Case report

Congenital isolated bilateral upper lid coloboma

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Abstract

A case of congenital isolated coloboma of both the upper lids from just lateral to the lacrimal punctum up to the medial half, with symblepharon in the region of lower eyelid, was studied in a 7-year-old female child. She did not have any other associated anomalies. The birth and family histories were normal. The puncta were normal in position and well apposed to the globe. The closure of the lid coloboma was done by release of symblepharon along with direct closure of the defect, for the right eye first, and one month later, for the left eye.

Key-word: eyelid, coloboma

Introduction

Congenital eyelid colobomas are a partial or total absence of eyelid structures and are caused by failure of fusion of the mesodermal lid folds (Hoyama, 2007). It was first noted by Jacques Guillemeau (1585) who called it paupieres accurcies. It is a defect affecting primarily the lid margin where all portions of the lid structure are absent. It is usually seen in the upper lid in its medial one third but occasionally it maybe present in the lower lid as in mandibulofacial dysostosis Various associated findings which have been described are the presence of dermoids or dermolipo-mata (Betharia, 1988; Gribor, 1975), conjunctival chondroma, symblepharon, corneal opacities, cutaneous bridging, coloboma of the fundus and facial abnormalities (Betharia, 1988).

A case report

A case of congenital isolated coloboma of both the upper lids was studied in a 7-year-old female child. The coloboma was as large as half of the upper lids with symblepharon (Figure 1a). She also presented with diminution of vision in both eyes and had a history of redness, burning sensation, watering and discharge on both the eyes on and off. Family history didn't show association of such a defect in other family members. She had parents and an elder brother. She was born at term by normal delivery in hospital, without perinatal complications. There was neither any history of maternal illness during pregnancy nor the history of intake of any medications other than iron and folate tablets. She had normal developmental milestones. She had no other associated congenital anomalies on systemic evaluation.

On ocular examination, she had visual acuity of 6/24 in both eyes, which improved to 6/9 on right eye, and 6/18 on left eye. There was no abnormal head posture and no abnormality was detected on examination of forehead and eyebrow. She had upper lid coloboma from just lateral to the lacrimal punctum up to the medial half, with symblepharon in the region of defect and puncta were normal in position and well apposed to globe. Extra-ocular movement was normal, Bell's phenomenon was

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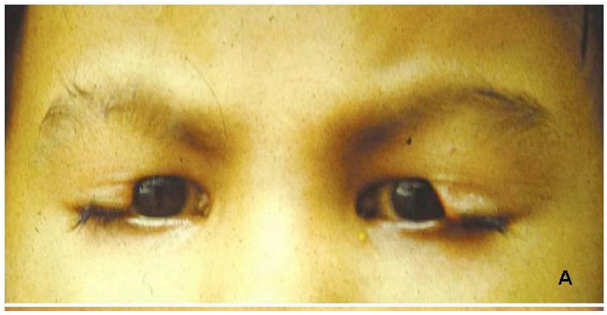
positive, convergence was normal. Cover test showed orthphoria. Symblepharon was present only in the region of upper lid coloboma of left eye, sclera and cornea were normal.

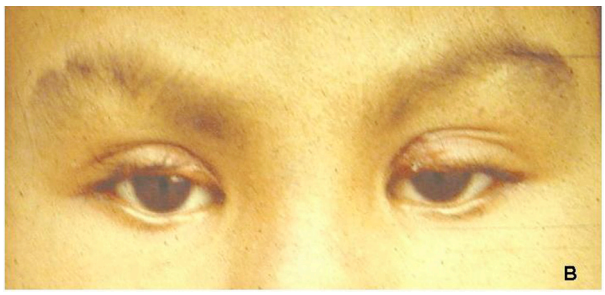
Anterior chamber was of normal depth and quiet, iris was normal, pupils were round, regular, reacting to direct and consensual light as well as accommodation reflex, no relative afferent pupillary defect (RAPD) was present. Lens and vitreous were normal. Fundus evaluation under mydriasis was also normal. Intra-ocular pressure was within normal limit, 11 mmHg in each eye with Perkins tonometer. Cycloplegic refraction revealed RE: +0.50DS 6/9 and LE: +2.00DS 6/18. Based on these observations, the diagnosis made was bilateral congenital upper lid coloboma with anisometropic amblyopia.

Her parents were informed of the condition and were advised for lid surgery. The closure of the lid coloboma was done by release of symblepharon along with direct closure of the defect for right eye first and one month later the left eye was done. The procedure of lateral canthotomy and cantholysis was performed. Margin of the coloboma incised to make the edges raw. Temporal upper lid was advanced nasally to close the colobomatous lid defect.

On follow up examination, the patient did well with the upper lids well apposed to the globe (Fig 2). Levator palpebrae superioris function was normal and no lagophthalmos was detected. A good functional and cosmetic result was achieved from the treatment. However amblyopia therapy did not help much to improve vision.

Figure 1a. Lid coloboma involving half of upper lids. Figure 1b. Closure of defect after surgery





Discussion

Most of the literature reports that surgical closure of upper lid coloboma is promising in terms of structural, function and cosmetic point of view. A flap from the lower lid using the lid switch technique and flap separation in a five month old baby presented with similar type of conditions was found to be successful (Adegbehingbe, 2005). The result is more promising when intervention is done earlier if the defect is larger than one third of the eyelid margin (Seah, 2002).

There are variable views regarding the normal develop-ment of the lids. However, it is believed that the normal development is the result of complex interactions and the union of mesodermal sheets of the frontonasal processes in the case of upper lid and similar maxillary processes for the lower lids. Any delay or interference in the interrelationship of developmental components can produce a lid defect (Betharia, 1988).

In the management of congenital coloboma, the presence of symblepharon or dermolipomata need initial treat-ment in the form of excision and mucous membrane grafting. The eyelid defect is repaired subsequently. Regarding the repair of such defects there seems to be some difference from the post-traumatic colobomas and eyelid defects following tumour excision. Since there is a clear and regular line of demarcation between skin & mucosa and absence of surrounding scar tissue in cases of congenital colobomas, the excision of the margin of the coloboma is unnecessary. Only proper freshening of the edges of the coloboma is required (Betharia, 1988).

Congenital colobomas involving one third of the lid can easily be repaired by direct suturing after proper

freshen-ing of the edges of the defect instead of one fourth of the eyelid defect due to other causes. A defect up to one half or more will require canthotomy and cantholysis in addition to direct suturing as was done in our case. Colobomas involving more than half of the lid requires lid 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 semicircular flap from the lateral canthal region.

These cases should be taken up for surgical manage-ment at the earliest to give good functional and cosmetic results (Betharia, 1988).

This case shows that lid coloboma could be an isolated problem in a child. Such a defect of the size mentioned can be corrected by canthotomy, cantholysis followed by direct closure, with good results.

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