Case Report

Congenital Ectropion in Down's Syndrome: A Case Report

Islam MR[1], Akther KU[2], Ghosal S[3], Siddiqua A[4]

Abstract

Congenital bilateral ectropion of the eyelids is a rare, benign condition reported in ophthalmic literature. It is more frequently associated with Down's syndrome, ichthyosis and sporadic cases in newborns from the black population. A rare case of primary congenital ectropion of all four eyelids in a child with Down's syndrome is reported to emphasize the problems of surgical management. Congenital ectropion is associated with other eyelid abnormalities and usually requires surgical measures to protect the cornea.

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Introduction

Congenital ectropion (eyelid eversion), a rare medical condition where the eyelids turn outward, can cause concern among parents and caregivers of affected newborns. Though the etiology is unknown, several intrauterine and genetic factors have been associated[1]. Using medications and watchful waiting, and through surgical correction are the treatment options for this condition[1]. Although this condition is said to be rare, this article reports a case of congenital ectropion with down syndrome and common cold.

Case Report

An 8-month-old boy presented with clinical features of common cold and Down's syndrome like generalized hypotonia, upward slant of eyes, ectropion (figure-1), low set ears, small mouth with protruding tongue (figure-2), simian crease and clinodactyly. He was the first child of a 25-year-old woman. Pregnancy was uneventful with regular antenatal follow-ups. The child was full-term and was born with normal vaginal delivery, with no instrumentation. His bilateral ectropion of the upper eyelids since the neonatal period. His karyotyping was 47, XY, +21, cardiovascular examination revealed normal findings. Epicanthic folds, bilateral ectropion, euryblepharon and lid retraction were observed on ophthalmic examination. Exposure keratitis was present and there was no retinal detachment or cataract. Vision was grossly intact but visual acuity cannot be assessed due to the uncooperativeness of patient. His intraocular pressure was normal. The case was diagnosed as congenital ectropion with Down's Syndrome.

Figure-1: Bilateral ectropion in the study case

Figure-2: Ectropion with protruded tongue in the study case

[1]Md. Rafiqul Islam, Assistant Professor, Dept. of Pediatrics, Gonoshasthaya Samajvittik Medical College Hospital, Dhaka, Bangladesh.
[2]Khandokar Urmina Akther, Resident, Dept. of Pediatric Neurology, Bangabandhu Sheikh Mujib Medical University, Dhaka, Bangladesh.
[3]Sharmistha Ghosal, Junior Consultant, Dept. of Pediatrics, Government Employee Hospital, Dhaka, Bangladesh.
[4]Ayesha Siddiqua, Assistant Registrar, Dept. of Pediatric Gastroenterology, Dhaka Medical College Hospital, Dhaka, Bangladesh.

Address of Correspondence: Dr. Md. Rafiqul Islam, Assistant Professor, Department of Pediatrics, Gonoshasthaya Samajvittik Medical College Hospital, Dhaka, Bangladesh. Mobile: +8801723775820. Email: rafiqulislamgsvmc@gmail.com

Discussion

Congenital ectropion of the upper eyelid was initially described by Adams in 1896[2-4]. After that, Gilbert and co-workers described two more cases of congenital ectropion associated with Down's syndrome[5,6]. There have been more reports of this uncommon disease in Black infants[1-4,7,8] associated with ichthyosis[1,4,9,10] and trisomy 21[2,5,8,11-14]. Usually the condition is bilateral and asymmetrical, some unilateral cases also have been identified[8,11,13]. Down's syndrome encompasses numerous ocular abnormalities like myopia, keratoconus, nystagmus, epiblepharon, brushfields spots, hypertelorism, epicanthus, convergent strabismus, cataracts, blepharoconjunctivitis with the epicanthal folds, and the most noticeable periocular findings are the characteristic mongoloid slant to the eyelid fissures[6,12]. Although the exact causes of congenital upper eyelid ectropion is unknown, several factors have been suggested to play a role in its development, including lack of functional lateral canthal ligaments, lateral eyelid elongation, orbicularis hypotonia, vertical shortening of the anterior lamella and orbital septum's inability to fuse with the levator aponeurosis[1,2,4,6,11-14].

Treatment for congenital ectropion of the upper eyelid is controversial. Numerous methods have been recommended[15]. For mild cases, simple medical management such as using lubricating ointments and creating a moist environment may suffice to prevent dryness of the exposed conjunctiva, reduce swelling and allow the eyelid to naturally invert within a few weeks[3,7,11,13]. More severe cases that do not respond to conservative treatment may require surgical options such as injecting hyaluronic acid under the conjunctiva[4,8,13], tarsorrhaphy (partial or complete closure of the eyelids)[2,3,6,7], excising excess conjunctiva during tarsorrhaphy[5,7], suturing the fornix[3,13], using a full-thickness skin graft[1,2,5,11], performing full-thickness horizontal lid shortening[2,6] or attaching the orbital septum to the levator aponeurosis[2]. While most cases of congenital ectropion not associated with Down's syndrome improve with methods like eyelid patching, taping and ointment use[3,6,7,11,13]. However, surgical treatment may be necessary for those with Down's syndrome[6,12,14].

Exposure keratopathy prevention is one of the major goals of management. Treatment for congenital ectropion in such cases should involve shortening the anterior lamella by using full-thickness skin grafts that extend beyond the horizontal limb of the canthal tendon to compensate for potential graft contraction. Furthermore, full-thickness pentagonal lid resection as well as lateral and medial tarsal strip procedures are required to treat horizontal lid laxity. In our case, we advised surgical correction and regular follow up. However, the parents declined the surgery and did not continue the follow up.

Conclusion

Congenital ectropion is a rare abnormality that can threaten the cornea and visual acuity if not treated early. Creating awareness among pediatricians and ophthalmologists about the eye findings in Down's syndrome is the main concern for this report. It may resolve spontaneously or may need surgical correction to avoid exposure to keratopathy.

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