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A Case Report Of Ptosis Correction In Bpes Blepharophimosis – Ptosis Epicanthus Inversus Syndrome

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Abstract-

The blepharophimosis-ptosis-epicanthus inversus syndrome is characterized by shortening of the horizontal orbital fissure (blepharophimosis), congenital ptosis and epicanthus inversus. The condition may occur either as an autosomal dominant trait (blepharophimosis-ptosis-epicanthus inversus syndrome types 1 and 2), or sporadically. Blepharophimosis-ptosis-epicanthus inversus syndrome type 1 is associated with female infertility. Mental subnormality may occur, especially in the sporadic cases. Chromosome analysis from a few patients suggests that the genetic defect causing the syndrome is localized to chromosome 3q22.

A 43-year-old male presented with bilateral ptosis, reduced visual acuity, and classical BPES features. Family history suggested autosomal dominant inheritance. The patient underwent a left-eye frontalis sling operation to address severe ptosis. Postoperative outcomes showed significant improvement, with palpebral fissure height increasing from 3 mm to 14 mm by day 7. Bell's phenomenon was preserved, and no complications occurred. Frontalis sling surgery proves

effective for BPES, enhancing functionality and aesthetics while ensuring patient good visual outcome.

Introduction-

Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) is a highly rare genetic developmental disorder primarily characterized by distinct ocular malformations. This syndrome is divided into two subtypes, both of which feature the defining oculofacial abnormalities. Type I is associated with premature ovarian failure (POF) in addition to the ocular malformations, while Type II presents only with the physical features. The global prevalence of BPES is estimated at 1 in 50,000 births. Current research indicates that there are no significant differences in the occurrence of BPES based on factors such as ethnicity, sex, race, or age. The condition remains exceptionally rare, with both subtypes exhibiting similar hallmark traits, though Type I's additional ovarian failure sets it apart from Type II. Despite the rarity of the disorder, it provides valuable insight into genetic developmental conditions with ocular manifestations. [1]

BPES is diagnosed based on the presence of characteristic oculofacial features and, in Type I, primary ovarian insufficiency. Both subtypes of BPES result from mutations in the FOXL2 gene, a forkhead transcription factor located on the long arm of chromosome 3 (3q23). These genetic mutations are central to the development of the condition's hallmark features and associated ovarian dysfunction in Type I. [2]

The FOXL2 protein appears to be involved in the development of eyelid muscles as well as in the growth and development of ovarian cells. One family pedigree has been reported with BPES II previously. [3]

Case Report-

43 year old male present in Ophthalmology OPD RMCH with complaints of drooping of both upper lids and DOV since childhood. No history of systemic illness or ocular trauma or ocular surgery in past.

There is family history of similar eyelids complaint in father, brother and his son and daughter.

On examination-

BCVA RE 6/12, LE- PL +ve PR accurate

On Ocular examination-

Resting gaze of patient- chin up, head backward tilt, arching of eye. Lower lid – vertical folds of skin arose that were inserted in the upper lid suggestive of epicanthus inversus.

Patient also has increased distance between medial canthus of both eyes around 35mm which is suggestive of Telecanthus.

Patient has an over action of the frontalis muscle.

The length of vertical palpebral fissure height= 3 mm

The length of horizontal palpebral fissure = 20 mm --- Suggestive of Blepharophimosis

Conjunctiva, Cornea, Lens, Fundus = WNL

Management of BPES should address the eyelid malformation and any related visual impairment.

The ptosis in BPES has generally been corrected surgically and tends to be severe due to poor levator function.

In our patient we planned for the left eye Frontalis Sling Operation for correction of ptosis as patient have good visual prognosis in the left eye.

POD 1- Palpebral fissure height = 13 mm

Mean reflex distance 1 = 6 mm

Mean reflex distance 2 = 7 mm

with good bell's phenomenon

On POD 7- Palpebral fissure height = 14 mm

Mean reflex distance 1 = 7 mm

Mean reflex distance 2 = 7 mm

with good eye closure and good bell's phenomenon.

Cornea, conjunctiva, lens, fundus is WNL

BCVA LE 6/12

Discussion-

Blepharophimosis Ptosis Epicanthus Inversus Syndrome (BPES) is a rare congenital eyelid disorder, occurring in approximately 1 in 50,000 births and inherited in an autosomal dominant manner. Up to 75% of cases exhibit FOXL2 mutations, commonly intragenic, including nonsense, missense, duplications, and deletions. [4,5]

Management requires a collaborative, multidisciplinary approach with clinical geneticist, pediatric ophthalmologist, oculoplastic surgeon, endocrinologist, reproductive endocrinologist and gynecologist. Eyelid surgery in BPES is based on benefits and risks, with early surgery preventing deprivation amblyopia and late surgery allowing for more reliable ptosis measurements, the latter of which provides a better surgical outcome. Ptosis surgery may be affected by dysplastic structure of the eyelids. Surgery traditionally involves a medial canthoplasty for correction of the blepharophimosis, epicanthus inversus, and telecanthus at age three to five years, typically followed a year later by ptosis correction; recently, a one-stage surgical procedure has been described. [6]

Conclusion-

In conclusion, due to the rarity of Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES), the current body of literature remains limited, making treatment approaches varied and not universally standardized. Different surgical techniques are used to correct the condition which led to significant improvements in ptosis, epicanthus inversus, blepharophimosis, and a reduction in the inner canthal distance, effectively addressing both functional impairments and aesthetic concerns. Not only did the procedure improve the patient's appearance, but it also enhanced visual function and helped reduce the psychological burden often associated with BPES. The positive outcomes demonstrate the value of personalized surgical treatments that focus on the unique needs of each patient, resulting in substantial improvements in their overall quality of life. Despite these successes, further research is needed to refine surgical techniques and establish comprehensive treatment guidelines. More extensive studies will be instrumental in optimizing management strategies for BPES and providing clinicians with evidence-based recommendations to improve patient outcomes. Ultimately, ongoing research is essential for better understanding and treating this complex congenital condition.

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