Case Report

Blepharophimosis syndrome in a Nigerian male child

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Abstract

This report is a case of blepharophimosis syndrome in a six-year-old Nigerian male child who presented to the Eye clinic of Federal Medical Centre, Owo in June 2010 on account of poor vision of one-year duration. There was no history of similar occurrence in the family of the patient. The patient’s vision was 6/18 and 6/18-1 in the right and left eyes respectively. The patient had cycloplegic refraction which revealed hyperopic astigmatism, but the vision did not improve with post cycloplegic test that was done due to the ambyopia the patient had. In view of this and for cosmetic reasons, the patient was encouraged to have ptosis surgery.

Keywords: Blepharophimosis syndrome, amblyopia, male, sporadic, Nigeria.

Introduction

Blepharophimosis syndrome is a congenital eyelid malformation. It was first reported in 1841 by von Ammon(1). It is inherited in an autosomal dominant fashion(2). In humans, the upper and lower eyelids normally fuse together in the eight-week of development and separate again between fifth and seventh months(3). 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(4,5). However, some progress in understanding the molecular genetic basis of blepharophimosis syndrome has already been made(6). 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(7). 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
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\(^{(4,7,8)}\). 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\(^{(6)}\). 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\(^{(2)}\). Other features of blepharophimosis syndrome include mental retardation seen mainly in sporadic cases\(^{(2)}\). Refractive errors, amblyopia and strabismus are commonly associated with blepharophimosis syndrome\(^{(6)}\).

It is also often associated with nasolacrimal drainage problems. There was an incidence of 18% of nasolacrimal drainage problems in a previous study\(^{(10)}\).

In view of the rarity of blepharophimosis syndrome in this environment, we decided to highlight the case of blepharophimosis syndrome with associated amblyopia in a six-year-old Nigerian male child. We are not aware of similar reports in this part of the world.

**Case history**

A six-year-old Nigerian male presented to the Eye Clinic of Federal Medical Centre, Owo in June 2010. The patient presented on account of not seeing the chalk board very well of one year duration and history of discharge of one week duration. The discharge was associated with redness of both eyes and itching. There is no history of trauma to the eyes and no history of use of recommended glasses. There is no family history of drooping of the lids.

On examination, the visual acuity was 6/18 on the right and 6/18-1 on the left. 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 6mm in both eyes. The margin reflex distance (MRD) was zero. The patient also had bilateral epicanthus inversus and telecanthus (Fig 1).

**Fig1:** Blepharophimosis syndrome (showing telecanthus, bilateral symmetrical ptosis and epicanthus inversus)

There was mild discharge from both eyes and hyperaemia of the conjunctiva of both eyes. The remaining structures in the anterior segment were normal. Funduscopic examination revealed pink optic disc with a cup-disc ratio of 0.3 with normal vessels in both eyes. An assessment of blepharophimosis syndrome was made. Cycloplegic refraction was done two weeks later following resolution of the hyperaemia in both eyes and the patient was discovered to have mild hyperopic astigmatism. The post cycloplegic test done did not result in any appreciable improvement in visual acuity, thus recommended, glasses was not prescribed. The patient was to be referred to an Oculo-plastic surgeon for ptosis surgery. However, the grand mother declined in view of the fact that she did not see the need for surgical intervention. We counselled the grand mother on the need for her to allow the child to have ptosis surgery, but she was yet to accept at the time of this report. In view of this, we decided to review the child periodically and we intend to use the opportunity to encourage the maternal grand mother as well as the parents of the child to consent to surgical intervention most especially in view of the fact that the child had developed amblyopia. The periodic review...
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. In a case series report by Beckingsale et al, 39% of the 28 patients with blepharophimosis syndrome had amblyopia. The authors concluded that patients with blepharophimosis syndrome have a high rate of amblyopia. 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. 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. It is a pity that the case highlighted presented late and his maternal grandmother was yet to consent to ptosis surgery as at the time of this report. It is however, to the advantage of the patient that he did not have co-existing strabismus which could have doubled the risk of amblyopia in him.

Blepharophimosis syndrome is often bilateral and symmetrical. The patients usually have smaller than normal eyelid opening. There was however a report of unilateral blepharophimosis syndrome in China by Cai et al. Blepharophimosis syndrome is often associated with nasolacrimal drainage problems. This association is one of the reasons why we encouraged this patient to come for review periodically. Athapilly et al reported congenital alacrima in a patient with blepharophimosis syndrome.

The severity of ptosis in blepharophimosis syndrome may make children to adopt a chin-up, backwards head-tilt position and to recruit the frontalis in elevating the lids leading to raised arched eye brows. However, the later findings were not observed in our patient in spite of the severity of the bilateral ptosis. We suspect that the case highlighted is likely to be sporadic in view of the fact that we could not establish family history. It is interesting to note that our patient was said to be performing well in primary school. This is noteworthy considering the fact that blepharophimosis syndrome most especially the sporadic type is associated with mental retardation. Cai et al in China reported a novel case of unilateral blepharophimosis syndrome and mental retardation associated with de novo trisomy for chromosome 3q.

We conclude that early corrective surgery to prevent development of amblyopia in blepharophimosis syndrome is advised. This explains our decision to refer the patient for ptosis surgery for cosmetic reasons and to ensure restoration of normal visual development in the child.

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References


