Blepharophimosis Ptosis Epicanthus Syndrome: A Case Report

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Introduction: Blepharophimosis ptosis epicanthus syndrome (BPES) is a rare disorder associated with maldevelopment of oculofacial structures.[1] It is characterized by shortened horizontal palpebral fissure, ptosis, epicanthus inversus and telecanthus[2,3,4]. It is usually bilateral and may be asymmetrical[5] . Other features include ectropion, low nasal bridge, microphthalmos, divergent strabismus and amblyopia. Also, cardiac defects, mild mental retardation and psychological challenge from facial appearance may occur.[2,4] In females, premature ovarian failure may be present.[1,2,6]

BPES is usually inherited in autosomal dominant fashion, but may be sporadic.[5,7,8] It is due to mutation of the FOXL2 gene, located in 3q23, causing maldevelopment of eyelids and ovaries.[5,9] Different phenotypes, BPES types 1 and 2 occur.[1,10] Staged surgery is advocated, usually at age 3-5 years.[3]

Case Report: Master B.S, a 3 month old male infant, presented with bilateral non-progressive drooping upper lids from birth. There was no tearing, redness or discharge. Ante- and peri-natal histories were normal. Developmental milestones were age appropriate, with no known family history of similar eye features. Review of systems was normal. On examination, he followed light and objects well; head tilt and chin up position were present. He had bilateral ptosis, phimosed lids, epicanthus inversus and telecanthus (Figure 1). Anterior segments and fundi were normal. Refraction was +1.50DS -0.25DCx180 (right) and +1.75DS -0.25DCx180 (left).

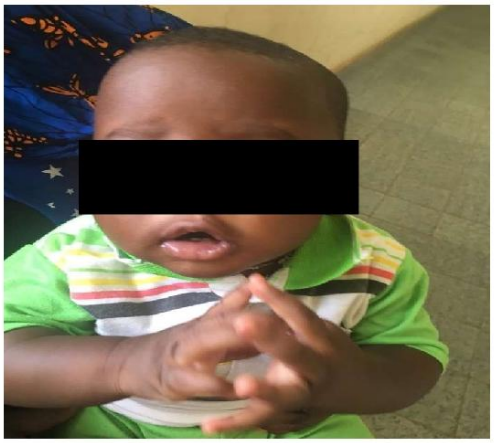

Figure 1: Bilateral ptosis, phimosed lids, telecanthus and epicanthus inversus

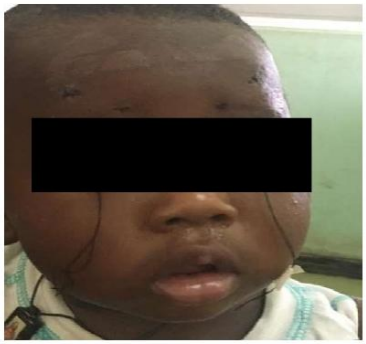

Figure 2: Immediate Post-operatively. Upper

A diagnosis of Blepharophimosis ptosis epicanthus syndrome with imminent amblyopia was made. He subsequently had bilateral tarso-frontalis suspension with silicone sling. Postoperatively, upper lid lift was satisfactory (Figure 2). Repair of epicanthus and telecanthus were scheduled for 4 years of age. Follow-up at the 4th post – operative month showed good lift with no head tilt (Figure 3).

Figure 3: Four month Post-operatively. Lifted lids. Residual telecanthus and epicanthus inversus.

Discussion: Clinical features and onset in index patient bear similarity to documented studies.[4,9] No evidence of mental retardation was obvious in this patient. Refractive status was age-appropriate. Head tilt with marked ptosis suggested visual axis obstruction, a risk for amblyopia. This necessitated early ptosis correction to be followed later by correction of the phimosed lids.[3] Published reports note an adequate balance between benefits of early surgery, avoiding amblyopia, and later surgery (just prior to school age), thus allowing for facial maturation before any intervention.[9]

Traditionally, staged surgery is done for BPES.[4] First, a medial canthoplasty to correct blepharophimosis, epicanthus and telecanthus; then tarso frontalis sling 6-12 months later to correct ptosis.[11] However, severe ptosis and imminent amblyopia may warrant ptosis surgery before 3 years, as in this index patient.[4,13]

One stage surgery has been advocated.[1,11-12] It consists of lateral canthotomy, medial canthoplasty, transnasal wiring and frontalis sling. Genetic studies are recommended, to identify responsible gene and in addition, to identify premature ovarian failure in susceptible females. This was not available for this patient.

Conclusion: Knowledge of the defining features of BPES, adequate assessment and surgical procedures are essential for patient evaluation and rehabilitation.

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