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ORIGINAL ARTICLE

A One-Stage Correction of the Blepharophimosis Syndrome Using a Standard Combination of Surgical Techniques

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Abstract
Background The aim of this study was to evaluate the efficacy of a one-stage treatment for the blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) using a combination of standard surgical techniques.
Methods This is a retrospective interventional case series study of 21 BPES patients with a 1-year minimum follow-up period. The one-stage intervention combined three different surgical procedures in the following order: Z-epicanthoplasty for the epicanthus, transnasal wiring of the medial canthal ligaments for the telecanthus, and a bilateral fascia lata sling for ptosis correction. Preoperative and postoperative measurements of the horizontal lid fissure length (HFL), vertical lid fissure width (VFW), nasal intercanthal distance (ICD), and the ratio between the intercanthal distance and the horizontal fissure length (ICD/HFL) were analyzed using Student's *t* test for paired variables.
Results The mean preoperative measurements were 4.95 ± 1.13 mm for the VFW, 20.90 ± 2.14 mm for the HFL, 42.45 ± 2.19 mm for the ICD, and 2.04 ± 0.14 mm for the ICD/HFL ratio. The mean postoperative measurements were 7.93 ± 1.02 mm for the VFW, 26.36 ± 1.40 mm for the HFL, 32.07 ± 1.96 mm for the ICD, and 1.23 ± 0.09 mm for the ICD/HFL ratio. All these values and their differences were statistically significant ($P < 0.0001$). All of the patients developed symmetric postoperative inferior version lagophthalmus, a complication that tended to decrease over time.
Conclusion One-stage correction of BPES is safe and efficient with the surgical techniques described.

Keywords Blepharophimosis · BPES · Epicanthus · Telecanthus ptosis

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Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) is characterized by a horizontal and vertical shortening of the lid fissure, with a classic triad represented by epicanthus, telecanthus, and blepharoptosis. This syndrome is inherited as an autosomal disease and it is classified into two subtypes: (1) BPES that causes female infertility and hypogonadism, transmitted only by men with complete penetrance, and (2) BPES that can be transmitted by both sexes and is characterized by incomplete penetrance (96.5%) [1].

The syndrome also can be manifested as a result of new genetic mutations and sporadic cases without family history of the disease can occur. The first study about the involvement of chromosome 3 in a patient who, among

numerous phenotypic changes, presented the features of the syndrome was published in 1981 [3]. Several studies confirm the presence of mutations such as deletion or translocation of the *FOXL2* gene, which maps to chromosome 3q21–24 [2].

The global prevalence of BPES has not been precisely established but various authors report an incidence of 1/50,000 syndrome carriers among the newborn [2]. The prevalence is higher in Asian than in Caucasian populations [4].

The majority of authors recommend a multiple-stage surgical correction for BPES [5–9]. Some reports defend a single-stage surgical correction of this complex syndrome, although the surgical techniques used in those reports vary widely [4, 10–13]. Therefore, there is no unanimous consensus on BPES correction with respect to either staging or choice of surgical technique.

The purpose of this report is to determine the efficacy of a one-stage BPES correction using three standard surgical techniques in the following order: Z-epicanthoplasty [14], transnasal wiring of the medial canthal ligaments [15], and a final bilateral lid suspension to the frontalis muscle with fascia lata [16].

Methods

This retrospective noncomparative interventional case series study was performed on 43 BPES patients who were treated between January 1992 and December 2007 at the Oculoplastic Departments of the Ivo Pitanguy Institute and Antonio Pedro University Hospital of the Federal Fluminense University. This study was approved by the Ethics for Research Committee of the Federal Fluminense University School of Medicine and informed consent was obtained from all patients or their legal representatives.

The inclusion criteria for this study were bilateral BPES with the characteristic classic triad, no previous surgical treatment, and a minimum postoperative follow-up period of 1 year. The exclusion criteria were noncharacteristic BPES eyelid deformities, any previous eyelid or orbital surgery, and a less than 1-year postoperative follow-up period. Twenty-two patients were excluded from the total sample of 43 BPES patients, 12 because of previous surgery and 10 for not having a 1-year follow-up. The remaining 21 patients were included in this study.

All of the patients underwent a routine ophthalmological exam that included visual acuity, eye motility, a fundus exam, biomicroscopy, and an intraocular pressure reading. All patients were checked with orbital and nasal images (routine X-ray and CT scans) to rule out any previous disease.

Twenty-one BPES patients underwent a one-stage bilateral surgical intervention comprising three consecutive procedures: a Z-epicanthoplasty [14], transnasal wiring of the medial canthal ligaments [15], and a final lid/brow suspension with autogenous fascia lata [16]. All procedures were performed by the same surgeon. The horizontal lid fissure length (HFL), the vertical lid fissure width (VFW), the nasal intercanthal distance (ICD), and the ratio between the intercanthal distance and the horizontal fissure length (ICD/HFL) were recorded and analyzed using Student's *t* test for paired variables.

Preoperative and 1-year postoperative photographic images were recorded for comparison. All postoperative complications were rigorously dealt with.

Treatment

Z-Epicanthoplasty and Transnasal Wiring of the Medial Canthal Ligaments

The epicanthus was corrected with the Z-epicanthoplasty procedure as described by Lessa and Sebastiá [14]. The first step of this procedure is to draw a point for the new inner canthus (point 1), which is repeated on the other side so that the future intercanthal distance will be one-half the size of the interpupillary distance, as recommended by Mustardé [17, 18]. Point number 2 is bilaterally drawn adjacent to the preexisting medial canthii, and points 1 and 2 are then joined by a line (Fig. 1). The skin is then incised and the orbicularis muscle is separated until the medial ligament is dissected (Fig. 2).

The telecanthus is then corrected with a transnasal wiring procedure of the canthal ligaments as previously described by Callahan [15]. A small hole is drilled posterior the anterior lacrimal crest, in the lacrimal fossa, and the medial ligaments are simultaneously tied and tightened using a thin 4-0 steel wire suture threaded through the previously drilled hole (Fig. 3).

At this stage points 1 and 2 are approximated with a 6-0 nylon suture which results in a superior and inferior dog-ear skin fold. Two Z-plasties, one above and one below the line of incision, are then marked so that the future scars will run parallel and adjacent to the lid margins (Fig. 4). Then the skin flaps are dissected and transposed (Fig. 5) and finally sutured with a 6-0 nylon or silk suture (Fig. 6).

Lid Ptosis Correction

A bilateral frontalis lid sling with fascia lata, as described by Lexer [16], was finally used to correct lid ptosis. Four strips of fascia (60 × 2 mm) were removed from the middle lateral portion of the thigh and sutured to the upper anterior part of the tarsus. These were then passed beneath

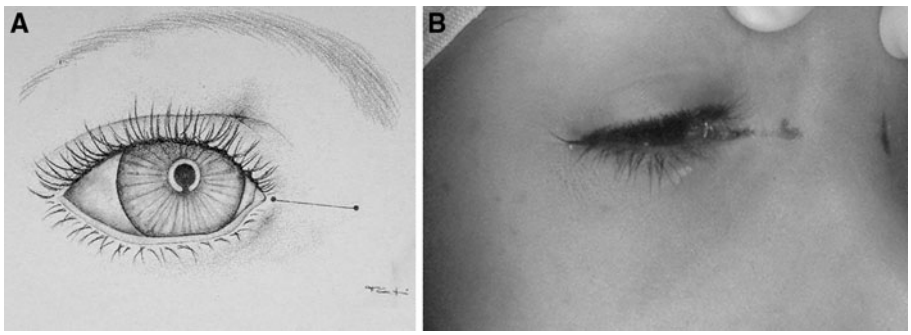

Fig. 1 Marking and joining points 1 and 2 (from [14])

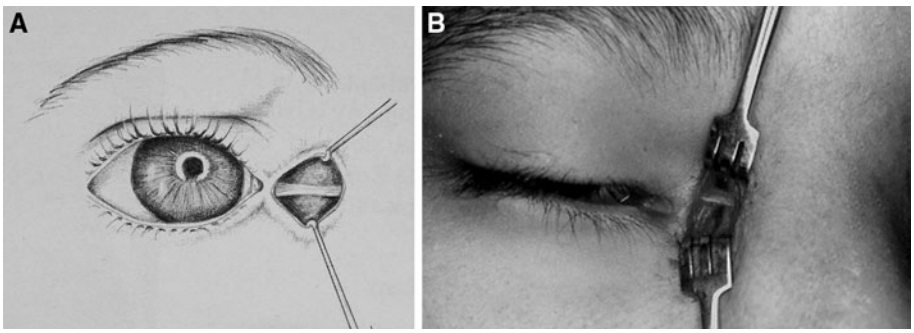

Fig. 2 Dissection of the medial canthal ligament (from [14])

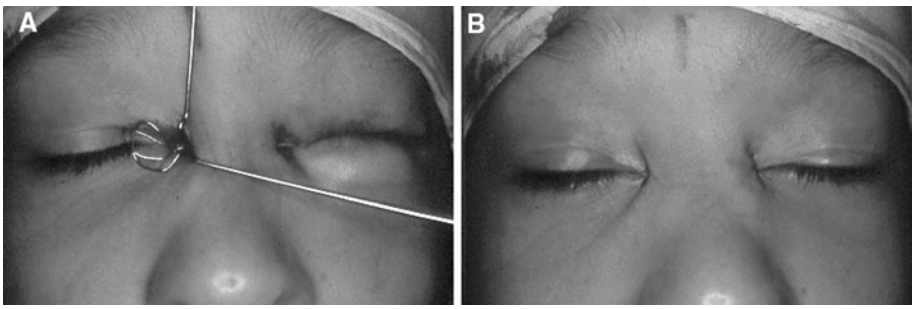

Fig. 3 Transnasal fixation of medial canthal ligaments. **a** Passage of steel wire with the aid of a small drill hole and attachment to the canthal ligaments. **b** Pulling and fixing of steel wires, correcting telecanthus

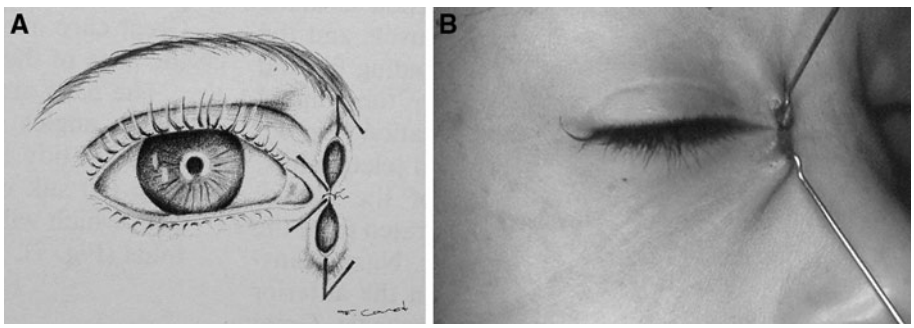

Fig. 4 Union of points 1 and 2 and marking of Z-plasty [14]

the orbicularis muscle and sutured to the frontalis muscle above the brow in the manner of a "W" (Fig. 7). The central arm of the W determines the height of the lid margin and the other two arms of the W are placed to regulate lid contour.

Statistic Analysis

Student's *t* test for continuous paired variables was used to compare the means of the preoperative and postoperative measurements of HFL, VFW, ICD, and ICD/HFL. A

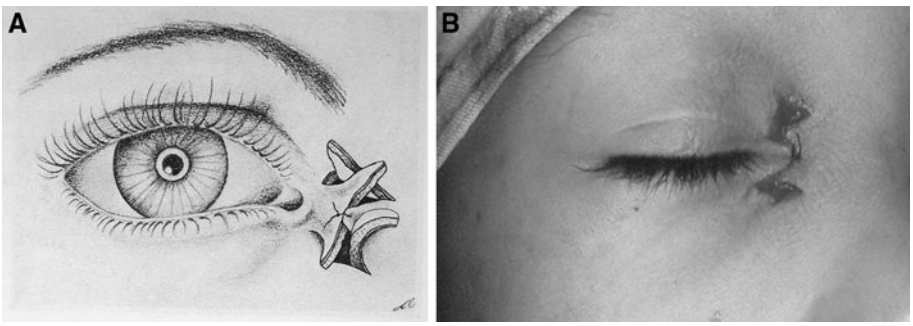

Fig. 5 Incision and transposition of the flaps [14]

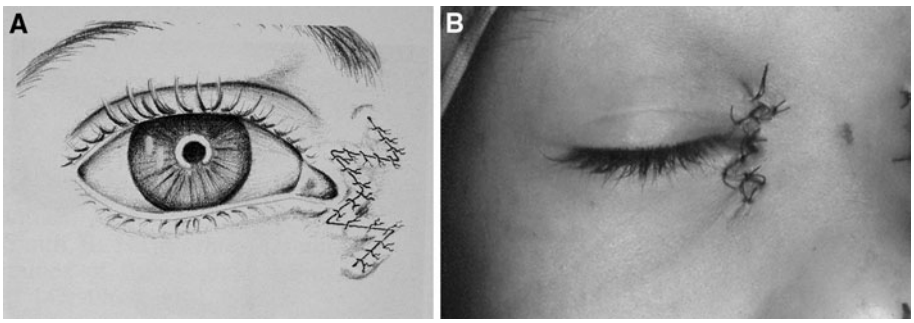

Fig. 6 Suturing of the flaps with 6-0 silk thread (from [14])

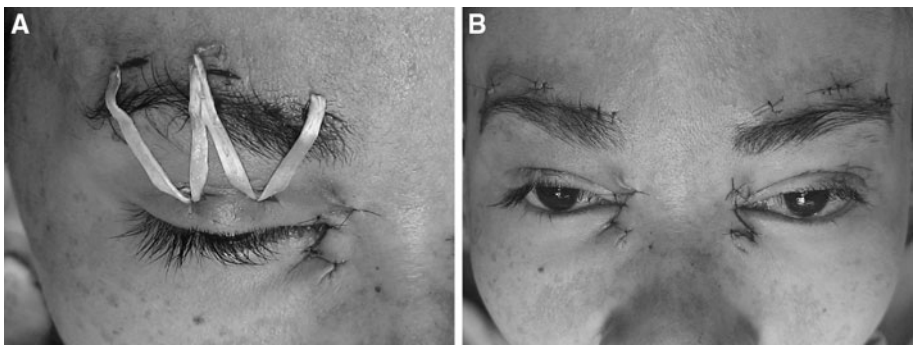

Fig. 7 Removal of thin strips of fascia lata which are set initially on the anterior face of the tarsus. These are passed below the orbicularis muscle and then fixed to the frontalis muscle

$P < 0.05$ level of probability was considered statistically significant. The statistical analysis was performed using the S-PLUS® 8.0 program for Windows (Insightful Corporation, Seattle, WA, USA).

Results

Nine of the 21 patients were male. The age of the total sample ranged from 5 to 42 years with an average of 21.4 years. The follow-up period ranged from 1 year to 11 years 4 months with an average of 4 years 3 months (Figs. 8, 9, 10).

The preoperative and postoperative VFW, HFL, and ICD differences (Table 1) were all statistically significant ($P < 0.0001$). The preoperative and postoperative values for ICD/HFL (Table 2) were also statistically significant. The perioperative complications were limited to nasal bleeding after the transnasal wiring in two patients who required nasal packing for 48 h, but after the removal of the packing progressed normally. The only postoperative complication registered was lagophthalmus with inferior version gazing, which is a frequent finding in lid/brow suspension procedures. No patient showed corneal suffering due to the lagophthalmus, which was more accentuated in the immediate postoperative period and tended to decrease over time (Figs. 8, 10).

Discussion

BPES is a relatively rare, autosomal dominant deformity characterized by overall narrowing of both lid fissures and classically demonstrated by the clinical triad of epicanthus inversus, telecanthus, and severe myogenic ptosis [1, 2].

Fig. 8 Pre- and postoperative appearance of a 37-year-old female patient. **a** Preop photo. **b** Postoperative photo. **c** 3 Months after surgery with moderate lagophthalmus. **d** 6 Months after surgery with lagophthalmus reduction

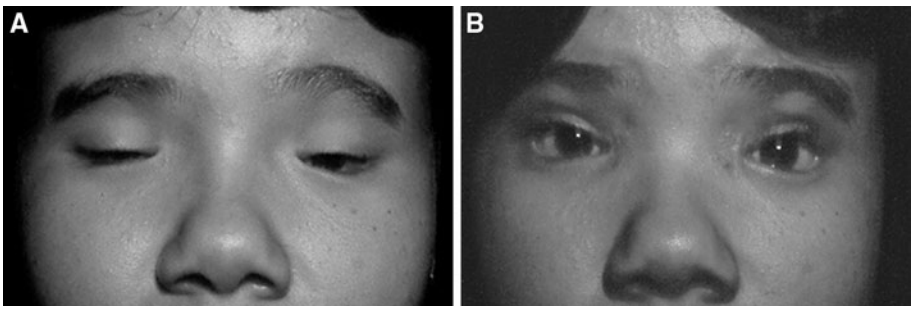

Fig. 9 Pre- and postoperative appearance of a 14-year-old female patient

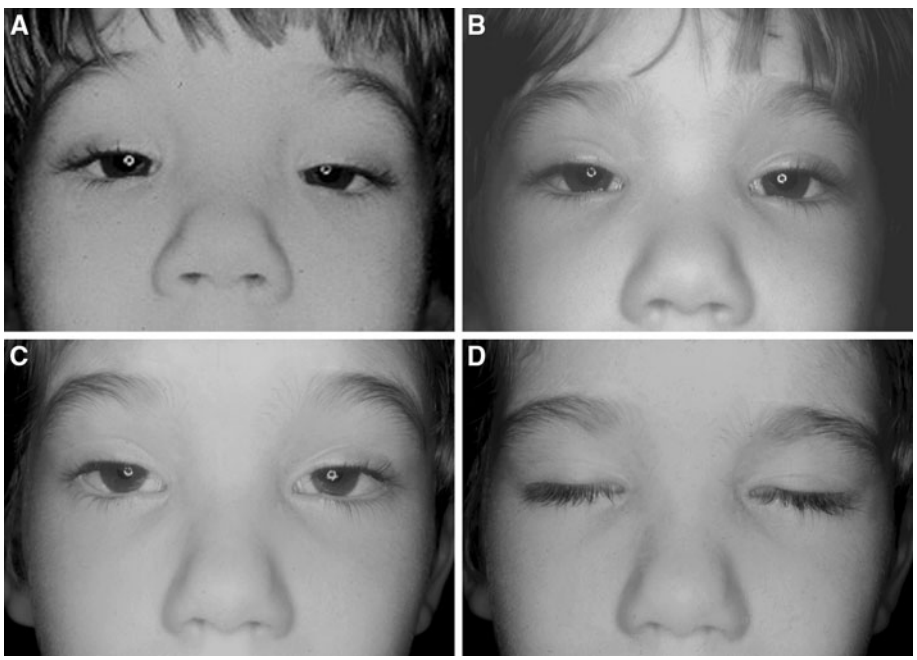

Fig. 10 Pre- and postoperative appearance of a 5-year-old male patient. **a** Preoperative photo. **b** One-year postoperative photo. **c** 4 Years after surgery. **d** 4 Years postoperative photo showing a mild lid-lag lagophthalmus

Table 1 VFW, HFL, and ICD values

	Preoperative (mm)	Postoperative (mm)	*P* value
VFW	4.95 ± 1.13	7.93 ± 1.02	<0.0001
HFL	20.90 ± 1.24	26.36 ± 1.40	<0.0001
ICD	42.45 ± 2.19	32.07 ± 1.96	<0.0001

HFL horizontal lid fissure length, *VFL* vertical lid fissure width, *ICD* nasal intercanthal distance

Table 2 ICD/HFL ratios

	Preoperative	Postoperative	*P*
ICD/HFL	2.04 ± 0.14	1.23 ± 0.09	<0.0001

ICD/HFL ratio between the intercanthal distance and the horizontal fissure length

It is generally accepted by the majority of practicing surgeons that this condition should be corrected in young children unless there is the risk of developing amblyopia due to severe eyelid ptosis. The risk of developing amblyopia in unilateral and asymmetric ptosis is higher than in symmetric ptosis as is found in the majority of BPES patients because in this condition the patient adopts a characteristic elevation and extension of the chin that seems to diminish the risk of amblyopia development. We had five patients with amblyopia. Three of these patients had a small visual deficiency and two had a moderate visual deficiency. The majority of our patients had symmetrical ptosis which we believe is responsible for the small incidence of amblyopia observed in this series. Beaconsfield et al. [19] reported a 56.4% prevalence of amblyopia, although some of his patients, unlike ours, had unilateral or asymmetric ptosis.

Although our average patient age was 21 years old, we agree that BPES patients should be operated on at a younger age, but in our experience this was not always possible due to the delay by the patient in finding and reaching an experienced medical institution. On the other hand, some younger children were not included in the study because they had previous surgery.

Treatment of BPES is difficult and controversial due to the complexity of the defects, the large number of different techniques employed, and the possibility of using a one-stage or a multiple-stage procedure. There are advocates for a single-session surgical correction [4, 11–13] and those who defend a multistage correction [5–10]. A staged approach is recommended by authors with the provision of adequate time between the two stages. The delay is justified because of the opposite direction of the tension created by the two procedures. The authors prefer to correct the epicanthus and telecanthus first and about 6 months later the ptosis.

Various techniques for the epicanthus correction have been described. The most widely used is probably that which was described by Mustardé [17, 18]. Li et al. [7] report better results with the V–Y procedure in older (>10 years) patients. Karacaoglan et al. [11], in association with the other procedures, use a bone graft to the base of the nose with the intent of projecting and stretching the skin of the nose and therefore obtaining a better epicanthus correction. We defend the Z-epicanthoplasty [14] technique because it is simpler to perform and has similar efficacy to the Mustardé [17, 18] procedure from which it was derived. The resulting scars become cosmetically acceptable with time and are well positioned and very similar to those resulting from Mustardé's technique.

There is also controversy about the correction of telecanthus. Some authors use simple attachment to the periosteum [4, 7, 11, 13], others [5, 6, 10, 20] argue for transnasal wiring. Despite the fact that it is a treatment with more morbidity and is difficult to implement, it provides a more effective traction of the ligaments. Thus, we believe that for a better reduction of the intercanthal distance and a proper positioning of the inner canthus in a one-stage procedure with no recurrences, transnasal wiring is a safe procedure for treatment of telecanthus. We are aware that it is a technique with higher morbidity and potential risks of infection, but in our cases it did not occur. Complications were limited to two cases of nasal bleeding that evolved well after buffering for 48 h. There was no need for revision of the procedure or withdrawal of the wire.

Ptosis surgery is also a controversial area related to BPES surgery. Some authors base their treatment on the dependence of levator muscle function, i.e., when it is higher than 4 mm, they prefer shortening the muscle; when it is below 4 mm, they use silicone or fascia lata brow suspension [8–10]. Nakajima et al. [4] invariably recommend a levator muscle reduction approach independent of the muscle function or ptosis severity. They prefer the levator approach because they dislike the static appearance and residual lagophthalmus induced by the lid/brow suspension techniques. Tronina et al. [13] suggest a tarsal plate reduction associated with reinsertion of the elevator aponeurosis.

There seems to be general agreement that the lid/brow sling is the procedure of choice [7–9, 12] with severe ptosis and very poor levator muscle function as usually occurs in BPES patients. In our experience with the patients studied in this report, the levator muscle function was always very restricted, below 4 mm, which is the main reason for using a fascia lata brow suspension procedure in all of our patients. We prefer the procedure described by Lexer [16] with a slight change from the "U" design to a "W" design, which we believe transmits a more effective lid lift with contraction of the frontal muscle. The frontalis sling can be

approached with alloplastic materials [8] or with a fascia lata autograft [9]. We prefer the latter due to its excellent tissue integration and relative lack of postoperative complications.

We agree with the allegations of those who recommend multiple-stage surgery. These authors [5, 7, 8] state that when ptosis and medial canthoplasty correction are performed at the same time, the tightened vertical and horizontal tissues pull against each other in opposite directions inducing loosening and therefore reducing the effect of the medial canthoplasty and/or the ptosis surgery. We believe that this concept is partially true when the surgical procedure on the medial canthal tendon involves shortening, suturing, or tucking of the ligament [4, 7, 11, 13]. On the other hand, when the tendon is tightened with a steel wire by transnasal fixation [5, 6, 10, 20], the added strength practically ensures that the medial ligament will not loosen as the superior lid is opened. Because both medial ligaments are simultaneously tightened one against the other, as with the procedure discussed in this report, both medial ligaments and canthii tend to be more symmetrical and the goal for medial tendon reduction seems to be, in our opinion, easier to achieve; even though this technique is difficult to perform and may be associated with more complications than a simple shortening of the medial ligament. The fascia lata for the lid suspension also yields very strong traction that is resistant to pulling and loosening.

Huang et al. [12] have a different approach. Although they prefer a one-stage procedure, they believe that the medial canthal tendons should be freed and repositioned to the periosteum and tightened only after the ptosis surgery, which they purposely overcorrect.

Taylor et al. [9] report a 26% improvement in the intercanthal distance using one-stage treatment that includes the shortening and fixation of canthal ligament to the periosteum.

Wu et al. [10] prefer to treat ptosis early to avoid amblyopia. They propose different treatment according to the severity of ptosis. In patients with eyelid opening below 2 mm, they use a multiple-step treatment. In blepharoptosis eyelid opening up to 2 mm, they use a single-stage treatment, and get good results when the eyelid opening is larger than 4 mm.

We believe that the effectiveness of a one-stage procedure is more possible with the use of the transnasal wiring technique described and the simultaneous use of a resistant fascia lata lid/brow sling. Even so, the potential risks of the nasal-wiring procedure (possible infection and bleeding) should be taken into account when planning a surgical procedure in a BPES patient.

Transnasal wiring and fascia lata brow suspension are more difficult to implement and have greater morbidity than some simpler techniques, thus the use of these procedures by a broader surgical practitioner base with less experience may not be the most appropriate.

It is our opinion that the ICD/HFD ratio is the best parameter for measuring success of BPES surgery because it depends on the relative proportions of the nasal intercanthal distance and the horizontal fissure length, which are independent of the patient's age or physical characteristics. Most of the literature agrees that it is very difficult to attain an ICD/HFD ratio of 1 which would be the ideal result. Ratios nearing 1.3 are considered both functionally and aesthetically satisfactory. The ICD/HFD ratio results found in this case series study are comparable to those found by others who used a two-stage procedure [5–10], therefore confirming the effectiveness of the one-stage procedure practiced in this report. We also believe that a one-stage procedure helps to reduce health costs and patient/parental discomfort.

The average age of the patients in our study reflects the social conditions and hardships that these patients had in finding a medical center capable of offering treatment for BPES. One possible reason for the success with this combination of techniques could be the older age of the patients. Despite the fact that the surgery has worked well in some younger patients, we cannot state that this combination of techniques would have the same results in all younger patients due to our relatively small size. Additional studies are necessary to determine whether this combination of techniques would have the same success rate in a series with a lower mean age.

It is very important to take into account that in some specific situations (anatomical variations, experience of the surgeon) other approaches should be considered. The one-stage treatment was necessary in our patients because of the difficulty they had accessing specialized medical care and their low socioeconomic status. The multiple-stage treatment would have been much more costly and traumatic.

Disclosure The authors have no conflict of interest or financial ties to disclose.

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