Bilateral upper lid coloboma at Bugando Medical Centre: A case report

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ABSTRACT

This is a rare case of bilateral upper eyelid coloboma in a neonate managed with early primary surgical correction. A three-day-old term neonate weighing 3 kilograms was referred from primary health facility to Bugando Medical Center, a tertiary referral hospital in Mwanza, Tanzania, following a spontaneous vaginal delivery. Ophthalmological examination revealed bilateral upper eyelid colobomas with exposure keratopathy with dryness and loss of luster. Systemic physical examination did not reveal any associated congenital anomalies such as craniofacial deformities, polydactyly, ear abnormalities, genital anomalies, or choanal atresia. On the first day post-admission, the patient underwent successful surgical repair via primary closure of the eyelid defects. Frost sutures were applied bilaterally to protect the cornea, and a good Bell's phenomenon was observed postoperatively. We conclude that prompt recognition and early surgical repair of eyelid colobomas are critical in preventing serious complications such as exposure keratopathy, corneal ulceration, xerosis, and permanent vision loss. This case highlights the importance of early intervention, even in resource-limited settings, and underscores the need for comprehensive systemic evaluation to rule out syndromic associations.

Key words: Lid coloboma, Eyelid defects, Lid reconstruction, Goldenhar Syndrome, Fraser Syndrome

INTRODUCTION

Congenital upper eyelid coloboma may be unilateral or bilateral and associated with systemic and ocular anomalies[1]. Untreated eyelid colobomas lead to vision loss and blindness from exposure keratopathy. Coloboma is mostly associated with systemic illnesses such as Goldenhar syndrome, Fraser syndrome, and Charge syndrome. The management varies according to site, defect size and associated complications[2].

All eyelid defects larger than one-third need early surgical management and a close follow-up for exposure keratopathy and amblyopia.

This case report notes that early diagnosis and treatment are required for a good cosmetic and visual outcome and quality of life of a neonate.

CASE PRESENTATION

A full-term three-day-old baby girl was referred to Bugando Medical Centre, with a history of normal spontaneous vaginal delivery at home who cried immediately after delivery and with a history of only a single antenatal clinic visit, no history of prior antenatal medications given and no history of ultrasound done prior.

On examination, her vitals were 97% oxygen saturation in room air, 37.1^{0}C temperature, 140 pulse rate, 44 respiration rate, 4.4 mmol/dl random blood glucose, and 3.0-kilogram weight. She also had a normal ECHO cardiography and abdomino-pelvic ultrasound with no signs of a syndromic baby.

On Ophthalomology review, she had bilateral upper eyelid defects of more than one-third. All preoperative evaluations were stable from ECHO, to serological blood work-ups (Full blood pictures, electrolytes) done with the help of the paediatric team. She was immediately booked for eyelid reconstruction to prevent corneal exposure.

Intra operative examination: The patient was examined under general anaesthesia. There was eyelid notching on the middle third of the upper eyelids bilaterally, with chemosis and conjunctival hyperemia, with a corneal diameter of 10mm, horizontal and 9.5mm vertical, with hazy cornea on the left eye more than the right eye. The anterior chambers are well-formed with the iris clearly seen bilaterally.

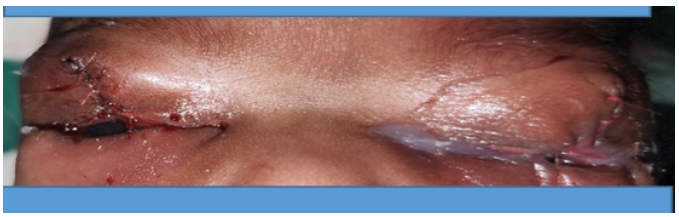
After a full ophthalmologic examination under general anaesthesia, the operation to repair the defects followed. The upper eyelid defect margins were freshened by splitting the edges at the grey margin, margins were

opposed with a stay suture of 4-0 silk and sutured with slight eversion with 6-0 vicyrl suture then tarsal plates were identified and sutured with the same half thickness 6-0 vicyrl suture and the subcutaneous tissue was opposed frost sutures applied and hemostasis achieved, ocular chloramphenicol ointment was applied in the fornices and the eyelids with vaseline impregnated gauze applied on top.

She had an uneventful postoperative period in the neonatal ICU one day post op with swelling of eyelid margins and sutures holding and was later discharged. The patient had good right eyelid movement and decreased left eyelid movement post-operatively.

Figure 1: Preoperative bilateral upper eyelid notching

Figure 2: Bilateral upper eyelid repair with frost suture on the left eye



DISCUSSION

Eyelid coloboma represents a congenital, full-thickness discontinuity of the eyelid margin. The defect may present unilaterally or bilaterally and can involve any of the four eyelids. Its presentation ranges from a subtle notch in the lid margin to complete absence of the eyelid[3]. Anatomically, colobomas most commonly affect the medial aspect of the upper eyelid and the lateral portion of the lower eyelid. Although relatively rare, eyelid colobomas may constitute an ophthalmic emergency in neonates due to the risk of corneal exposure and subsequent complications. They are frequently associated with a spectrum of ocular and systemic anomalies[4,5].

Ocular anomalies that may co-occur include dermoids, lipodermoids, keratoconus, iris colobomas, and microphthalmia. One notable genetic association is the nasopalpebral lipoma–coloboma syndrome, which is inherited in an autosomal dominant pattern with high penetrance. Furthermore, eyelid colobomas may form part of the phenotypic presentation of first branchial arch syndromes, such as Treacher Collins and Goldenhar syndromes, and may also coexist with craniofacial clefts[6].

The embryogenesis of eyelid coloboma is attributed to failure of eyelid fusion during critical developmental windows. Normal eyelid morphogenesis requires the adherence of eyelid folds by the ninth gestational week, with continued integrity through to the sixth month. Disruption of this process, whether by mechanical, genetic, or vascular influences, can result in colobomatous defects. Additionally, aberrations in neural crest cell migration and vascular development have been implicated in the pathogenesis of associated craniofacial syndromes. In certain cases, facial clefting may result from incomplete fusion, potentially due to anomalous fibrous bands[6]. Management of eyelid coloboma is guided primarily by the degree of corneal exposure. In instances where the corneal surface remains adequately protected, conservative measures such as topical lubricants, moisture chambers, and nocturnal patching may be employed to delay surgical intervention until the child is physiologically better suited for anaesthesia[5]. When surgical correction is indicated, the choice of technique is based on the size of the defect. Direct closure is typically feasible for defects involving less than 35% of the horizontal eyelid margin. For moderate defects (35–45%), adjunctive procedures such as lateral canthotomy or cantholysis may be required to reduce closure tension. In cases where the defect exceeds 45% of the eyelid length, staged reconstructive approaches, including the use of local flaps or grafts, are generally necessary to achieve optimal functional and cosmetic outcomes[7]. Surgical intervention is typically determined by the size of the defect. Defects involving less than 35% of the eyelid length can often be corrected with direct closure. When the defect extends between 35% and 45%, a lateral canthotomy or cantholysis may be necessary to ease closure and reduce wound tension. For defects exceeding 45%, a staged surgical approach is usually warranted[7].

Eyelid colobomas are commonly associated with syndromic conditions such as Delleman, Fraser, and Goldenhar syndromes. Although isolated occurrences are rare, they may present with midfacial anomalies. Surgical reconstruction of eyelid colobomas is considered a complex yet gratifying aspect of oculoplastic surgery. Lesions are frequently localized to the junction of the medial and central thirds of the eyelid. When significant corneal exposure is present, early surgical repair is essential to prevent irreversible complications such as corneal scarring or vision loss. Definitive reconstruction may be achieved in a single stage or require a staged approach depending on the extent of the defect[7].

CONCLUSION

Eyelid colobomas, though rare, represent a significant congenital anomaly with potential for serious ophthalmic complications if not promptly recognised and appropriately managed. Their association with systemic and ocular anomalies necessitates a thorough multidisciplinary evaluation. Early diagnosis, coupled with careful assessment of corneal exposure, is critical in guiding timely intervention. While minor defects may be managed conservatively in the short term, larger or vision-threatening lesions often require surgical correction, tailored to the size and location of the defect. Ultimately, successful outcomes depend on individualized treatment planning and may involve staged reconstructive procedures. Awareness of associated syndromic presentations and embryological underpinnings further enhances diagnostic accuracy and optimizes patient care.

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