Unilateral anterior persistent fetal vasculature in a child with blepharophimosis-ptosis-epicanthus inversus syndrome: A surgical challenge

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Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) is a rare autosomal dominant genetic disease. It is clinically characterized by four major features; blepharophimosis, ptosis, epicanthus inversus, and telecanthus. We report a case of a 1-year-old female with BPES with unilateral anterior persistent fetal vasculature (PFV). On examination, she was found to have all the clinical features of BPES, along with calcified and partially absorbed cataract with elongated ciliary processes in her left eye. B-scan of left eye showed attached retina with no evidence of posterior PFV. Systemic examination was normal. She underwent cataract surgery with primary posterior capsulotomy with intraocular lens implantation under general anesthesia. Literature search did not reveal any previous reports of unilateral anterior PFV and BPES. The clinical features, other associations, and the difficulties in the surgical management of this condition are discussed.

Key words: Anterior persistent fetal vasculature, blepharophimosis-ptosis-epicanthus inversus syndrome, congenital cataract

Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) (Online Mendelian Inheritance in Man accession number #110100) is a rare autosomal dominant genetic disease characterized by 4 major features; blepharophimosis, ptosis, epicanthus inversus, and telecanthus.[1] Persistent fetal vasculature (PFV) is a congenital developmental anomaly of the eye caused by the failure of regression of the primary vitreous.[2] The clinical ocular manifestations include microphthalmia, progressive cataract, retrolental fibrovascular tissue, persistent hyaloid vessel remnants, and tunica vasculosa lentis remnants.[3] We describe a case of BPES associated with anterior PFV. Literature search did not reveal any previous reports of BPES associated with unilateral anterior PFV.

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Case Report

A 1-year-old girl born of nonconsanguinity at term presented to us with the complaints of squinting of both eyes since birth and white pupillary reflex in the left eye noticed at 2 months of age. There was no history of similar complaints in the family.

On examination, she was not cooperative for visual acuity testing with Teller Acuity Cards. She was able to fix and follow light and objects with her right eye. There was strong resistance to occlusion of the right eye, proving that the vision in the left eye was very poor. External examination revealed epicanthus inversus, telecanthus, and ptosis [Fig. 1]. Horizontal fissure width was 20 mm in both eyes, and the inner intercanthal measurement was 42 mm.

On examination, there was pseudoesotropia due to the presence of epicanthal folds.

Anterior segment examination of the right eye was normal. Left eye showed elongated ciliary processes with partially absorbed calcified cataract [Fig. 2a].

Fundus examination of the right eye was normal. There was no view of the fundus in the left eye. B-scan of the left eye confirmed the anterior PFV and the retina was attached. Retinoscopy of the right eye showed a refractive error of +7.0DS. Retinoscopy of the left eye was not possible due to the cataract. The systemic evaluation was normal.

Diagnosis of anterior PFV with BPES was made based on the clinical features. Lens aspiration with primary posterior capsulotomy and anterior vitrectomy with posterior chamber intraocular lens (PCIOL) in the left eye was done under general anesthesia under guarded visual prognosis.

Two side ports were made at 2 and 10 o'clock. A corneal tunnel was made at 12 o'clock. Since the anterior capsule was calcified and fused with the posterior capsule and could not be separated, it had to be cut with micro scissors. An outcome was then used to make an opening in the visual axis. A foldable hydrophobic three piece PCIOL was inserted into the ciliary sulcus (20% under correction was done). There was no intraoperative or postoperative complication.

Difficulties were encountered during surgery, due to limited surgical space because of the short horizontal palpebral fissure and epicanthal folds. There were difficulties in doing

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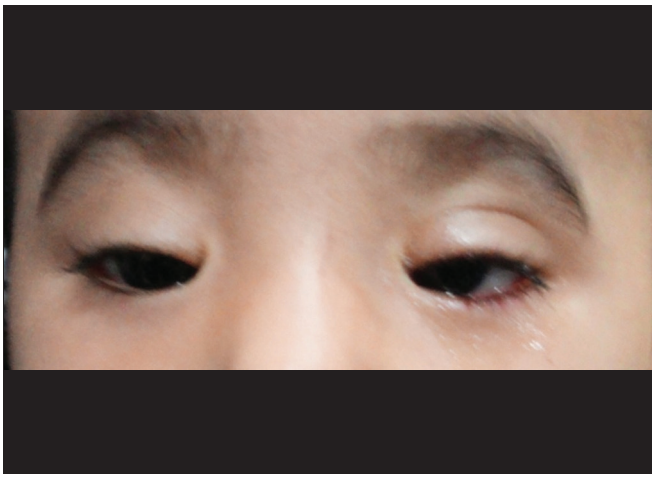

Figure 1: External photograph of the eyes showing blepharophimosis, ptosis, epicanthus inversus, and telecanthus

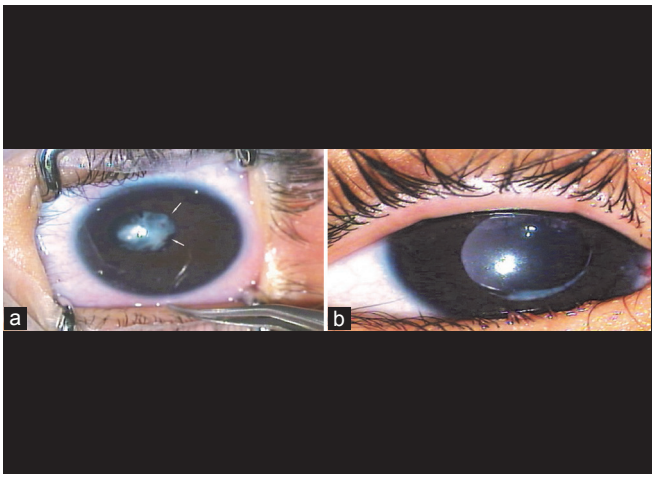

Figure 2: (a) Clinical photograph of the left eye taken just before surgery showing a rigid pupil, elongated ciliary processes (white arrows) and a calcified, partially absorbed cataract. (b) Clinical photograph of the left eye with pseudophakia and a clear visual axis

continuous curvilinear capsulorhexis, cortex aspiration, and primary posterior capsulotomy due to the presence of a rigid pupil, calcified anterior capsule, and partially absorbed calcified cataract.

Postoperatively, [Fig. 2b] the refraction in both eyes was + 7DS. She was advised glasses and part-time occlusion of the right eye for 4 h in a day. She was lost to follow-up 1 year after surgery.

Discussion

BPES is diagnosed clinically and can be categorized into two types. BPES Type I is characterized by the four eye findings along with female premature ovarian failure. BPES Type II also has the four eye findings but is not associated with premature ovarian failure in the female patients.[1]

Our patient had all the four clinical features of BPES. The normal palpebral fissure width in a 1-year-old child is about 23 mm[4], and the intercanthal distance ranges from 26 to 35 mm.[5] Our patient had a short horizontal palpebral fissure width (20 mm) and a large intercanthal distance (42 mm).

Other ophthalmic manifestations associated with BPES include strabismus[6] microphthalmos, retinochoroidal and optic disc coloboma[7] lacrimal drainage apparatus abnormality, congenital alacrima,[8] facial dysmorphism and congenital cataracts[5] and with bilateral blue dot cataracts with retinitis pigmentosa and primary inferior oblique overaction.[9] To the best of our knowledge, anterior PFV has not been described with BPES in literature. It is difficult to explain the association of anterior PFV with blepharophimosis syndrome since different genes on different chromosomes code for the two diseases (FOXL2 gene on chromosome 3q23 region codes for BPES[1] and a gene on the chromosome 10q11-q21 region codes for PFV).[2] This could be a coincidental finding rather than an association. Further studies are needed to prove the association.

Treating the cataract in anterior PFV is by itself a challenge because of the leathery fibro-vascular plaques.[10] Complications associated with the surgery are intraocular hemorrhage, retinal tears, secondary glaucoma, extensive inflammatory response, and visual axis opacification.[10] When this is associated with BPES, the surgery becomes more demanding because of the small horizontal palpebral fissure width and less working space. A lateral canthotomy can be considered to overcome this problem. A modified approach to cataract surgery had to be adopted because of the partially absorbed, membranous, and calcified nature of the cataract. Since the anterior capsule was stuck to the posterior capsule and could not be separated, capsulectomy with micro scissors was preferred. An outcome had to be used to enlarge the opening in the visual axis.

Conclusion

This is a unique case of BPES with a unilateral PFV significant enough to need surgical intervention. The surgeon should be aware that the surgery could be technically more challenging due to the associated short horizontal palpebral fissure and epicanthal folds.

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Conflicts of interest

There are no conflicts of interest.

References

1. Allen CE, Rubin PA. Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES): Clinical manifestation and treatment. Int Ophthalmol Clin 2008;48:15-23.
2. Shastry BS. Persistent hyperplastic primary vitreous: Congenital malformation of the eye. Clin Experiment Ophthalmol 2009;37:884-90.
3. Cheng LS, Kuo HK, Lin SA, Kuo ML. Surgical results of persistent fetal vasculature. Chang Gung Med J 2004;27:602-8.
4. Walia BN, Bhalla AK. Palpebral fissure length in normal infants. Indian Pediatr 1989;26:57-60.
5. Keating PD, Hepler RS. Blepharophimosis and acquired somato-facial dysmorphism. Associated with congenital cataracts. Arch Ophthalmol 1969;82:1-3.
6. Dawson EL, Hardy TG, Collin JR, Lee JP. The incidence of strabismus and refractive error in patients with blepharophimosis, ptosis and epicanthus inversus syndrome (BPES). Strabismus 2003;11:173-7.

7. Lee LR, Sullivan TJ. Blepharophimosis syndrome: Association with colobomatous microphthalmos. Aust N Z J Ophthalmol 1995;23:145-7.
8. Athappilly GK, Braverman RS. Congenital alacrima in a patient with blepharophimosis syndrome. Ophthalmic Genet 2009;30:37-9.
9. Vedantham V, Jethani J, Agarwal A, Vijayalakshmi P. Retinitis pigmentosa associated with blepharophimosis, blue dot cataract and primary inferior oblique overaction: A new syndrome complex? Indian J Ophthalmol 2007;55:150-1.
10. Tartarella MB, Takahagi RU, Braga AP, Fortes Filho JB. Persistent fetal vasculature: Ocular features, management of cataract and outcomes. Arq Bras Oftalmol 2013;76:185-8.