Bilateral Congenital Microblepharon Associated with Down’s Syndrome – A Case Report

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Abstract. Microblepharon is an atypical condition of the eyelids typically coupled with various ocular and systemic inherited abnormalities, as mentioned in several works of literature. To prevent corneal exposure and to attain an excellent visual outcome, urgent surgical intervention is needed in such cases. Our reported case is a bilateral upper lid congenital microblepharon with ectropion associated with Down’s syndrome and its management.

Keywords: Eyelid anomaly, microblepharon, ectropion, down’s syndrome.

INTRODUCTION

A microblepharon is characterized by shortening of the upper eyelid lamella anteriorly. Cornaz first stated this condition in 1848 [1], and Fuchs described its features in 1885 [2]. Associated congenital anomalies include microphthalmos, lid coloboma, absent puncta, and tetrastichiasis with anomalies of the limbs, genitals, face, and skull [3–7]. Isolated microblepharon case without systemic and ocular abnormalities has also been reported.

CASE REPORT

Since birth, a 9-month-old Down’s syndrome infant has been complaining of inability to close eyelids, photophobia, white staining of the corneas, and discharge from both eyes. Although we did not do karyotyping, Down’s syndrome was diagnosed based on facial phenotypic features. An ophthalmologic examination revealed bilateral upper eyelid microblepharon associated with ectropion and significant lagophthalmos in both eyes, as well as exposure keratopathy, with a greater degree in the right eye (Figure 1). The patient was prescribed topical antibiotics and frequent lubrication and referred to the paediatrician for thorough systemic evaluation. The pediatric consultation revealed no systemic anomaly, and we scheduled for urgent surgical modification of the bilateral upper lid microblepharon with ectropion correction to attain eyelid closure. A graft from the retro-auricular area was used for a full-thickness skin graft on the upper eyelids on either side (Figure 2). Ectropion was almost corrected following skin graft, although lateral lid tightening was done by lateral tarsal strip (LTS) procedure. Postoperatively there was minimal lagophthalmos, and keratopathy was resolved (Figure 3).

DISCUSSION

Microblepharon is a rare hereditary anomaly categorized by vertical shortening of anterior lamellae of the eyelids [1].
Microblepharon is a bilateral condition, but it may occur unilaterally on rare occasions [8]. Microblepharon presents various clinical signs, i.e., mild lagophthalmos with watering, photophobia, ectropion, and severe lagophthalmos, experiencing corneal exposure due to inadequate eyelid closure. In some literature, microblepharon has been reported in a still-born [9]. Children with ectropion and bilateral aphakic microphthalmos have also been reported [10]. Merriem et al. described microblepharon as a combination of multiple congenital anomalies [11]. Additionally, it has been reported with chromosomal abnormalities such as trisomy 21 and trisomy 12p [12]. Among the ocular deformities associated with Down’s syndrome are myopia, keratoconus, nystagmus, epiblepharon, brush field spots, hypertelorism, epicanthus, convergent strabismus, cataract, blepharoconjunctivitis with the epicanthal folds, and the mongolid slant to the eyelid fissures [13]. In our case, no other ocular comorbidities were noted except mongolid slant to the eyelid fissures on examination under anaesthesia.

From an embryological perspective, the lower eyelids develop from the maxillary process, while the upper eyelids develop from the frontonasal prominence [14]. Around day 45, the upper and lower lid folds begin to form, grow towards each other, and fuse around the 3rd month, and they separate again around the 6th month. During developmental stages, the mesenchyme of the lid fold is formed from the neural crest, and its deficiency results in microblepharon.

The management of the microblepharon depends on its severity. In the case of mild lagophthalmos, the cornea is well protected from being treated with lubricating eye drops and ointment. In severe lagophthalmos, cornea exposure leads to keratitis and may need surgical intervention to preserve vision. The surgical processes principally involve eyelid reconstruction with free, full-thickness skin grafts or pedicle flaps. In our case, we did the upper lid reconstruction with full-thickness skin grafts. Postoperatively, there was negligible lagophthalmos. Subsequently, the keratopathy healed, the left cornea is clear and only a mild scar on the right cornea. Our observation is urgent surgical intervention is needed to attain good corneal coverage and preserve sight. The child is on follow-up and may require additional visual rehabilitation.

**Statement of Ethics:** The patient’s parents provided knowledgeable consent to publish this case report (text and photographs). The institute’s committee approved the study protocol on human research. The study involved no animal research.

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**REFERENCES**