinoblastoma

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SUMMARY

We describe a case of bilateral persistent fetal vasculature (PFV) in a 3-month-old boy who presented with bilateral white pupillary reflex and a possible diagnosis of retinoblastoma. On ultrasonography, there was an echogenic band in the posterior segment of both eyes which connected the posterior surface of the lens capsule to the optic disc. Colour Doppler revealed the presence of vascularity in the band along its entire length. No calcification or mass lesion was seen. These findings are diagnostic of PFV. Most cases of PFV are unilateral and sporadic in nature and closely mimic retinoblastoma. Although rare, PFV should be considered in the differential diagnosis when examining a case of bilateral leukocoria. Paediatricians should be aware of this rare but serious entity.

BACKGROUND

Persistent fetal vasculature (PFV), previously called persistent hyperplastic primary vitreous (PHPV), is a rare vitreoretinal malformation characterised by a retrolental fibrovascular mass due to defects in primary vitreous regression.¹ It is the second most common cause of acquired cataract during the first year of life.² It is unilateral in around 90%–95% of cases and sporadic in nature. When bilateral (2.4%–11%), it is commonly associated with other conditions like Norrie's disease, Warburg syndrome, Patau syndrome or retinal dysplasia.^{3,4} It usually presents as leukocoria, microphthalmia and cataract. It is one of the most important differential diagnoses of retinoblastoma.⁴ Often it is a diagnostic challenge for paediatricians and ophthalmologists to differentiate PFV from retinoblastoma. However, PFV must be considered as a differential diagnosis when evaluating a case of leukocoria. It has characteristic radiological findings on Doppler ultrasonography and hence can be reliably differentiated from retinoblastoma. Therefore, such children must be evaluated with a detailed ophthalmic examination along with ultrasonography before CT is requested in order to avoid unnecessary radiation exposure. We present the case of a 3-month-old boy referred to us for imaging with a clinical suspicion of retinoblastoma.

CASE PRESENTATION

A 3-month-old boy was taken to the local paediatrician with a complaint of bilateral white pupillary reflex. At the initial evaluation the possibility of bilateral retinoblastoma was considered and

the patient was referred for a CT scan of the orbit and brain. We reviewed the patient's history and performed a detailed clinical examination. The patient was a term, normal birthweight baby with an uneventful perinatal and postnatal period. His parents had noticed a white reflex in both eyes since the age of 1 month. There was no history of ocular trauma or oxygen administration during the neonatal period. On examination, there was bilateral leukocoria (figure 1), but visual fixation and following was preserved in both eyes. There was no nystagmus or squint. Systemic examination (including neurological) was normal. For a child of this age, retinoblastoma was the strongest possibility, but we thought that it could also be PFV and so consulted an ophthalmologist.

INVESTIGATIONS

In consultation with an ophthalmologist, an ultrasound B-scan was performed to identify the underlying pathology. The ultrasound study was performed with a high-frequency transducer operating at 10 MHz (Prosound Alpha 7; Hitachi Aloka Medical America). Greyscale evaluation revealed a medium increased echogenicity band in the posterior segment of both globes extending from the posterior surface of the lens capsule to the optic disc. Colour Doppler revealed the presence of vascularity along the entire length of the band (figure 2a,b). No calcification or mass lesion was seen.

DIFFERENTIAL DIAGNOSIS

PFV is one of the most important differential diagnoses of retinoblastoma, which is the most common cause of bilateral white pupillary reflex in children. As our findings were diagnostic of PFV, bilateral PFV was considered in this case.

OUTCOME AND FOLLOW-UP

The parents were informed and counselled about the diagnosis, treatment options and prognosis. They were happy with the news that it was not malignant. The patient was referred to an ophthalmologist for further management but could not be followed up further.

DISCUSSION

PFV was first described by Reese in 1955 in his Jackson Memorial Lecture.⁵ In the embryo, the primary vitreous is formed during the first month of development and is supplied by the hyaloid blood

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Figure 1 Bilateral white pupillary reflex.

system. The hyaloid artery generally begins to regress during the formation of the avascular secondary vitreous at around 9 weeks. Incomplete regression of the embryonic vitreous and the hyaloid vasculature leads to this condition.⁴

PFV is usually found in otherwise healthy, full-term infants, however, systemic and neurological abnormalities are also described in the literature. Its severity can range from an isolated Mittendorf dot to more severe forms of retrolenticular membranes, retinal dysplasia or detachment.⁶ The most common presenting signs and symptoms are leukocoria, poor vision and strabismus.⁷ PFV is classified into three types: anterior, posterior, or a combination of anterior and posterior. Complications of PFV can be cataract, intraocular haemorrhage, secondary

glaucoma, lens capsule rupture, traction retinal fold, and subsequent phthisis bulbi.⁴

PFV has a very characteristic ultrasonographic appearance, which helps to reliably differentiate it from other conditions with white pupillary reflex. On 2D ultrasonography, an echogenic inhomogeneous band is seen which extends from the posterior surface of the capsule of the lens to the optic disc. On colour Doppler, arterial flow may be seen in this band which represents persistent hyaloid artery.⁸ CT findings of PFV are microphthalmia, a small or irregular lens, infiltration of the vitreous cavity giving the appearance of a hyperdense vitreous body, and tubular intravitreal density (Cloquet's canal). Calcification is characteristically absent and retinal detachment may be noticed.⁴ MRI is considered superior to CT scanning in the diagnosis of PFV. Anterior-type PFV is characterised by a shallow or collapsed anterior chamber along with a retrolental vascular membrane which is hyperintense on T2 images. The findings described in posterior-type PFV are microphthalmia, tubular Cloquet's canal, funnel-shaped retinal detachment, hyperintense subretinal fluid, hypointense retrolental mass and vitreous haemorrhage. In one study, funnel-shaped retinal detachment and a retrolental mass were the most common findings of PFV on MRI.⁹

The differential diagnosis of PFV includes retinoblastoma, Coat's disease, vitreoretinal dysplasia, ocular toxocariasis, retinal astrocytoma and retinopathy of prematurity (ROP). Retinoblastoma is the most serious of these and as it closely resembles PFV, must be excluded in all cases of leukocoria. ROP is a disease of prematurity but can occur in late preterm and term babies if they have received an excess of oxygen/mechanical ventilation in neonatal life. It a preventable causes of visual morbidity and is of public health importance. Almost all these entities can be differentiated by ocular examination along with ocular imaging.

Management and prognosis depend on the extent of disease, age at diagnosis and timing of surgery. Mild PFV can run a relatively benign natural course without surgery, while moderate and severe cases need surgical interventions aimed primarily at visual rehabilitation. Purely anterior PFV has a good visual outcome

Figure 2 a,b Ultrasound along with Doppler showing the fibrous vascular band extending from the posterior aspect of the lens to the optic disc.

with aphakic correction and amblyopia therapy, in contrast to anteroposterior PFV where vision may be unsalvageable.⁶

Although rare, bilateral PFV should be considered in the differential diagnosis when evaluating a case of bilateral leukocoria. Retinoblastoma must be differentiated from this entity with the help of imaging.

Learning points

- ▶ Retinoblastoma is the most common and serious cause of bilateral leukocoria in infancy, but other differential diagnoses must be kept in mind.
- ▶ Persistent fetal vasculature (PFV) is a common, but easily overlooked, mimicker of retinoblastoma.
- ▶ Ultrasound is extremely useful for diagnosing PFV and must be performed before advanced imaging such as CT and MRI.
- ▶ Paediatricians should be aware of this rare but important entity.

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