Congenital Bilateral Coloboma of Upper Eyelid

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ABSTRACT

Congenital coloboma of eyelid is a rare anomaly. There is partial or total absence of eyelid structures. A seven year male child had coloboma of both the upper lids lateral to lacrimal puctum affecting the medial half of lid symmetrically with symblepharon in region of defect bilaterally. The study was carried out at Maharaja Krushna Chandra Gajpati Medical College Berhampur, Odisha in year 2010.

Both eyebrows were abnormal. He presented on and off diminution of vision, burning sensation, redness and watering from both the eyes on and off. On examination high refractive error was detected (visual aquity was 6/18 in righteye and 6/24 in left eye). Cornea was dry and opacities were present in both the eyes. There was limitation of ocular movement in both sides due to symblepharon. Nystagmus was present. The subject did not have any other associated anomaly. The birth and family history was normal. This case can be surgically treated and earliest management can give good fuctional as well as cosmetic results.

KEY WORDS

Congenital coloboma, eyelid, frontonasal process, symblepharon

INTRODUCTION

Congenital eyelid coloboma is a rare anomaly, was first noticed by Jacques Guillemeau in 1985 and also named it as paupieres accurcies.[1] It can be presented by neonate as well as later by children. Neonates came to clinician as relative emergency for corneal protection. In age of childhood, coloboma cases usually came for refractive error correction as well as cosmetic purpose.[2] There is partial or total absence of eyelid structures. It is caused by failure of fusion of mesodermal lid folds.[3] It usally seen in upper lid affecting medial third of the lid, but sometimes can affect lower lid as in mandibulofacial dysostosis.[2]

CASE REPORT

A seven year old male child came to outpatient department of ophthalmology at Maharaja Krushna Chandra Gajpati Medical College Berhampur, Odisha with bilateral upper eye lid defect (congenital coloboma) on 02-08-2010. He complained of diminution of vision, burning sensation, redness and watering from both eyes on and off for one year.

On examination, his visual equity in right eye was 6/18 and 6/24 in left eye. Cornea was dry with opacities seen in the both eyes (fig. 1 and 2). Nystagmus was also present. Medial half of upper eyelids lateral to lacrimal punctae

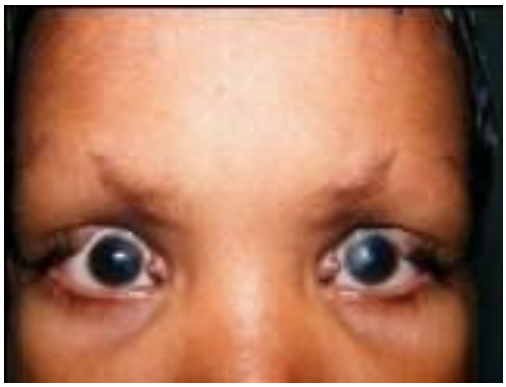

Figure 1. Bilateral coloboma involving medial half of upper eyelid symmetrically

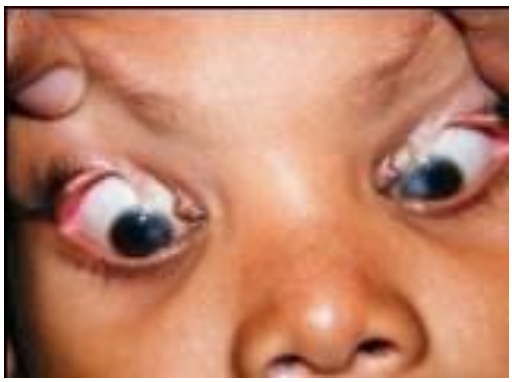

Figure 2. Symblepharon in the upper lid defect bilaterally

were absent in both sides symmetrically (fig. 1). Lacrimal punctae were normal on either side and well opposed to globe. Symblepharon, a fibrous adhesion or band between eyelid and conjunctiva were seen in both the eyes in the region of defect (fig. 2). Ocular movements was limited due to presence of symblepharon. Eyebrows were not normally developed on the both sides (fig. 1). But lower eyelid and punctae were well developed without any abnormality. Pupils, anterior chamber and fundi, iris were all normal. Intraocular pressure was also in normal limits. No other associated anomalies were found. The boy was born to non consanguineous parents after full term normal delivery. Post natal period was uneventful. There was no family history of similar or any other congenital anomalies. He was treated conservatively and sent for treatment for symblebharon, later on for reconstructive surgery of upper lids after taking proper consent.

DISCUSSION

Coloboma is a congenital disease with an incidence of 1 in 10000 live births.[4] Defect involves full thickness of the eyelid margin.[5] It can occur unilaterally or bilaterally, involving one or all four eyelids. It can present as a small notch to a complete absence of eyelid. Colobomas are frequently associated with systemic and other ocular anomalies. Keratoconus, coloboma of iris, dermoid, lipodermoid, and micro-ophthalmia are usually associated with congenital coloboma of eyelid. Sometimes it may occur as a part of multisystemic syndrome like Goldenhar and Treacher Collins syndrome or may be associated with a facial cleft.[2,6] Upper lid colobomas are more common than lower lid coloboma. Lower lid colobomas affects lateral half of lid as in mandibulofacial dysostosis in contrast to upper lid colobomas where lid defect present medially. Lower eyelid colobomas may present medially in case of lacrimal drainage channel abnormalities.[7]

A study was done by Betharia et al. on five cases of congenital colobomas of lids revealed that colobomas involve more than half of upper eyelids on medial side bilaterally in all cases.[1] Nystagmus was present in four out of five cases. They found an unusual abnormality of eyebrow (vertically placed) in one case.[1] Our present study goes in favour of Betharia et al. studies by the presence of both nystagmus and eyebrow.[1]

In studies carried out on twenty two patients of congenital upper lid colobomas, thirteen cases were associated with strabismus, fibrous adhesions (symblepharon), high refractive error and opacities in ocular media.[2] The present study had all the features except strabismus.

Studies have reported the congenital bilateral upper lid coloboma in infant of full term delivered normally. The postnatal period was uneventful. Baby was treated with lubricants and night time patching with plan for delayed surgical repair.[8]

In the management of congenital coloboma, symblepharon should be treated initially by excision and mucous membrane grafting. Subsequently eyelid defect has to be repaired. There is different line of treatment for congenital coloboma and coloboma secondary to trauma or tumor excision. Only proper freshening of edges of coloboma is required. Congenital coloboma involving one third of lid can be repaired by direct suturing after freshening of edges of defect. Canthotomy and cantholysis in addition to suturing should be done coloboma involving one half of lid. A defect more than half of lid requires sharing procedures e.g. Cutler Beard operation and Hughe's procedure, or lid sliding techniques e.g. Mustarde's rotation flap repair and Tenzel's semi- circular flap from the lateral canthal region.[1]

Embryological basis / Pathogenesis:

There are various theories regarding pathogenesis of colobomas. During intrauterine life lid developed normally by complex interaction and the union of mesodermal sheets of frontonasal process (upper lid) and maxillary process (lower lid). Any alteration, delay or interference in this interaction of developmental component can produce lid defect.[1]

Complete epithelial adherence of lid folds occurred at ninth week and its maintenance continued from tenth week to sixth month of gestation. Any local factor altering

this normal growth of lids and marginal structures results in palpebral colobomas and maldevelopment of eyelashes, punctae and other adjacent structures.[9]

Isolated colobomas usually caused by defect in eyelid fold. The most severe idiopathic type is due to failure of development of eyefold and less severe type is due to failure or defect in fusion of two lid folds at ninth week of gestation.[10] Interference in neural crest development and abnormal vascular system results in first arch syndromes and failure of fusion in some cases possibly associated with abnormal fibrous bands causes facial clefts.[2]

This case study on coloboma has revealed that coloboma may be a contributing factor for the occurrence of diminution of vision since birth. This case can be corrected surgically in order to bring back his vision. Hence it has been studied in detail and reported.

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