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Blepharophimosis-ptosis-epicanthus inversus syndrome : about two cases

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Abstract
Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) is a rare autosomal dominant disorder characterized by orbito-palpebral anomalies.
Other ophthalmic manifestations can be associated with BPES including ,euryblepharon, strabismus, microphthalmos, lacrimal drainage abnormality, and optic disc coloboma .
We reporttwo case of BPES in both eyes .Referedfor a congenital ptosis reported by their parents . No other ophthalmologic manifestations wasassociated with this syndrome.

Keywords: *congenital ptosis ,pediatric ophthalmology , Blepharophimosis*

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I. INTRODUCTION

Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) is arare autosomal dominant genetic disorder characterized by different orbito-palpebral abnomalities: blepharophimosis, ptosis, epicanthusinversus, and telecanthus[1]. No differences in prevalence has been reported, but this syndrome is not very rare , different cases have been described in the literature [2]

Other ocular signs morefrequently present including squint, nystagmus due to amblyopia, microphthalmus, microcornea and lacrimalstenosis [2].

II. CASE REPORT

- Case 1: This study reports the case of a 2 years oldboy referred to the pediatric ophthalmology department for poor vision and ptosis.

The infant had small palpebral fissure a drooping eyelids since birth , the examination revealed a vicious attitude of theinfant with head tilted backwards, the examination of the eyelids showed an epicanthus inversus.

The distance between the two medial canthus was 45 mm defining the telecanthus(Fig. 1).

The infant had horizontal nystagmus in the left eye. The patient had severeptosis on the left eye with a difficulttoassess levator course and a flat nasalbridge .The boy had also a bilateral ear dysplasia .

Anterior segment examination of both eyes was normal.

A surgery was scheduled when the boy will reach 5 or 6 yearsold .

- Case 2 : the second case shows a 6 years old girl , refered for fision loss in the left eye , ptosis .
The ophthalmological examination has shows an epicanthus inversus in both eyes , ptosis was severe on the left eye compared with the right , a telecanthus
The visual acuity was 8/10 in both eyes ,anterior and posterior segment examination where normal . (fig 2)

III. DISCUSSION

The diagnosis of blepharophimosisptosis epicanthus inversus syndrome (BPES) is easily made when we find the characteristic eyelid abnormalities are found and a family history of BPES [3]
For a long time we define any causes of BPES , actually some studies shown that it could be a genetic implication due to mutation in FOXL2 gene on chromosome 3q23 region[4].
FOXL2 gene is implicated in the developing eyelid and also the ovary[4]
Surgical management of these syndromes is and requires the coordination of different specialties, pediatric ophthalmology, oculoplastic, endocrinologist, gynecologist and geneticists.
The goals of an ophthalmologist caring for these patients to promote normal visual development, improve aesthetics and abnormal chip posture [5].
Surgical management of eyelid malformation is also complex.
Many different surgeries have been described, involving correction of the telecanthus, epicanthus inversus and ptosis

Canthoplasty is performed at 3 to 5 years of age [6]
BPES patients have several other clinical extraocular findings. They are often noted to have broad, flat nasal bridge, arched palate, ear abnormalities like our first case who has ear dysplasia . Mental disorders can be associated with BPES in some cases[7]

CONCLUSION :

This study presents two rare cases of blepharophimosis ptosis epicanthus inversus syndromes followed in the pediatric ophthalmology department . The surgical goal in BPES is not only to improve the unusual facial and abnormal posture of the associated, but also the patient's peripheral visual field. Surgery remains difficult for surgeons ophthalmologists, so the time of surgery must be early because of the risk of amblyopia [3].

CONFLICT OF INTEREST

Authors declare that they do not have any conflict of interest.

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