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Case report

Bilateral cataract in a child with blepharophimosis-ptosis-epicanthus inversus syndrome: A surgical challenge

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ABSTRACT

Introduction: Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) is a rare autosomal dominant genetic disorder characterized by complex orbito-palpebral anomalies. We report a rare case of BPES associated with bilateral congenital cataract.

Observation: This study reports the case of a 6-month-old infant with BPES in whom a bilateral congenital cataract was diagnosed, after the parents noticed leukocoria and signs of poor vision in their child. No other ophthalmologic manifestations commonly associated with this syndrome were found. The infant underwent cataract surgery first, with lens phacoaspiration and posterior capsulotomy coupled with anterior vitrectomy and placement of a 3-piece foldable hydrophobic posterior chamber lens in the capsular bag. The surgery was a real challenge due to the orbito-palpebral anomalies that limited a small surgical space, and the placement of the IOL was a matter of discussion.

Discussion: Publications on the association of congenital cataract with BPES are very rare. The link between these two anomalies is difficult to establish since different genes on different chromosomes code for the two diseases. A lateral canthotomy can be considered to overcome the surgical difficulties due to the reduced working space. The surgical management of pediatric cataract varies in the literature.

Conclusion: This case highlights the difficulty of cataract surgery in children, even more so when associated with BPES, and the challenge of improving vision in these children given the high risk of amblyopia.

1. Introduction

Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) is a rare autosomal dominant genetic disorder characterized by a complex of four orbito-palpebral anomalies: blepharophimosis, ptosis, epicanthus inversus, and telecanthus [1]. No differences in prevalence based on sex, race, or ethnicity have been reported. The prevalence of BPES remain unknown [2], however the condition is not very uncommon and more than 150 families with the blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) have been described [3] but its association with congenital cataract has been rarely reported. Other ocular signs more frequently present include squint, nystagmus, microphthalmus, microcornea and stenosis of the lacrymal canaliculi [3].

.Congenital cataract corresponds to a loss of lens transparency related to genetic mutations. it is known as a treatable cause of childhood blindness.

We report a rare case of BPES associated with bilateral congenital cataract and discuss the clinical features and difficulties in the surgical management of this unusual association.

This study has been reported in accordance with the SCARE criteria [4].

2. Observation

The present study reports the case of a 6-month-old infant, brought back by his parents to an ophthalmologic consultation for signs of poor vision, with bilateral leukocoria. There was no consanguinity between the parents, the infant was an only child.

The pregnancy was not monitored but was presumed to be full term, the delivery took place vaginally, without incident, in a hospital setting, there was no instrumental delivery (forceps). The infant had small palpebral fissure with drooping eyelids since birth and there was a family history of similar eyelid appearance in his paternal cousin.

The ophthalmological examination revealed a vicious attitude of the

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child with head tilted backwards, the pursuit and threat reflexes were absent, the examination of the eyelids showed an epicanthus inversus. The distance between the two medial canthus was 38 mm defining the telecanthus, the inner canthal distance at 6 months being at 2,2 mm ± 0,5 mm [5] (Fig. 1).

The vertical and horizontal diameter of the palpebral fissure was 4 mm and 12 mm, respectively, while the normal width of the palpebral slit varies between 18 and 23 mm [6], which is suggestive of blepharophimosis.

The palpebral crease of the upper eyelid was absent (Fig. 2).

The infant had horizontal nystagmus. On the cover test, the presence of a true tropia was difficult to assess due to the presence of the telecanthus and the narrow palpebral fissure. The patient had severe bilateral ptosis, with a difficult-to-assess levator course and a flat nasal bridge (Fig. 3).

Corneo-conjunctival examination of both eyes was normal with a corneal diameter of 9.5 mm. The lenticular examination showed a nuclear cataract with no passage to the fundus. Ocular ultrasound showed a normal posterior segment and an axial length of 19 mm.

Visual evoked potential and electroretinogram were normal. A genetic investigation was requested but not done due to lack of funds.

Because of the high risk of amblyopia, which is already increased by the orbito-palpebral malformation, and in order to give this child a better chance of recovering good vision after eyelid surgery, he underwent cataract surgery first, with lens phacoaspiration and posterior capsulotomy coupled with anterior vitrectomy.

The placement of an artificial implant was a matter of discussion due to the borderline size of the cornea. The decision to implant was made intraoperatively, the IOL power calculation was done with the ultrasound biometry, with the applanation technique, and the undercorrection was 20%. A 3-piece foldable hydrophobic posterior chamber lens was implanted in the capsular bag.

There were surgical difficulties, due to the small surgical space limited by the epicanthal folds. The continuous curvilinear capsulorhexis was difficult to perform because of the limitation of rotational movements caused by the small palpebral slit and the blocking of the rhexis forceps with the eyelid retractor (Fig. 4).

The surgery was then longer, with small jerky movements.

Postoperative follow-up was good, with spherical refraction at +1D and cylindrical refraction at −0.75D in both eyes. The fundus was normal without any sign of retinitis pigmentosa.

At 6 months postoperatively, the refraction evolved to a myopic shift of -1D which may correspond to a keratometric flattening in relation with the increase of the axial length.

A palpebral reconstruction surgery will be scheduled around the age of 5 or 6 years.

3. Discussion

Blepharophimosis was first described by VonAmmon in 1841, then in1889 it was described by Vignes, and since then several cases have been published [7].

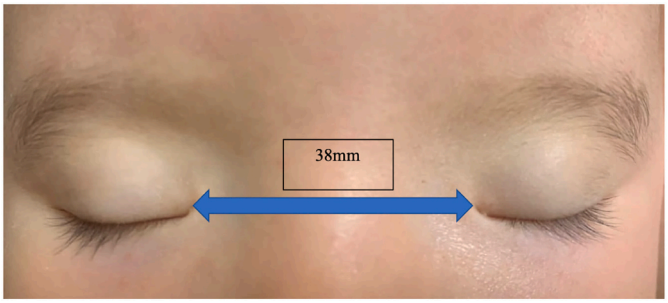

Fig. 1. Image of the large inner intercanthal distance, 38 mm (télécanthus).

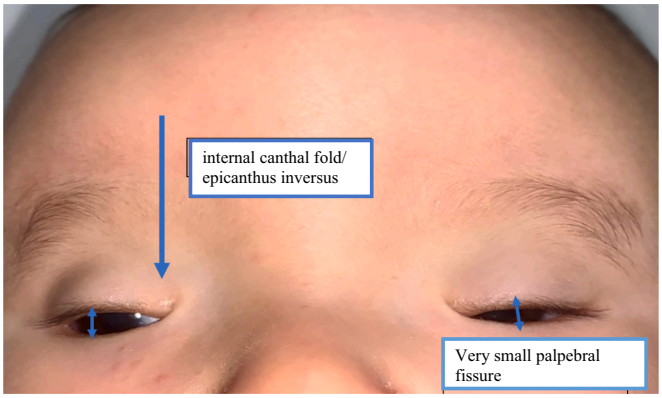

Fig. 2. Image of the eyelids of the 6 months-old infant where we can see the large inner intercanthal distance (télécanthus), the epicanthus inversus, and the shortening of the horizontal orbital fissure (blepharophimosis).

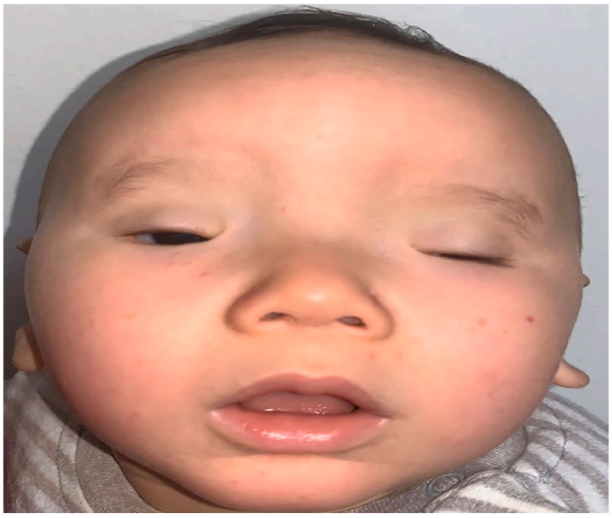

Fig. 3. Maximum palpebral opening with vicious attitude: rejection of the head backwards, elevation of the eyebrows, and recruitment of the frontal muscle. Flat and low nasal bridge.

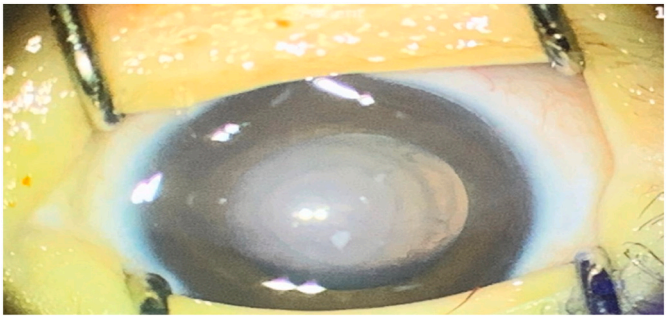

Fig. 4. Photograph showing the small palpebral fissure limiting the surgical space.

BPES can be classified clinically into two types. Type I BPES is characterized by the four ocular manifestations: blepharophimosis, ptosis, epicanthus inversus and telecanthus [8] as well as premature ovarian insufficiency in female patients.Type II BPES is limited to ocular manifestations without other associated manifestations [9]. Our patient had all the clinical features and was classified as BEPS type II.

Normal width of the palpebral slit varies between 18 and 23 mm [6]

and the normal inner canthal distance in Centimeters by Age: Mean (2SD), is detailed below [5]:

- Premature newborn 1.6 (0.4)
- Full-term newborn 2.0 (0.4)
- 1–6 months: 2.2 (0.5)
- 7–12 months: 2.5 (0.5)
- 13–18 months: 2.5 (0.6)
- 19–24 months: 2.5 (0.4)
- 25–30 months: 2.6 (0.6)

Our patient had a short horizontal diameter of the palpebral slit (12 mm) and an intercanthal distance at 38 mm.

Other ophthalmologic manifestations associated with BPES can be found such as microphthalmia, strabismus [10], retinal coloboma [8], alacrymia [4] and congenital cataract [11].

Publications on the association of congenital cataract with BPES are very rare [8]. It is difficult to establish the link between these two anomalies since different genes on different chromosomes code for the two diseases (the FOXL2 gene on the region of chromosome 3q23 for BPES [12] and a gene on the region of chromosome 10q11-q21 for cataract and primary vitreous persistence) [11]. Further genetic studies are needed to establish a clear association, which remains possible.

Treating congenital cataracts when associated with BPES becomes a real surgical challenge [11]. The surgery is more difficult because of the small palpebral fissure and the reduced working space. A lateral canthotomy can be considered to overcome this problem. Other situations can make surgery even more difficult, such as a cataract with a calcified capsule or when associated with persistent fetal vasculature [13].

It is controversial to implant an IOL at the time of cataract surgery in infants, versus to leave the child aphakic with a secondary procedure for IOL implantation later in childhood [14].

Pediatric cataract surgery is evolving with advances in microsurgical techniques. Currently there is an increasing trend toward IOL implantation in children with increasing evidence of better visual outcomes in infants managed with IOL implantation. More surgeons are starting to implant between 6 months and 1 year [15].

Primary IOL implantation in children <2 years is a safe surgical procedure with excellent long-term results compared with aphakia and secondary IOL implantation after the age of 2 years. The myopic shift is well-controlled, and final visual acuity achieved is reasonably good, and it leads to lower incidence of complications. However, care must be taken in children <6 month-old because the incidence of adverse events is high [16].

Determining IOL power in congenital cataract surgery is an important issue due to increasing axial length and the refractive power of pediatric eyes after cataract surgery and also an additive effect of IOL implantation on eye growth, there are recommendations for under correction of IOL power and hyperopic outcomes so the patient will be postoperatively hypermetropic which will shift to emmetropic during adulthood to reduce the necessity of IOL exchange [17]. Therefore an under correction of about 20% in infants and 10% in toddlers is highly recommended [18].

However, in our case, the visual defects that may occur in childhood cannot be blamed on cataract alone. The orbitopalpebral anomalies encountered in this syndrome are also very amblyogenic.

Timing of eyelid surgery is controversial; it involves weighing the balance of early surgery to prevent deprivation amblyopia and late surgery to allow for more reliable ptosis measurements, the latter of which provides a better surgical outcome, furthermore, ptosis surgery is hampered by the dysplastic structure of the eyelids [2].

4. Conclusion

We reported a case of BPES associated with a bilateral congenital cataract requiring surgery. The surgeon should be aware of the technical difficulty of performing this surgery due to the small palpebral fissure which makes handling difficult.

The frequency of ophthalmologic follow-up should depend on the age of the child, the therapeutic attitude adopted, and the results of the visual acuity evaluation In this case, cataract surgery represents one step, among many others to follow, to reach an optimal visual rehabilitation, eyelid surgery remains necessary at an older age.

It is also important to mention that all women with BPES should see a clinical geneticist and, at puberty, an endocrinologist to evaluate the development of PFO [19].

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Ethical approval

I certify that this kind of manuscript does not require ethical approval by the Ethical Committee of our institution.

Consent

Written informed consent was obtained from the parent's patient for publication of this case report. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

O. Nabih: drafting and writing the article, acquisition of data.
L. Arab: revising the article.
L. Elmaaloum: study design.
B. Allali: study concept.
A. Elkettani: final approval.

Registration of research studies

This is a case report that does not require a research registry.

Guarantor

O. Nabih.

Declaration of competing interest

The authors declare that they have no competing interests.

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