http://jmscr.igmpublication.org/home/
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v8i5.31

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Blepharophimosis Syndrome- A Rare Case

Authors
Dr Preeti Rawat[1], Dr Rekha Soni[2], Dr Shruti Choudhary[3]
[1]Professor Department of ophthalmology MGM Medical College Indore
[2]PG resident MGM medical College and MY hospital Indore

Abstract
Background: *BPES is complex of eyelid malformation.*
It is very rare and dominantly inherited condition approximately 6% of children with congenital ptosis present with this syndrome associated with primary amenorrhea in some family lines.
Case Description: *17 year male present with chief complaint of diminution of vision with small eyes since birth associated with watering, on ptosis workup he has absent lid crease, good levator function with good bells phenomena*
Prognosis: *Excellent eyelid surgery results have been published typically requiring multiple staged surgeries.*

Introduction

It is an uncommon inherited dysmorphic syndrome which primarily affect soft tissue of mid face with signs including blepharomiosis, telecanthus, ectropion, stabismus, high arched brows
Type 1-associated with primary ovarian failure
Type 2- no systemic association.

Case Report

-17 yr male
-small eyes
-drooping of eyelids since birth

Past History

History of oculoplasty surgery done in 2008 (V-Y plasty with MCL tightening)

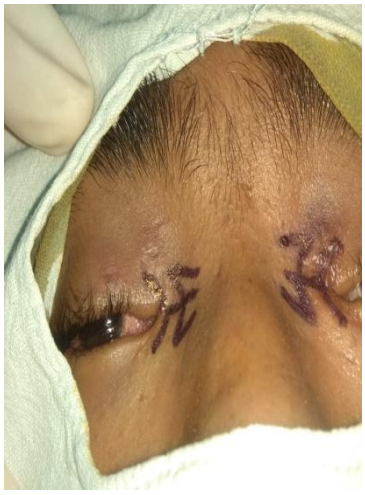
Clinical Features

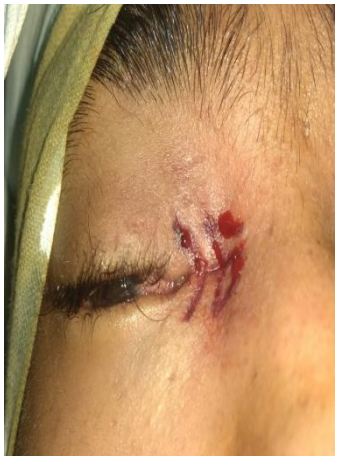
Drooping of eyelids with head tilted slightly backwards with chin up
With Interpupillary distance-60 mm
Intercanthal distance-40 mm
Horizontal palpebral aperture-27 mm

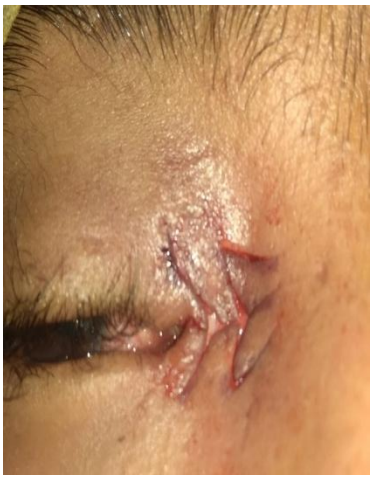
Surgical Management

Treatment needs to address both the eyelid malformation with purpose of correcting blepharophimosis, epicanthus inversus, telecanthus and ptosis
Mustarde' technique (double Z plasty) done under local anaesthesia.

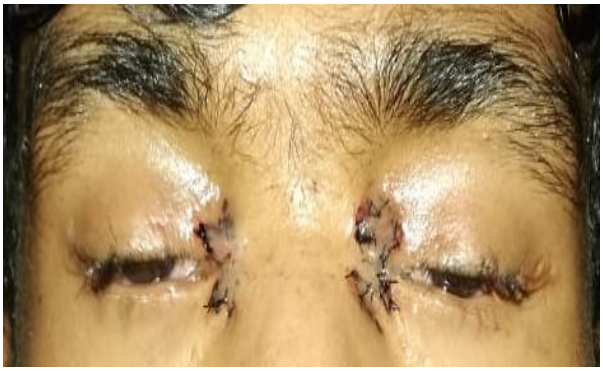
Intra operative Pictures

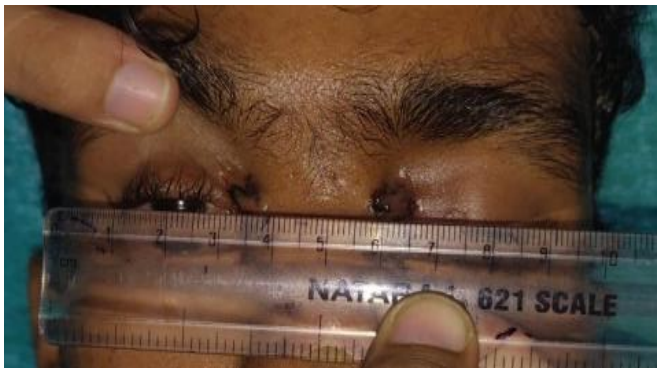


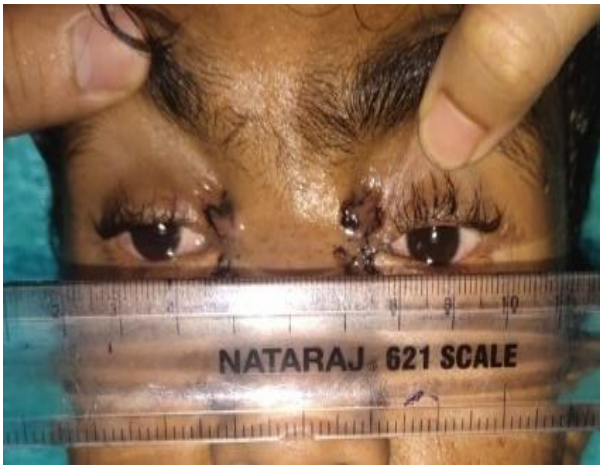




Postoperative Pictures







Intercanthal distance - 35 mm
Other options which includes Y to Vflap, five flap
New surgical approach involves modified skin redraping method, telecanthus corrected by shortening medial canthal tendon fixing it to subcutaneous tissue.

Post Operative
Interpupillary distance-55 mm
Intercanthal distance-35 mm
Horizontal palpebral aperture-27 mm

Discussion
It is very rare only 50-100 cases have been described n dominantly inherited n severity of finding varies among affected family members. Retrospective analysis of BPES patients also identified amblyopia (41%), Refractive errors (34%), strabismus (20%), nystagmus (6%),. Of the patient with strabismus 70% showed esotropia 25% exotropia and 5% hypertropia.

Conclusion
The prevalence BPES is unknown but there are no difference in prevalence based on ethnicity, sex, race and age.

References

1. Vignes A. Epicanthus hereditaire. Rev Gen Ophthalmol (Paris) 1889;8:438–439
2. Beaconsfield M, Walker JW, Collin JR. Visual development in the blepharophimosis syndrome. Br J Ophthalmol 1991;75: 746–748.
3. Verdin H, De Baere E. Blepharophimosis, Ptosis, and Epicanthus Inversus. 2004 Jul 8 [Updated 2015 Feb 5]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews ® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019.
4. Tyers, A. G. (2011). "The blepharophimosis-ptosis-epicanthus inversus syndrome (BPES)." Orbit 30(5): 199-201.
5. Batista F Vaiman D, Dausset J, Fellous M Veitia R. 2007. Potential targets of FOXL2 a transcription factor involved in craniofacial and follicular development identified by transcriptomics. Proc Natl Acad Sci USA 104:3330-3335.