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RESEARCH ARTICLE

BLEPHROPHIMOSIS PTOSIS AND EPICANTHUS INVERSUS SYNDROME

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ABSTRACT

Introduction: Blepharophimosis, ptosis and epicanthus inversus syndrome (BPES) is a developmental disorder for which diagnosis is based on 4 major features: Blepharophimosis, Ptosis, Epicanthus inversus and telecanthus). This condition is caused by mutations in the FOXL2 gene and is inherited in an autosomal dominant pattern.

Case report: A 12 year old girl presented ptosis with strabismus in eye OPD. The patient had bilateral ptosis. On basis of ophthalmic examination diagnosis of BEPS made. There is a high incidence of bilateral strabismus than the general population which can be detected in our patient.Occasional ocular findings reported in some patients include microphthalmos, anophthalmos, microcornea, hypermetropia, and nystagmus which are not detected in our patient.

Conclusion: Management of blepharophimosis syndrome type 1 requires the input of several specialists including a clinical geneticist, pediatric ophthalmologist, eye plastic surgeon, endocrinologist, reproductive endocrinologist and gynecologist.

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INTRODUCTION

Blepharophimosis, ptosis and epicanthus inversus syndrome (BPES) is a developmental disorder for which diagnosis is based on 4 major features: Blepharophimosis (shortened horizontal palpebral fissure); Ptosis (drooping of eyelids), Epicanthus inversus (vertical fold of skin that stretches from the lower eyelid up toward either side of the nose) and telecanthus (lateral displacement of inner canthi with normal interpupillary distance). All these features are typically detected in our patient.

Case report

A 12 year old girl presented ptosis with strabismus in eye OPD.Visual acuity was 6/60 in right (not improving with pinhole) and 6/6 left eyes. Esotropia of 45 degree was present in right eye. There was no improvement in visual acuity with pin hole. The patient had bilateral ptosis. Evaluation of the ptosis revealed palpebral fissure height of 6mm on the right and 7mm on the left. The lid excursion was 5 mm in both eyes. The margin reflex distance (MRD) was zero.

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The patient also had bilateral epicanthus inversus and telecanthus (Fig 1). Fundus examination and indirect ophthalmoscopic examination has not revealed any abnormality. Rest of eye examination was with in normal limit.Systemic examination was with in normal limit. On basis of ophthalmic examination diagnosis of BEPS made. There is a high incidence of bilateral strabismus than the general population which can be detected in our patient. Occasional ocular findings reported in some patients include microphthalmos, anophthalmos, microcornea, hypermetropia, and nystagmus which are not detected in our patient. In addition our patient has some facial features as antimongoloid slant, flat nasal bridge, high arched palate as well as low set ears with less corrugations of ear cartilage. These features were also reported previously in BPES cases. Patient's guardian was advised for surgical correction of ptosis. She is on follow up gyanae department as there is higher incidence of amenorrhea in BEPS cases. In view of the rarity of blepharophimosis syndrome in this environment, we decided to highlight the case of bleparophimosis syndrome with associated strabismus and ambylopia in a 12 year old Indian male child. We are not aware of similar reports in this part of the world.

DISCUSSION

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 amblyopia (Jethani et al., 2007). In case series report by Beckingsale et al, 39% of the 28 patients with blepharophimosis syndrome had amblyopia (Beckingsale, 2003). The authors concluded that patients with blepharophimosis syndrome have a high rate of amblyopia (Allen, 2008). In a case series of one hundred and one of blepharophimosis syndrome reviewed by Beaconsfield et al in London, 56.4% of them had amblyopia (Beaconsfield, 1991). It has been advocated that patients with severe ptosis should have their ptosis corrected before three years of age and all other patients should undergo surgery before five years of age (Beckingsale, 2003).

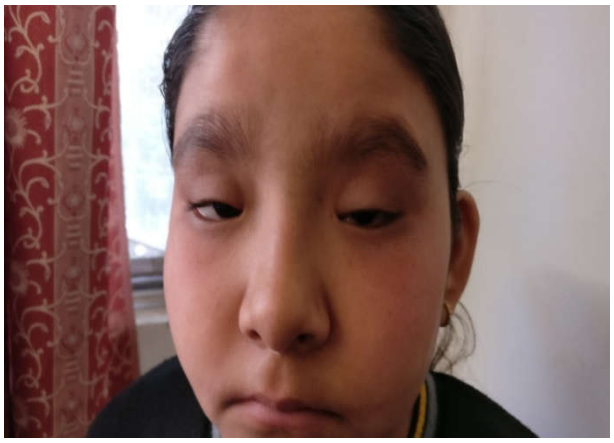

Figure 1

Blepharophimosis syndrome is a congenital eyelid malformation. It was first reported in 1841 by von Ammon. It is inherited in an autosomal dominant fashion (Cai, 1997). In humans, the upper and lower eyelids normally fuse together in the eight-week of development and separate again between fifth and seventh months (Sevel, 1988). Abnormal eyelid development has been observed in both mice and humans, but the molecular events governing both normal and abnormal eyelids development are not fully understood (Oley, 1988; Vassalli et al., 1994). However, some progress in understanding the molecular. Other chromosomal regions have been implicated in the aetiology of blepharophimosis syndrome. Maw et al reported linkage of blepharophimosis syndrome in large Indian pedigree to chromosome 7p13-p21 (Maw et al., 1996). Blepharophimosis syndrome is associated with dominantly inherited mutation in the FOXL2 gene on chromosome 3q23. The gene is expressed in the development of eyelid and ovary (Allen, 2008). Up to 75% of patients with blepharophimosis syndrome have relatives who have FOXL2 mutation, the remaining 25% of cases represent new mutation or milder expression in previous generations. Type 1, blepharophimosis syndrome is characterised by complete penetrance and transmission through males because of impaired female fertility due to premature ovarian failure. In type 2, there is incomplete penetrance and transmission by both males and females (Oley, 1988; Allen, 2008; Zlotogora).
Blepharophimosis syndrome features include epicanthus inversus, low nasal bridge and ptosis of the eyelid resulting in narrowing of the palpebral fissures.Associated features of the eye include nystagmus, microphthalmos, microcornea and stenosis of the lateral canaliculi (Cai et al., 1997). Other features of blepharophimosis syndrome include mental retardation seen mainly in sporadic cases (Cai et al., 1987). Refractive errors, amblyopia and strabismus are commonly associated with blepharophimosis syndrome. It is also often associated with nasolacrimal drainage problems.In view of the rarity of blepharophimosis syndromet, we decided to highlight the case of bleparophimosis syndrome.

Conclusion and Manaement

Management of blepharophimosis syndrome type 1 requires the input of several specialists including a clinical geneticist, pediatric ophthalmologist, eye plastic (oculoplastic) surgeon, endocrinologist, reproductive endocrinologist, and gynecologist. Eyelid surgery should be discussed with an oculoplastic surgeon to decide on the method and timing that is best suited for the patient. Traditionally, surgical correction of the blepharophimosis, epicanthus inversus, and telecanthus (canthoplasty) is performed at ages three to five years, followed about a year later by ptosis correction (usually requiring a brow suspension procedure). If the epicanthal folds are small, a "Y-V canthoplasty" is traditionally used; if the epicanthal folds are severe, a "double Z-plasty" is used. Unpublished reports have indicated that advanced understanding of the lower eyelid position has allowed for more targeted surgery that results in a more natural appearance. Generally, premature ovarian failure (POF) is treated with hormone replacement therapy. There is no specific treatment for POF caused by blepharophimosis syndrome type 1. Hormone replacement therapy is generally estrogen and progesterone and sometimes also includes testosterone. Birth control pills are sometimes substituted for hormone replacement therapy. Although health care providers can suggest treatments for some of the symptoms of POF, currently there is no scientifically established treatment to restore fertility for women diagnosed with POF (Premature Ovarian Failure, 2009).

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