April 2010

Blepharophimosis-ptosis-epicanthus inversus syndrome in a Pakistani pedigree

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INTRODUCTION

Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES), a rare developmental eye condition, 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). In a Chinese study, the vertical fissure length was reported from 2 to 4 mm, the horizontal fissure length from 13 to 22 mm, and the inner-canthal distance was reported between 35-39 mm. BPES has largely been reported as a hereditary autosomal dominant entity. Genetic studies done in Indian and Chinese populations have implicated mutations in the Forkhead transcription factor FOXL2 (chromosome 3q) as responsible for BPES.

BPES can be categorized into two types, based on the variable expressivity of female infertility, which is present in type-1 but absent in type-2 BPES.

Here we report a case of BPES in a Pakistani family with three other members having the same syndrome across multiple generations.

CASE REPORT

A 2-year-old girl child was brought to the ophthalmology clinic by her father who sought consultation for the correction of her shortened palpebral fissures. Detailed history of the girl showed that the condition had been present since birth. Her antenatal, natal and postnatal histories were otherwise unremarkable. On physical examination, the girl had 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) and an increased inner canthal distance (telecanthus).

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Key words: Blepharophimosis. Blepharoptosis. Epicanthus inversus. Pakistan. Pedigree.
blepharoptosis for the patient was discussed with her father; however, it has been deferred until the patient is older.

DISCUSSION

Von Ammon first used the term “blephaerophimosis” in 1841, and Vignes first associated blepharophimosis with blepharoptosis and epicanthus inversus in 1889. Since then, BPES cases have been sporadically reported in literature. In this report, 4 members of the same family were affected across 3 generations. In a Chinese study, 12 affected patients were identified across 4 generations in a single family. Moreover, the ocular measurements—the vertical and horizontal fissures lengths—of this case closely matched those reported in another Chinese study.

BPES may also be rare in Pakistan, but it remains to be seen how rare this condition is because of lack of BPES-specific published literature in the country.

In a 10-year, case series of 101 patients of BPES from the United Kingdom, 31 patients had a positive family history of the condition. This underscores the importance of subjecting patients to a thorough family history. In the UK study, there was a 5:1 predominance on the father’s side. In the same study, it was noted that 27 cases presented for oculoplastic consult by 18 months of age and another 25 cases by the age of 5 years. This patient presented for the consultation at 2 years of age.

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 stages. However, absolute consensus on the most appropriate and effective approach is still lacking. Surgery has been deferred in this patient for the time being; however, the patient’s father has been encouraged to maintain regular follow-ups.

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 counseling. Family pedigree construction is also important to identify the pattern of inheritance of the condition.

REFERENCES