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Blepharophimosis Syndrome

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ABSTRACT

Blepharophimosis syndrome (BPES) is a rare congenital condition primarily affecting the eyelids, leading to distinctive facial features. It is characterized by bilateral ptosis (drooping of the upper eyelids), shortened horizontal palpebral fissures (narrowed eye openings), epicanthus inversus (an upward fold of skin near the inner corner of the eye), and telecanthus (increased distance between the inner corners of the eyes). These features can significantly impact both vision and facial aesthetics. BPES is classified into two types based on its associated systemic manifestations. Type I BPES is linked to premature ovarian insufficiency (POI), which can result in infertility in affected females. In contrast, Type II BPES presents without ovarian involvement, primarily affecting only the eyelids. The condition is caused by mutations in the FOXL2 gene, which plays a crucial role in eyelid and ovarian development. Genetic testing is essential for confirming the diagnosis and guiding management, particularly in distinguishing between the two types. A multidisciplinary approach is vital in the evaluation and management of BPES. Ophthalmologic assessment is necessary for determining the extent of eyelid malformation and planning surgical correction. Genetic counseling is recommended for affected families, particularly for females with Type I BPES due to reproductive implications. Endocrinologic evaluation may also be required to assess ovarian function. Early surgical intervention, such as frontalis suspension or eyelid reconstruction, can significantly improve visual function and cosmetic outcomes. Regular follow-up is crucial to monitor for potential complications and ensure optimal management of both ocular and systemic aspects of the condition.

Keywords: Blepharophimosis syndrome, BPES, palpebral.

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INTRODUCTION

Blepharophimosis syndrome (BPES) is a rare genetic disorder that affects the development of the eyelids and the face.[1] It was first described in 1921 by KOMOTO.[2] BPES is caused by a mutation in the FOXL2 gene which is located on chromosome.[3] It is usually inherited in an autosomal dominant pattern but can also be caused by a new mutation. BPES type I is associated with premature ovarian insufficiency (POI) in females. The syndrome is characterized by a) Telecanthus: increased distance between the inner corners of the eyes; b) ptosis: drooping of upper eyelid c) epicanthus inversion: upward fold of skin near the inner corner of the eye; where the medial eyelid skin fold appears more prominent in the lower eyelid extending across the canthus and tapering into upper eyelid. The structural defects observed in BPES are caused by the interruption of the development of the eyelid and related tissues during embryogenesis.

CASE REPORT

A 9 year old female presented in the OPD of Bharatpur Eye Hospital with complain of photophobia and structurally face down since childhood. She complains of opening of mouth while attempting to open her eyes. Her unaided visual acuity was 6/18 in both eyes and pinhole visual acuity was 6/6 P in right eye and 6/12 in the left eye. Refraction was performed under cycloplegic effect. The refractive error of right eye was found to be +1.50 DSPH/-1.25 DC *150 and +0.50 DSPH/-2.25 DC *150 respectively. All ocular structured are examined using slit lamp. Bilateral ptosis was found with epicanthus inversion and telecanthus. Cornea was clear with pupil being round regular reactive with no relative afferent pupillary defect. Anterior chamber was normal and lens shows clear. Fundus evaluation was done with dilated pupil and it unfolds normal fundus. Complete ptosis evaluation was done and the values are indicated below: Different compensatory head posture was present with raising eyebrow, opening of mouth, chin depression. Distance between two medial canthus and two pupils were 35 mm and 55 mm.

Measurements / Tests	RE	LE
Hirschberg test	Central	Central
Lid crease	Absent	Absent
Palpebral fissure height	5 mm	4 mm
Marginal reflex distance 1	1 mm	2 mm
Marginal reflex distance 2	5 mm	4 mm
Bells phenomenon	Good	Good
Marcus Gunn jaw winking	Negative	Negative
Corneal sensation	Intact	Intact

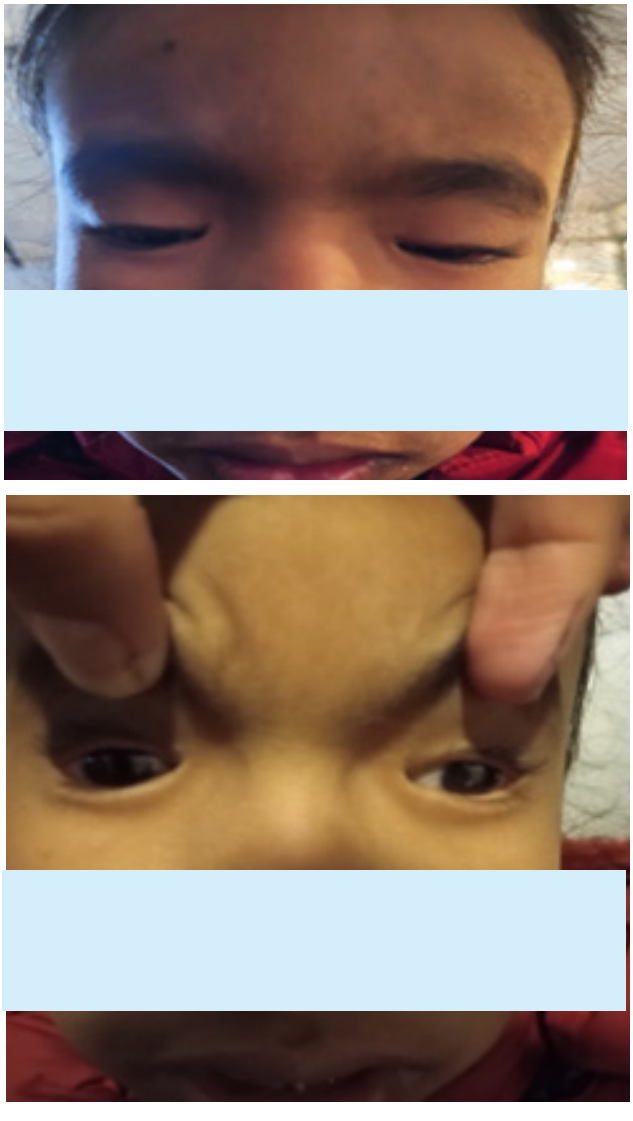

Figure 1. Clinical picture of patient showing bilateral ptosis, absent lid crease, epicanthus inversus

IMPRESSION

A diagnosis of Blepharophimosis syndrome was established based on finding of telecanthus, epicanthus inversus and ptosis.

MANAGEMENT

Ptosis surgery was advised with the aim to promote normal visual development, improve cosmesis, alleviate neck strain from compensatory head posture. Glass was prescribed for the associated refractive error.

DISCUSSION

Blepharophimosis was first described in medical literature in 1841. Later it was associated with ptosis and epicanthus inversus in 1889. Zlotogora et al. first described two phenotypes of BPES, with the differentiating factor being the concomitant presence of premature ovarian insufficiency.[4] A family with 21 affected subjects in five generations was reported in 1921 by Dimitry 5 as having ptosis alone and did not specify any other features, although photographs in the report show that they probably had the full syndrome. Dimitry's pedigree was updated by Owens et al. in 1960.[6] In Pakistan, a case report of BPES in a Pakistani family with three other members having the same syndrome across multiple generations has been described1 primarily diagnosed during ophthalmologic evaluation. From India, a report has described a family with two female siblings, who have typical eye manifestations of BPES along with amenorrhea, while their father only has ophthalmic manifestations.[7] While others have described families affected with POF and eye features of BPES.[8] A retrospective study in 204 individuals with BPES showed strabismus in 20%, a significant refractive error in 34%, and bilateral or unilateral amblyopia in 21% and 20%, respectively.[9]

CONCLUSION

Blepharophimosis syndrome has 4 traditional clinical signs; telecanthus, epicanthus inversus, ptosis and blepharophimosis. Type I is linked to early ovarian failure and only the traditional facial traits define type II. If left untreated these characteristics are linked to a high amblyopia risk. Early surgical intervention is necessary for both types to ensure normal development of eyesight.

Conflict of interest: None

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