

Sporadic Blepharophimosis Syndrome: A Case Report



Medical Science

KEYWORDS : Blepharophimosis syndrome; sporadic blepharophimosis.

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ABSTRACT

Blepharophimosis syndrome is a rare congenital eyelid malformation characterized by shortened horizontal palpebral fissures (blepharophimosis), impaired function of levator palpebrae superioris of upper lid (ptosis), a vertical skin fold arising from the lower eyelid that inserts medially in the upper lid (epicanthus inversus) and an increased inner canthal distance (telecanthus). Here we are reporting a case of blepharophimosis syndrome in a 8 year old female patient. She presented with drooping of eyelids which was constant throughout the day. On examination found to have chin lift, bilateral blepharoptosis with a broad and flat nasal bridge, epicanthus inversus, telecanthus, and blepharophimosis. Based on the constellation of these findings, a diagnosis of BPES was reached for the girl.

Patient then was planned for a two stage procedure. In the first sitting we undertook lateral canthotomy and medial canthoplasty (Modified Mustarde technique).Ptosis was corrected with bilateral Frontalis Sling surgery in the second sitting. Patient was followed up weekly & showed clinical improvement with minimal scarring.

Introduction:

Blepharophimosis was first reported by Von Ammon in 1841¹, Vignes first associated blepharophimosis with blepharoptosis and epicanthus inversus in 1889. BPES is a rare congenital eyelid malformation. It is characterized by shortened horizontal palpebral fissures (blepharophimosis), impaired function of levator palpebrae superioris of upper lid (ptosis), a vertical skin fold arising from the lower eyelid that inserts medially in the upper lid (epicanthus inversus) and an increased inner canthal distance (telecanthus)². BPES largely a hereditary autosomal dominant entity having mutation in transcription factor FOXL2 (chromosome 3q)^{3,4,5,6,7}. BPES type-1 associated with female infertility which is absent in type-2⁸. In this case report we demonstrate a sporadic case of Blepharophimosis syndrome in 8 year old female.

Case Report:

An 8 years old female patient was brought to Ophthalmology clinic with complains of drooping of eyelids since birth. Patient wanted correction for the same as it was cosmetically hampering. Amount of drooping of eyelids was constant throughout the day. Her antenatal, natal and post-natal history were otherwise unremarkable. Had no history of similar occurrence in family. On examination, chin lift and normal facial symmetry was noted. There was bilateral blepharoptosis with a broad and flat nasal bridge. There was a vertical skin fold arising from the lower eyelid that inserted medially in the upper lid (epicanthus inversus). The distance between the medial canthi was 40 mm (telecanthus). Bilaterally, the length of the vertical palpebral fissures was 4 mm whereas that of the horizontal palpebral fissures was 20 mm (blepharophimosis) which were shorter than the normal measurements for this age.

Rest of the ocular and detailed systemic examinations were within normal limits. Based on the constellation of these findings, a diagnosis of BPES was reached for the girl.

Patient then was planned for a two stage procedure. In the first sitting we undertook lateral canthotomy and medial canthoplasty. Lateral canthotomy was addressed to lengthen the horizontal palpebral aperture. Modified Mustarde technique was employed for medial canthoplasty which dealt with Epicanthus Inversus. Ptosis was corrected with bilateral Frontalis Sling surgery in the second sitting. Patient was followed up weekly & showed clinical improvement with minimal scarring.

Table 1

	Pre-Operative Measurements		Post-Operative Measurements	
	RE	LE	RE	LE
MRD1	-2mm	-2mm	3mm	2mm
MRD2	6mm	6mm	8mm	8mm
Vertical Palpebral Aperture	4mm	4mm	11mm	10mm
LPS Function	1mm	1mm	-	-

MRD – Margin reflex distance, LPS – Levator palpebral Superioris

Figure 1: Pre operative Image



Figure 2: Post Operative image

Discussion

The management of blepharophimosis is primarily surgical and over the years, the surgery for

BPES have evolved from a multi-staged approach to one that involves fewer stages⁹. However, absolute consensus on the most appropriate and effective approach is still lacking. Blepharophimosis can either present in the constellation of BPES or can be associated with other developmental ocular as well as systemic abnormalities. Therefore, a thorough and complete physical examination, in addition to a thorough family history of the patient, is necessary. BPES must be considered an important differential diagnosis in any patient presenting with ptosis and blepharophimosis. If available, molecular characterization and genetic evaluation may be useful for a more definitive diagnosis and genetic counselling. Family pedigree construction is also important to identify the pattern of inheritance of the condition. The late presentation of this patient could have led to stimulus deprivation amblyopia. There have been previous reports of amblyopia associated with blepharophimosis syndrome. A study by Jethani et al in India reported that 31.5% of their patients with blepharophimosis syndrome had amblyopia¹⁰. In case series report by Beckingsale et al, 39% of the 28 patients with blepharophimosis syndrome had amblyopia¹¹.

The severity of ptosis in blepharophimosis syndrome may make children to adopt a chin up, backwards head-tilt position and to recruit the frontalis in elevating the lids leading to raised arched eye brows. We suspect that the case highlighted is likely to be sporadic in view of the fact that we could not establish family history. It is interesting to note that our patient was said to be performing well in primary school. This is noteworthy considering the fact that blepharophimosis syndrome most especially the sporadic type is associated with mental retardation. Cai et al in China reported a novel case of unilateral blepharophimosis syndrome and mental retardation associated with de novo trisomy for chromosome 3q¹².

We conclude that, cosmetic factors associated with this condition mandates a wellplanned and appropriate surgical management.

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