Case Report

**Bilateral Congenital Eyelid Coloboma and its Complications in a 2-year-old Child**

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**Abstract:** Congenital eyelid coloboma is a partial or total absence of eyelid structures. It is present at birth and may cause damage to the cornea. This case report highlights the ocular complication of a bilateral coloboma in a 2-year-old child.

**Keywords:** Congenital eyelid, coloboma, cornea

**INTRODUCTION**

Coloboma is derived from the Greek “koloboma” meaning mutilated or curtailed. The malformation refers to a notch, gap, hole, or fissure in any of the ocular structures \([1, 2]\).

Congenital eyelid coloboma is a unilateral or bilateral, partial or full-thickness eyelid defect \([3, 4]\). It is an abnormality in which ocular structures are incompletely formed due to failure of the embryonic optic fissure to fuse. It occurs in 1: 10000 births \([5]\). Eyelid colobomas are most commonly triangular in shape with the base at the eyelid margin. It is usually located on the medial half of the upper lid or laterals half of the lower eyelid and can vary in size from a small indentation of the eyelid to near absence of the entire eyelid. Eyelid coloboma is rare and commonly occurs as an isolated finding. However, they have been reported to occur as part of a multisystem syndrome \([6]\). The eyelid coloboma usually presents with exposure keratopathy due to poor Bell's phenomenon; so it requires immediate attention, otherwise the cornea inevitably ulcerates and the eye becomes blind \([7]\).

**CASE REPORT**

A 2-year-old boy presented to us for exposure of eyeballs since birth. Birth and prenatal history was normal, the child was born at term. Family history did not show similar defect in the siblings. Best corrected visual acuity was light perception in the right eye, whereas in the left eye it was 6/5. On examination, the right eye had a large coloboma with almost the 2/3 of the upper lid missing medially but sparing the punctum. The cornea presented a total opacification with exposure keratitis; in addition, there was a symblepharon. In the left eye, the coloboma occupied the 1/3 of the upper lid medially; the punctum was spared; exposure keratitis was also noted. Both eyes were watering and discharging. No systemic disease was found after a checkup performed by an Internist; the patient was then scheduled for surgery.

**Fig. 1: Anterior view of the child with bilateral upper eyelid coloboma**
DISCUSSION

Congenital upper eyelid coloboma is a rare anomaly, with the unknown incidence. The size of the defect is different, but it always involves all layers of the eyelid. This malformation is more frequent at the upper eyelid, and unilaterally, at the junction of the medial two thirds [8].

It may be associated with several abnormalities such as conjunctival chondroma, symblepharon, corneal opacities, cutaneous bridging, coloboma of the fundus and facial abnormalities [9]. In some cases, coloboma may be associated with CHARGE syndrome; CHARGE stands for: Coloboma (eye), heart defects of any type, atresia (choanal), retardation (of growth and/or development), genital anomaly and ear anomaly. It was recommended that diagnosis of the syndrome be based on the presence of four of these physical features [10].

The pathogenesis of eyelid colobomas is not well understood since normal eyelid development does not produce clefting. It is possible that interference in the epithelial adhesion of the eyelid folds during gestation may cause maldevelopment of the eyelid and associated structures [11]. Alternatively, it has previously been postulated that central defects such as disturbances in the migration of neural crest cells may produce symmetric eyelid colobomas such as those seen in Treacher-Collins syndrome [12]. For some authors, eyelid coloboma is caused by failure of fusion of the mesodermal lid folds [13]. The optimal period for the operation depends on the size of the defect. If coloboma is smaller, operation can be delayed until the school time; but if the defect is larger, operation must be done as soon as possible in order to prevent corneal lesions [14].

Congenital colobomas involving one third of the lid can easily be repaired by direct suturing after proper freshening 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 [15].

CONCLUSION

Eyelid coloboma exposes the eyeball to external aggressive conditions with complication like keratitis, corneal sicca and even perforation. The current case illustrates the complications and the need to undertake the management very early.

REFERENCES