

Isolated Upper Eyelid Coloboma

İzole Üst Gözkapağı Koloboması

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Dear Editor,

We operated a 19-month-old child with congenital upper eyelid coloboma on the medial third of the right upper eyelid. The birth and family histories were ordinary. She did not have any other associated anomalies. The reconstruction of the eyelid coloboma was performed by release of symblepharon along with direct closure of the defect. The result was aesthetically acceptable.

Coloboma is a development failure of any structure (tarsus, iris, retina) related to the eyes. In the fourth week of embryonic development, a bud from the forebrain condenses into a globe. Normally, mesoderm and ectoderm migrate together in this stage. Divergences in this stage may cause defects on a wide spectrum varying from retina, macula, optic nerve, lens, or eyelid anomalies from a small notch to complete absence.

Congenital eyelid colobomas usually present with a full thickness defect of eyelids with a relation of bulbar conjunctiva and 90% of them affect the medial third of the eyelid.¹ In some cases, hereditary transition was reported. Chromosomal anomalies, such as Schmid-Fracaro syndrome and trisomy 18, and rare syndromes, such as CHARGE [C- coloboma, H- heart defects, A- atresia of choanae, G- genital defects, E- ear anomalies], Goldenhar syndrome (oculo-auriculo-vertebral dysplasia), Treacher Collins syndrome, Delleman syndrome, and frontonasal dysplasia, may cause coloboma and these can be detected on genetic analysis.²

Our patient was examined by pediatricians for associated anomalies. There were no special conditions in family history or the period of birth. There was an incomplete eyelid defect on middle third of the right upper eyelid which was eight mm in width and four mm in height and devoid of eyelashes (Figure 1). There were no other anatomic or functional anomalies related to the eye.

Surgery was performed under general anesthesia. A plastic globe protection device was applied for cornea protection. One millimeter of lateral portions of the upper eyelid and superior part of the defect areas was excised with a full thickness incision in a pentagonal shape. Conjunctival and epidermal parts of the tarsus were dissected carefully from the orbicularis oculi muscle. Two muscle flaps were prepared from two portions of the upper eyelid. Conjunctival flap was sutured with 6.0 absorbable sutures. After suturing two muscle flaps with 5.0 absorbable sutures, the skin was closed with 6.0 non-absorbable sutures under no tension. Dressings were applied to cover the wound (Figure 2). A good eyelid and skin contour was observed 6 months postoperatively (Figure 3).

Coloboma is a congenital malformation which occurs in 1:10000 births.³ In the fourth week of embryonic development, frontonasal and maxillary development begins, lasting up to the ninth week. In this period, external and internal factors, such as intrauterine amniotic band syndrome, chronic inflammation, lack of placental circulation, and mechanical stress, interrupt the normal development stages. However, these factors are still theoretical, and colobomas can be a part of various multi-systemic syndromes.⁴ An observational case series of 55 patients with eyelid coloboma treated by a single surgeon (JROC) between 1985 and 2005 revealed that only 29% of the eyelid colobomas are isolated and others are associated with other ocular (62%) and/or craniofacial (53%) abnormalities.⁵ Our patient had isolated and non-syndromic upper eyelid coloboma.

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Figure 1. Incomplete eyelid defect on the middle third of the right upper eyelid, 8 mm in width, 4 mm in height, and devoid of eyelashes



Figure 2. Intraoperative photo after the repair of the upper eyelid



Figure 3. A good eyelid and skin contour is observed 6 months postoperatively

A defect in the eyelid margin was first described in 1585 by Jacques Guillemeau. Colobomas generally present as a triangular defect on the middle third of the upper eyelid or a wide variety of defects ranging from a notch to total absence in the lateral portion of the lower eyelid. Eyelid colobomas predominantly affect the upper eyelid (93%) and are unilateral (76%). Lodhi et al.² reported 21 cases of congenital upper eyelid coloboma; of which 18 occurred in isolation with upper eyelid medial defect, 13 were bilateral, and five were unilateral. Our patient had a unilateral upper eyelid coloboma.

Surgical repair is not urgent unless there is an exposure on the cornea which may cause keratitis. Ideal correction can be performed 6 to 12 months after birth. In milder cases, surgery can be delayed to two to three years using natural tears. Our patient used lubricating solutions from the day she was diagnosed and operated on within a short period of time.

If the defect of the eyelid is smaller than 35% of the eyelid, it can be closed in primary manner. In defects ranging from 35% to 45%, a canthotomy and cantholysis should be performed for reducing the tension, but if the defect is greater than 45%, the patient needs a two-stage operation. In anterior defects narrower than 1/3, defect borders can be excised in a pentagonal shape and primary repair can be performed, but if there is an anterior and posterior defect, lamellar closure methods should be considered. We reconstructed the eyelid defect using a pentagonal primary repair procedure.

If lateral canthotomy and cantholysis are performed, it will reduce the tension on the horizontal plane. In smaller defects, semicircular rotation flap of Tenzel can be used. In defects greater than 1/3, Cutler Beard procedure should be considered for achieving a cosmetically acceptable result. Pendular eyelid flap is a novel technique used by Chalvatzis NT et al., and one stage-one site technique has been described by Dagi Glass LR for greater defects. In this procedure, a full thickness advancement flap from the lower eyelid is used to repair the upper eyelid defects.

In conclusion, anomalies related to eyelid can be in a wide spectrum. The causes of developmental failures are still theoretical, but a systemic and genetic analysis should be performed.

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