DOWN SENDROMLU YENİDOĞANDA ÜST GÖZ KAPAĞI EVERSİYONU

UPPER EYELID EVERSION IN A NEWBORN HAVING DOWN SYNDROME

Ümit Kamış,* Zekeriya Tosun,** Hürkan Kerimoglu,*** Banu Turgut Öztürk,*** Mustafa Avunduk.****

* Selcuk University, Meram Faculty of Medicine, Department of Ophthalmology
*** Selcuk University, Meram Faculty of Medicine, Department of Plastic and Reconstructive Surgery
*** Selcuk University, Meram Faculty of Medicine, Department of Ophthalmology
**** Selcuk University, Meram Faculty of Medicine, Department of Pathology

ÖZET

Olgu sunumu: Kliniğimize başvuran 3 günlük kız bebeğin her iki üst göz kapağı dışa dönüktü ve kapak aralığından protrude, konjonktival kemozis mevcuttu. Olgunun diğer sistemik muayene bulguları ve sitogenetik incelemeler sonucunda Down sendromu tanısı konuldu. Konservatif tedavi ile izlem sonrası klinik düzelme sağlanamaması üzerine konjontivadaki kemozisin cerrahi girişimle eksizyonunu yapıldı. Histopatolojik incelemede kapilerlerde dilatasyon ve kanama tespit edildi. Bunun kapağın eversiyonuna bağlı konjonktiva konjesyonundan kaynaklandığı düşünüldü.

Sonuç: Konjenital üst kapak eversiyonu nadir görülen sıklıkla Down sendromu ile birliktelik gösteren bir durumdur. Görsel aksı kapatacak kadar yoğun kemozise neden olabilmekte ve bu durumun tedavisinde cerrahi tedavi gerekebilmektedir.

Anahtar kelimeler: Kapak eversiyonu, Down sendromu, konjenital anomali

ABSTRACT

Case report: A 3- days- old female infant presented to our clinic with bilateral everted, upper eyelids and conjunctival chemosis protruding from interpalpebral area. The systemic findings and cytogenetic study revealed the diagnosis of Down syndrome. Following a period of conservative treatment without recovery, surgical excision of conjunctival chemosis has been peformed. Histopathological analysis revealed dilation of capillaries and hemorrhage which was assumed to be due to the congestion of the conjunctiva as a result of eyelid eversion.

Comment: Congenital upper eyelid eversion is a rare condition associated frequently with Down syndrome. It may cause serious conjunctival chemosis occluding the visual axis and may warrant surgical treatment.

Key words: Kapak eversiyonu, Down syndrome, Congenital Abnormalities

INTRODUCTION

Interruption of lid development during normal embryogenesis may cause a wide spectrum of congenital anomalies, however congenital eversion of eyelids are rarely seen. Its etiology is unknown, however it is frequently associated with Down syndrome. In this report we present bilateral eyelid eversion in a baby with Down syndrome to discuss its course and management.

CASE REPORT

A 3- days- old female infant, born after an uncomplicated pregnancy and delivery presented to our clinic with bilateral chemosis protruding from the everted, upper eyelids which obstructs the visual axis bilaterally (Figure 1).

Physical examination of the baby revealed normal findings except simian crease and drop ear deformity which warrants a cytogenetic study to investigate Down syndrome. Cytogenetic findings with G banding method demonstrated a regular type Down syndrome (47 XX, +21). MR Angiography showed bilateral well circumscribed lesions which demonstrate hyperintensity on T2 weighed images and homogeneous enhancement after administration of contrast agent. Conservative treatment with topical lubricants and ointments in addition to eyepatching after repositioning the eyelids were applied for 2 weeks. Unfortunately no improvement was observed. As visual axis was occluded totally, we decided to perform en bloc excision to prevent deprivation ambliyopia.

In her examination, under general anesthesia, before the operation, both eyes were normal in size

Geliş Tarihi :06-08-2009
Kabul Tarihi : 25-02-2011

Figure 1. Photograph of the newborn at her first visit. Upper eyelids are bilaterally everted and a red mass is protruding.

Figure 2. Postoperative photograph of the baby after total excision of the lesion.

and no abnormality was noted in the anterior segment structures. Intraocular pressures measured with Perkins tonometer were within normal limits and both fundi were unremarkable. Chemotic conjunctiva was excised and sent to the laboratory for pathological evaluation. The operation has been completed following the lid reconstruction with a wedge resection at the lateral cantus for both eyes (Figure 2).

Unfortunately the baby developed cardiac arrest a few hours after the operation which answered to resusitation initially. During his observation in the newborn unit cardiac insufficiency and lung infection developed within the following days, necessitating mechanical ventilation and respiratory support. Despite all efforts the patients condition deteriorated resulting in exitus in the 35th day after birth.

Although parents of the baby signed the consent form for publication, they did not allow a postmortem autopsy. The histopathological analysis of the excision material revealed dilation of capillaries and hemorrhage. This finding may be explained by congestion of the conjunctiva due to eversion of the eyelids.

DISCUSSION

Down syndrome, known as the most common chromosomal anomaly, encompasses numerious ocular abnormalities like epicanthal fold, hypertelorism, epiblepharon, ectropion etc. Among these eyelid deformities constitute a wide spectrum like upward slanting of the palpebral fissure, hypertelorism, epicanthus, epiblepharon, ectropion, and upper lid eversion.[1-3] Congenital eversion of the upper eyelids was first described by Adams in 1896 who called it as “double congenital ectropion”.[4] This acute ectropion is reported most frequently in Down syndrome, black babies and difficult deliveries.[5] Abnormalities like orbicularis hypotonia, birth trauma, vertical shortening of the anterior lamella or vertical elongation of the posterior lamella of the eyelid and failure of the orbital septum to fuse with the levator aponeurosis, absence of effective, lateral canthal ligament and lateral elongation of the eyelid have all been implicated as possible pathophysiological factors.[6]

This bilateral condition becomes usually evident intermittently when the child cries. Venous stasis

during delivery may cause marked chemosis and prolapse of the conjunctiva which may obscure the globe. This may recover spontaneously. Surgery is recommended only if initial attempts of pressure patching or repositioning of the lids and taping fails.[7-9]

In our case no remarkable change was noticed during the conservative treatment period. Surgical excision was undergone to avoid obstruction of the visual axis. In the literature other surgical treatment options like temporary tarsorraphy, subconjunctival injection of hyaluronic acid, fornix sutures and full thickness skin graft to the upper eyelid are reported.[10] We preferred to excise the excess conjunctiva to confirm the diagnosis.

Upper eyelid eversion is a rare abnormality and may threat the vision if not treated early. As it is to our knowledge synaptic connections are established permanently during the critical period for visual development and a retarded treatment may be less effective for restoring vision.[11] However risks of general anesthesia shoud be also kept in mind when deciding the for surgical treatment.

Dr. Banu Turgut Öztürk
Selçuk Üniversitesi, Meram Tıp Fakültesi, Göz Hastalıkları AD
42080 Akyokuş-Konya / TÜRKİYE
Tel: +90 (332) 223 6210
Fax: +90 (332) 223 6181
E-mail: ozturkbanuturgut@yahoo.com

REFERENCES:

1. Kim JH, Hwang J-M, Kim HJ, Yu YS. Characteristic ocular findings in Asian children with Down syndrome. Eye. 2002; 16: 710-14.
2. Berk AT, Saatci AO, Ercal MD, Tunc M, Ergin M. Ocular findings in 55 patients with Down's syndrome. Ophthalmic Genet. 1996; 17(1): 15-9.
3. da Cunha RP, Moreira JB. Ocular findings in Down's syndrome. Am J Ophthalmol 1996; 122(2): 236-44.
4. Adams AL. A case of double congenital ectropion. Med Forthnight 1896; 9: 137-8.
5. Sellar PW, Bryras JH, Archer DB. Late presentation of congenital ectropion of the eyelids in a child with Down's syndrome: A case report and review of the literature. J Pediatr Ophthalmol Strabismus 1992; 29: 64-7.
6. Al Hussain HA, Al-Rajhi AA, Al-Qahtani S, Meyer D. Congenital upper eyelid eversion complicated by corneal perforation. Br J Ophthalmol 2005; 89: 771.
7. Rabb EL, Saphir RL. Congenital eyelid eversion with orbicularis spasm. J Pediatr Ophthalmol Strabismus 1985; 22: 125-8.
8. Kronish JW, Lingua R. Pressure patch treatment for congenital upper eyelid eversion. Arch Ophthalmol. 1991;109:767-8.
9. Watts MT, Dapling RB. Congenital eversion of the upper eyelid: a case report. Ophthal Plast Reconstr Surg 1995: 11(4): 293-5.
10. Maheshwari R, Maheshwari S. Congenital eversion of upper eyelids: Case report and management Indian J Ophthalmol 2006; 54: 203-4.
11. Donahue SP. Screening Chapter 8. In Pediatric Ophthalmology and Strabismus ed. Taylor D, Hoyt CS. 3rd ed, Philedelphia, USA: Elsevier Saunders; 2005: 62.