Ankyloblepharon Filiforme Adnatum in a Newborn

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Abstract

Ankyloblepharon filiforme adnatum (AFA) is a rare congenital anomaly consisting of a partial or complete fusion of the eyelid margins. It is usually an isolated and benign malformation but its presence should alert the neonatologist because it can be rarely associated to other important multisystemic disorders. We report a case of a newborn presenting with isolated AFA at birth, treated in the first day of life.

Keywords

- ankyloblepharon filiforme adnatum
- neonatology
- ophthalmology
- congenital malformations

Case Report

Initial evaluation of a full term newborn some hours after birth showed the presence of partially fused left eyelids (►**Fig. 1**). The baby was born from a cesarean section for an altered cardiotochographic pattern at 41 + 5 weeks of gestation (birth weight 3,005 g). Delivery was uneventful, Apgar score was 9 at 1st minute and 10 at 5th minute. His 28-year-old mother was healthy except for a chronic isolated neutropenia without need of therapy. Prenatal screenings were unremarkable. Initial physical examination of the baby showed the presence of two thin tissue bands connecting left upper and lower eyelids (►**Fig. 1**), impairing full eye opening. The examination of the right eye was normal. The rest of the physical examination was unremarkable, except for a Mongolian spot in the trunk. A diagnosis of left Ankyloblepharon filiforme adnatum (AFA) was made. The two tissue bands between the eyelids were then excised by the ophthalmologist on the first day of life with subsequent normal eye opening (►**Fig. 2**).

Echocardiography and cerebral echography (both normal) were performed to exclude congenital heart disease and cerebral malformations. The baby was then discharged without complications.

Discussion

Ankyloblepharon filiforme adnatum (AFA) is a rare congenital anomaly,[1,2] consisting of a partial or complete fusion of the eyelid margins. Such condition is normal during the fetal life (eyelids normally remain fused until the 5th month of gestation),[3] but not after birth. It is described as a single or multiple tissue band between the upper and lower eyelid margins. Rosenman's 1980 classification divides AFA into four subgroups (1, isolated; 2, associated with cardiac or central nervous system anomalies; 3, associated with ectodermal syndromes; 4, associated with cleft lip and/or palate).[4]

More recent findings show that AFA can indeed occur isolated as well as along with other ocular anomalies,[5] trisomy 18,[6,7] or with other multisystemic syndromes, particularly Hay-Wells syndrome,[8,9] also known as ankyloblepharon-ectodermal defects–cleft lip/palate (AEC) syndrome[10]: this condition includes a great amount of congenital malformations and anomalies (ectodermal defects, cleft-lip/palate, limb anomalies) associated with AFA.[11]

The ankyloblepharon-ectodermal defects-cleft lip/palate syndrome and other related multisystemic disorders, such as ectrodactyly, ectodermal dysplasia, and cleft lip/palate (EEC) syndrome; limb-mammary syndrome (LMS); acro-dermato-

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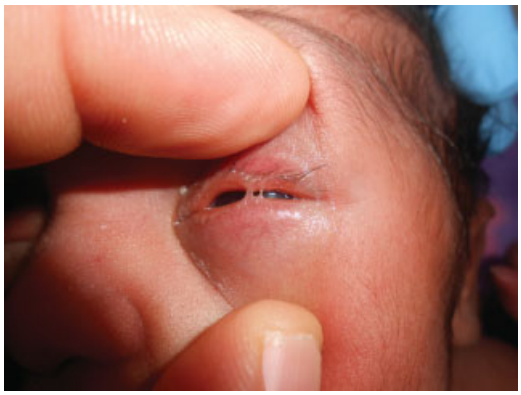

Fig. 1 Tissue bands connecting left upper and lower eyelids as seen at clinical examination at birth.

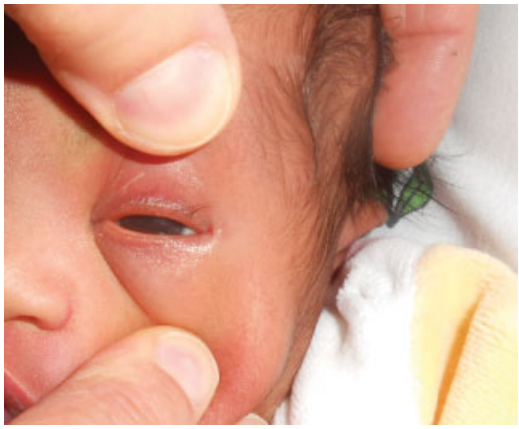

Fig. 2 Completely opened eye after tissue bands excision.

ungual-lacrimal-tooth (ADULT) syndrome; and Rapp-Hodgkin syndrome (RHS), are characterized by abnormal embryological development of ectodermally derived tissues such as skin, hair, teeth, nails, and exocrine glands.[12] All these rare multisystemic diseases share mutations in the p63 gene, a transcription factor related to tumor suppressors p53 and p73. Localized to chromosome 3q27, p63 plays a role in ectodermal, craniofacial, and limb development during fetal life.[13]

Early recognition of isolated AFA versus AEC and related syndromes is important for a correct diagnostic and therapeutic work-up and for prognostic stratification.[10]

Conclusion

Ankyloblepharon filiforme adnatum is a rare congenital condition, which can be easily missed at first clinical examination at birth due to ocular edema and eyelids swelling. Nevertheless, accurate examination of eyelids represents a fundamental part of neonatal physical evaluation before discharging the newborn, both to avoid future functional problems like impaired vision or amblyopia, and because eyelids malformations could be a sign of multisystemic disease. For this reason, in presence of an anatomic anomaly such as AFA, a thorough evaluation of other body systems (namely, jaw,[14] oral cavity,[11] and limbs[13]) to exclude associated malformations is strongly recommended.

Conflicts of Interest

None.

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