Blepharophimosis with Bilateral Duane’s Syndrome - An Unusual Combination

Dr. Vijaya Kumari MS

Introduction

Blepharophimosis is a rare congenital disorder, which occurs sporadically or inherited as an autosomal dominant trait. Duane’s syndrome is characterized by limitation of abduction, narrowing of palpebral fissure along with retraction of eyeball and face-turn. A novel coexistence of sporadic blepharophimosis with bilateral Duane’s Type 1 in a male patient is described here.

Case report

A fourteen-year-old male came with complaints of drooping of upper lid since birth. Patient also complains of inability to close the eyes fully. There is no corneal exposure on attempted closure of the eyes. There is no significant head tilt or face turn. Visual acuity is 6/6 bilaterally. His abduction is partly restricted in both eyes. Retraction of the eyes on attempted adduction is present in both the eyes. There is no strabismus in primary position. Apart from this, there is hypoplasia of superior orbital rim, flat nasal bridge and distichiasis. There is moderate degree of ptosis in both the eyes with the upper lid covering half the pupil. Levator function is only 4 mm bilaterally. Lid crease is also noted to be absent. There is also ectropion of the lower lid, telecanthus and epicanthus inversus, which completes the picture of blepharophimosis syndrome. Anterior segment and fundoscopy is normal.

Shows ptosis in primary position (1), eye retraction and abduction restriction on side gaze (2, 3)
Discussion

Blepharophimosis ptosis epicanthus inversus syndrome (BPES) is a distinctive congenital eyelid malformation, which can occur sporadically or inherited as an autosomal dominant fashion. Both types of BPES have been mapped to chromosome 3q23 and mostly due to mutation of a fork head transcription factor FOX-L2 gene, which locates at this region.\(^1\)

Primary amenorrhea is described in most of the affected women.\(^2\) Thus, BPES is characterized by facial dysmophy combined in some cases with primary ovarian failure.\(^3\) High degree of menstrual irregularities and infertility are common in affected women. Early recognition of this condition may allow appropriate counseling and treatment including egg donation. During the period of four to eight weeks of embryonic development the cranial nerves, their nuclei and corresponding innervation to extra ocular muscles undergo development and differentiation. This coincides with period of time that FOX-L2 is strongly expressed in developing eyelid and surrounding tissues.\(^4\)

Mental subnormality can occur at times in sporadic cases. Seventy percentage of BPES patients have refractive error. In a study by Kyung. S et al\(^5\) in Samsung medical centre, Seoul, Korea, out of 20 BPES patients, 45% had amblyopia, 25% unilateral and 20% bilateral. 67% of those with Amblyopia has coexisting strabismus.\(^5\)

In our patient, the only complaint was ptosis. There was no squint in primary position, refractive error or head tilt. Since the levator action is poor, the patient is advised sling surgery at a later date. Understanding this rare and complex syndrome will aid us in better management and counseling of such cases when need arises.

References

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3. Loffler.K.A, Zarkower.D. Etiology of ovarian failure in BPES, FOX-L2 is a concerned early acting gene in vertebrate ovarian development; Endocrinology. 2003 Jul 144(7); 3237-43