

Blepharo phimosis syndrome with squint in a 9-year-old boy

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Abstract: Blepharo phimosis syndrome comprises many ocular signs among others blepharo phimosis, ptosis, epicanthus inversus. It may speedily lead to amblyopia. Its management is difficult and may require the expertise of many specialists. The current case was complex, for in addition to the above features, there was a squint.

Keywords: Blepharo phimosis, syndrome, squint

INTRODUCTION

Blepharo phimosis-ptosis-epicanthus inversus syndrome (BPES) is an uncommon congenital disorder characterized by a narrowed horizontal palpebral aperture, ptosis, epicanthus inversus, and telecanthus. It typically has bilateral features, although not always symmetrical [1]. Blepharo phimosis occurring either per se or syndromally is part of over 100 conditions, but the combination with epicanthus inversus, ptosis and cleft lip and palate as phenotypic manifestations of a particular syndrome is very unusual [2]. The global prevalence of BPES has been estimated to be approximately 1 in 50,000 [3]. Main clinical features are: blepharo phimosis, blepharo ptosis, epicanthus inversus and telecanthus. Occasional findings include microphthalmos, anophthalmos, micro cornea, hypermetropia, strabismus, nystagmus, trichiasis, flat broad nasal bridge, high arched palate, minor ear anomalies, cardiac defects, mild mental retardation and infertility in females [4].

CASE REPORT

A-9-year- old boy presented to pediatric ophthalmology out patients department with short palpebral fissures since birth; He was the first and only child of non-consanguineous parents. The prenatal and postnatal history was unremarkable. Family history including the close siblings revealed no abnormal findings. Ocular examination showed a flat broad nasal bridge, blepharo phimosis, narrowed horizontal palpebral aperture epicanthus inversus, bilateral ptosis and left eye esotropia. Cycloplegic refraction results were +1.00 δ for the right eye and +1.50 δ for the left eye; then the best corrected visual acuity was 6/6 in the

right eye and 6/9 in the left eye. The fundus was normal both eyes.



Fig.1: Photograph of the child with blepharo phimosis syndrome

DISCUSSION

Congenital Blepharo phimosis (BPS) is an oculofacial maldevelopmental syndrome comprising the four classic anomalies of epicanthus inversus, telecanthus, palpebral phimosis, and bilateral ptosis.

Moreover, it is also accompanied by poorly developed nasal bridge and hypoplasia of the superior orbital rim. The condition may occur either as an autosomal dominant trait or sporadically [5].

Features in BEPES vary from one patient to another. Our patient had hyperopia; this condition is found by some authors, who noted the presence of refractive errors in some patients [6]. In some cases, there is an elevated intraocular pressure [7]; this condition was not present in our patient.

Surgical management of Congenital Blepharophimosis is not very easy. Some authors perform a single stage surgery [8]. Blepharophimosis syndrome is a complex condition and its management requires input of several specialists including pediatric ophthalmologist, orthoptist and oculoplastic surgeon. Various methods of treatment to improve cosmetic appearance have been advocated [8].

Timing of eyelid surgery is controversial. It involves weighing the balance of early surgery to prevent amblyopia and late surgery to allow for more reliable ptosis measurement [9].

CONCLUSION

Congenital blepharophimosis is a complex syndrome which can lead to amblyopia. Its management is sometimes tough and can call for the expertise of many specialists.

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