CASE REPORT

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Congenital upper eyelid coloboma with ipsilateral eyebrow hypoplasia

Urođeni defekt gornjeg kapka sa istostranom hipoplazijom obrve

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Abstract

Introduction. Coloboma is a Greek word, which describes the defect of all layers of the organ, and it can be congenital or as the result of an injury, operation, or some disease. 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. Sometimes, it can also involve the eye, and may be a component of many syndromes (Goldenhar, Fraser, Manitoba, CHARGE, Cat eye). **Case report.** We are describing the case of the upper eyelid coloboma with the rare eyebrow anomaly at the three- month old girl, and the result of reconstruction. The baby was treated conservatively with lubricants and overnight patching. Pentagonal excision of the defect was performed in general anesthesia. Three layers of the eyelid were prepared: the skin, muscle and tarsoconjunctival layer. Because of orbicularis muscle malposition, reinsertion and reposition of the muscle fibres were performed. Then, lateral canthotomy was made and the suture of three layers of the eyelid. Catgut suture 7–0 was used for the conjunctiva and muscle. Nylon 6–0 was used for skin suture. Z-plasty was done on the upper part of the pentagonal excision in order to reduce skin tension at the suture line. The operation lasted about 60 minutes and the hospitalization three days. The occlusive dressing was applied for two days. The stitches were removed after seven days. The postoperative swelling of the upper and lower eyelid disappeared in five days. There were no complications in the postoperative period. **Conclusion.** The main principle of the treatment of eyelid coloboma is surgical reconstruction of all layers of the eyelid, in optimal period, using different surgical methods, which depends on the size of the defect. An early diagnosis is of the greatest importance, as well as the treatment of associated anomalies. Complications of the upper eyelid coloboma depend on the size of the defect, presence of the eye anomalies and the method of reconstruction.

Key words:
eye abnormalities; eyelids; coloboma; eyebrows; reconstructive surgical procedures; treatment outcome.

Apstrakt

Uvod. Kolobom je grčka reč koja opisuje defekt svih slojeva organa, a može biti kongenitalni ili posledica povrede, operacije ili neke bolesti. Kongenitalni kolobom gornjeg očnog kapka je retka anomalija, sa nepoznatom incidencijom. Veličina defekta je različita, ali uvek uključuje sve slojeve kapka. Ova malformacija je češća na gornjem kapku i jednostrano, na spoju medijalne dve trećine. Nekada, može da zahvati oko, a može biti i sastavni deo mnogih sindroma (Goldenhar, Fraser, Manitoba, CHARGE, Cat eye). **Prikaz bolesnika.** U radu je opisan kolobom gornjeg očnog kapka udružen sa retkom anomalijom obrve kod devojčice stare tri meseca, kao i postrekonstruktivni rezultat. Beba je lečena konzervativno lubrikantima i u toku noći primenom zavoja. Petougaona ekscizija defekta sprovedena je u opštoj anesteziji. Ispreparisana su tri sloja kapka: koža, mišić i tarzokonjuktivalni sloj. Zbog malpozicije orbikularnog mišića urađena je reinsercija i repozicija mišićnih vlakana. Učinjena je, zatim, lateralna kantotomija i sutura očnog kapka u tri sloja. Ketgut 7–0 je korišćen za suturu konjunktive i mišića, a najlon 6–0 je korišćen za kožu. U gornjem delu ekscizione rane urađena je Z-plastika u cilju smanjenja napetosti kože duž suturne linije. Operacija je trajala 60 minuta, a hospitalizacija 3 dana. Okluzivni zavoj bio je postavljen 2 dana. Šavovi su uklonjeni nakon 7 dana. Postoperativni otok gornjeg i donjeg kapka iščezao je nakon 5 dana. Nije bilo komplikacija u postoperativnom periodu. **Zaključak.** Osnovni princip lečenja koloboma očnih kapaka je hirurška rekonstrukcija svih slojeva kapka, u optimalnom periodu, korišćenjem različitih hirurških metoda, u zavisnosti od veličine defekta. Rana dijagnoza je od najvećeg značaja, kao i tretman udruženih anomalija. Komplikacije koloboma gornjeg očnog kapka zavise od veličine defekta, prisustva anomalija očiju i metode rekonstrukcije.

Ključne reči:
oko, anomalije; očni kapci; oko, defekt; obrve; hirurgija, rekonstruktivna, procedure; lečenje, ishod.

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Introduction

Coloboma, a Greek word, meaning defect is ysed for a defect of all the layers of the eyelid. Coloboma may be congenital or a result of injury, operation, or some disease.

Congenital upper eyelid coloboma is a rare anomaly of unknown incidence. The size of the defect varies, but it always involves all the layers of the eyelid. This malformation is more frequent in the upper eyelid, unilaterally, and at the junction of the medial two thirds. Sometimes a defect can involve eye, and may be a component of many syndromes (Goldenhar, Fraser, Manitoba, Charge, Cat eye). The main principle of the treatment is surgical reconstruction of all eyelid layers in optimal period, using different surgical methods, which depends on the size of the defect. An early diagnosis is of the greatest importance, as well as the treatment of associated anomalies. Complications of untreated upper eyelid coloboma depend on the dimensions of the defect, the presence of eye anomalies and the method of reconstruction.

We presented a 3-month-old girl with upper eyelid congenital coloboma and the eyebrow anomaly, and the result of the reconstruction. The main purpose of this report was to describe a rare, combined congenital malformation and the patient's condition after surgical treatment, as well a, to paint out the importance of multidisciplinary approach in the early diagnosis and the treatment of this anomaly.

Case report

A 3-month-old girl was referred to the Plastic Surgery Unit by the ophthalmologist. The main diagnosis was congenital upper right eyelid coloboma. Coloboma was on the medial third of the upper eyelid. The girl had ipsilateral eyebrow hypoplasia and malposition. The rest of the physical examination was normal. There was no family history of similiar or other congenital anomalies (Figure 1).

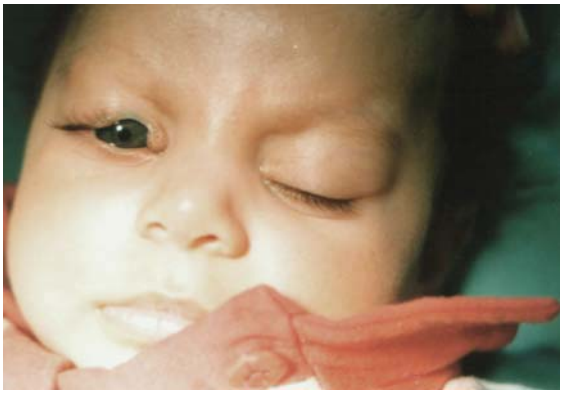

Fig. 1 – A three-month-old girl before surgical treatment

The baby was treated conservatively with lubricants and overnight patching. The pentagonal excision of the defect was performed in general anesthesia. The three layers of the eyelid were prepared: the skin, muscle and the tarsoconjunctival layer. Because of the malposition of the orbicularis muscle, reinsertion and reposition of the muscle fibres were performed. Then, lateral canthotomy was made and suture of the three layers of the eyelid. Catgut suture 7–0 was used for the conjunctiva and muscle. Nylon 6–0 was used for skin suture. Z-plasty was done on the upper part of pentagonal excision in order to reduce skin tension at the suture line. The operation took about 60 minutes and the hospitalization three days. The occlusive dressing was applied for two days. Stitches were removed after seven days. The postoperative swelling of the upper and lower eyelid disappeared in five days. There were no complications in the postoperative period. The girl underwent a regular control by the plastic surgeon and the ophthalmologist. Correction of eyebrow deformity was recommended in a later period (Figure 2).

Fig. 2 – The result of surgical treatment after 3 years

Discussion

Congenital eyelid coloboma was first described by Walter Eyre Lambert in 1901 considering the anomaly of the upper left eyelid. It is supposed that this malformation derives from the incomplete fusion of the mesodermal clefts during the gestation period, under the influence of PAX2 gene [1]. It is possible that many factors, such as radiation, may have some influence on the development of the eyelid. Upper eyelid coloboma is a rare congenital anomaly of unknown incidence.

Grover et al. [2] reported his results of a retrospective study which covered the period of 18 years and 51 patients with congenital coloboma of both eyelids. Upper eyelid coloboma was more frequent (74.5%), and defect of the eyelid was less than 40% in 57.9% patients. In our hospital the incidence of this malformation is about 1 : 9000 during a 5-year-period. There are no relevant data about sex, race and size distribution of this anomaly. Coloboma is the most frequent at the junction of the medial two thirds of the upper eyelid, and laterally in the lower eyelid. There are also some reports about bilateral upper eyelid coloboma [3].

Congenital upper eyelid coloboma is sometimes associated with other anomalies. Cryptophthalmos is present in the Fraser syndrome [4] and in Manitoba Oculotrichial syndrome (MOTA) [5, 6]. CHARGE syndrome is described as a syndrome with eyelid coloboma, lip and/or palate cleft, ear

anomaly, choanal athresia, brain and cardiovascular anomalies and slow psycho- somatical development [7]. Goldenhar syndrome Oculo Auriculo Ventebral (OAV) syndrome includes upper eyelid coloboma, ear, nose, palate, lip and mandibular hypoplasia, and eye lipodermoid cyst, as well as strabismus [6, 8]. In some cases, coloboma may involve eye (iris, lens, choroidea, retina, optical nerve) – Cat eye syndrome. Nouby [9] indicated in his study that 3 out of 26 children with eyelid coloboma had cryptophthalmos and 11 were found with some facial anomalies, like first cleft syndrome. Nouby made the classification of eyelid coloboma: coloboma without cryptophthalmos, coloboma with partial cryptophthalmos, coloboma with total criptophthalmos, classical cryptophthalmos and severe cryptophthalmos (severe nose deformities and lower eyelid ectropion). Lower eyelid coloboma is a rare anomaly, but it is usually a part of the Treacher Collins syndrome.

Eyelid coloboma treatment is surgical and depends on the size of the defect [10]. A team approach is necessary for the early diagnosis and treatment of accompanied anomalies, and sometimes there is a need for genetics consultation. The ophthalmological examinations of the newborn with eyelid coloboma must include the anterior segment (symblepharon, conjunctival caruncular malformation, keratopathy, choroidal coloboma, eyelid lypodermoid cyst), as well as the posterior segment (retinal and optical nerve coloboma). Computed tomography (CT) is reserved for Treacher Collins syndrome, for patients with lower eyelid coloboma.

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 [11], but if the defect is larger, operation must be done as soon as possible in order to prevent corneal lesions. In the meanwhile, topical lubricants and wet dressing are recommended. Surgical methods depend on the size of defects, but excision and reconstruction of all eyelid layers remain necessary. There are many surgical methods that can be used: direct suture, local, regional and composite skin flaps [12]. Sometimes, there is a need for two-stage operation. Skin or other flaps are necessary when the defect involves more than 40% of the upper eyelid. The most common methods in those cases are Cuttler-Beard, modified Hughes technique, Tensel or modified Tensel flap and full, thickness cross-flap from the lower eyelid.

Conclusion

We presented a very rare congenital, combined upper eyelid anomaly. In this case, there was no need for the use of a flap, but we performed lateral canthotomy and Z-plasty, because of the borderline defect size. In our opinion, the desinsertion and reposition of the orbicularis muscle are very important, like in cleft lip operations. Also, pentagonal excision may have some advantages on simple V-excision, because suture line is at the right angle and at the eyelid border. Reconstruction of eyebrow deformity can be performed later, using some of the numerous known surgical methods.

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