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Blepharophimosis Syndrome In The Case Of Two Brothers

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Abstract
Blepharophimosis syndrome (BPES) is a congenital eyelid malformation characterized by severe bilateral ptosis, telecanthus, and epicanthus inversus. We present a case report of two siblings, a 9-year-old boy and a 5-year-old girl, diagnosed with BPES. Clinical examination revealed classic features of the syndrome including bilateral ptosis, telecanthus, and epicanthus inversus. Both children exhibited compensatory postures to cope with visual impairment. BPES follows an autosomal dominant pattern, with mutations in a gene on chromosome 3. Management involves early ophthalmological assessment, surgical intervention, and multidisciplinary collaboration. Amblyopia and other ocular disorders may complicate the clinical course. Awareness of the psychological impact on patients and families is crucial. Early intervention is imperative for optimal visual outcomes and to mitigate the associated challenges of this complex syndrome

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I. Introduction

Blepharophimosis syndrome is a congenital palpebral malformation that combines bilateral ptosis with telecanthus and epicanthus inversus. It may also include other palpebral malformations(1). The syndrome can be divided into two main categories: type 1 is associated with ovarian anomalies, while type 2 is limited to palpebral involvement(2). Our case report illustrates a case of blepharophimosis syndrome identified in two brothers.

Observation of the patients

The first child, aged 9, was referred by a pediatrician for an ophthalmological examination in the presence of bilateral ptosis. Clinical examination revealed major bilateral ptosis associated with telecanthus (Figure 1), epicanthus inversus (Figure 1), and absence of the upper palpebral crease. The function of both upper eyelid levators is nil, and the child raises his eyebrows and adopts a vicious head posture (head thrown backwards) during fixation efforts. This clinical picture led to the diagnosis of blepharophimosis syndrome. The child also presented with esotropia and limited abduction in both eyes.

The sister is 5 years old. Clinical examination reveals bilateral ptosis associated with telecanthus (Figure 2), epicanthus inversus (Figure 2), and absence of the upper palpebral crease. The function of both upper eyelid levators is nil, and the child raises his eyebrows and adopts a vicious head posture (head thrown backwards) during fixation efforts. This clinical picture led to the diagnosis of blepharophimosis syndrome. The child also showed esotropia with limited abduction in both eyes. The rest of the examination was unremarkable.

Figure 1: 9-Year-Old Child With Blepharophimosis Syndrome

Figure 2: Little Sister With Blepharophimosis Syndrome

II. Discussion

Blepharophimosis ptosis epicanthus inversus syndrome (BPES) is an inherited condition that follows an autosomal dominant pattern. It is caused by a mutation in a gene on chromosome 3, which affects the development of the eyelid and also has a role in ovarian follicles (3)(4).

Blepharophimosis syndrome is a congenital eyelid malformation that is characterized by specific anatomical changes. The most common clinical signs include severe, bilateral, and symmetrical ptosis, as well as telecanthus, which is an increased distance between the inner corners of the eyes while the distance between the pupils remains the same. Another characteristic of this syndrome is epicanthus inversus, which is a skin fold of the lower eyelid that extends over the inner corner of the eye. Additionally, there is blepharophimosis, which is a significant narrowing of the eyelid opening. Understanding these anatomical features is important in grasping the nature of this congenital eyelid malformation (5)(6).

Patients with this syndrome adopt specific postural positions to compensate for the visual impairments. These include hyperextension of the neck and contraction of the forehead muscles, which help elevate the eyebrows (7). These compensatory mechanisms highlight the functional complexity and adaptive limitations associated with the eyelid anomaly seen in this syndrome.

Other ocular disorders may also be present in individuals with this syndrome, such as microphthalmia, strabismus, and colobomas of the optic disc. Amblyopia, commonly known as lazy eye, is frequently observed in these individuals and can be caused by various factors including severe ptosis, strabismus, and refractive errors. There are also extra-ocular manifestations, such as a broad and flat nasal bridge, arched palate, and cupped ears (6).

Initial management of BPES involves evaluation by a pediatric ophthalmologist to assess for amblyopia, presence of strabismus, and refractive errors. Surgical treatment aims to promote adequate visual development, improve aesthetics, and relieve cervical tension caused by compensatory postures. The surgeon must assess the severity of ptosis, blepharophimosis, and epicanthus inversus before proceeding with the appropriate surgical technique. In some cases, multiple surgical procedures may be required (8)(9).

Early medical and surgical interventions are crucial for a favorable prognosis.

Managing BPES requires a multidisciplinary approach involving collaboration between a pediatric ophthalmologist, an oculoplastic surgeon, a pediatrician, an endocrinologist, a gynecologist, specialized nurses, and a geneticist(10).

III. Conclusion

In conclusion, blepharophimosis syndrome presents significant challenges in its management due to its complex nature. The visual implications, particularly the risk of amblyopia, add to these concerns. Early intervention is essential to ensure optimal visual outcomes. It is also important to recognize the profound psychological impact that this condition can have on both the child and their parents.

References:

[1] Allen Ce, Rubin Pa. Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome (Bpes): Clinical Manifestation And Treatment. Intophthalmolclin. 2008;48(2):15–23

[2] De Baere E, Dixon Mj, Small Kw, Et Al. Spectrum Of Foxl2 Gene Mutations In Blepharophimosis-Ptosis-Epicanthus Inversus (Bpes) Families Demonstrates A Genotype-Phenotype Correlation. Hum Mol Genet. 2001;10(15):1591–1600

[3] Fokstuen S, Antonarakis Se, Blouin Jl. Foxl2-Mutations In Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome (Bpes); Challenges For Genetic Counseling In Female Patients. Am J Med Genet A. 2003 Mar 01;117a(2):143-6

[4] Chawla B, Bhadange Y, Dada R, Et Al. Clinical, Radiologic, And Genetic Features In Blepharophimosis, Ptosis, And Epicanthus Inversus Syndrome In The Indian Population. Invest Ophthalmol Vis Sci. 2013;54(4):2985–2991.

[5] Klein M. Hereditary Bilateral Ptosis And Blepharophimosis Associated With Other Developmental Abnormalities Of The Outer Eye. Proc R Soc Med. 1950 Dec;43(12):1025-6
[6] Beaconsfield M, Walker Jw, Collin Jr. Visual Development In The Blepharophimosis Syndrome. Br J Ophthalmol. 1991 Dec;75(12):746-8
[7] Choi Kh, Kyung S, Oh Sy. The Factors Influencing Visual Development In Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome. J Pediatrophthalmol Strabismus. 2006;43(5):285–288
[8] Karacaoğlan N, Sahin U, Ercan U, Bozdogan N. One-Stage Repair Of Blepharophimosis: A New Method. Plast Reconstr Surg. 1994 Jun;93(7):1406-9
[9] Savino G, Mandarà E, Calandriello L, Dickmann A, Petroni S. A Modified One-Stage Early Correction Of Blepharophimosis Syndrome Using Tutopatch Slings. Orbit
[10] Bhattacharjee K, Bhattacharjee H, Kuri G, Shah Zt, Deori N. Single Stage Surgery For Blepharophimosis Syndrome. Indian J Ophthalmol. 2012 May-Jun;60(3):195-201