Management of an Isolated Type I Ankyloblepharon Filiforme Adnatum (AFA) with Coloboma of the Upper Eyelid in a Newborn

Abstract

Introduction: Ankyloblepharon filiforme adnatum (AFA) is a rare congenital condition marked by one or more bands of tissue adhering partially or entirely to the ciliary margins of the upper and lower eyelids at the gray line. This case study illustrates the ease of treating the condition. A general ophthalmologist could do that. Case Presentation: A one-month-old infant was admitted to the outpatient clinic. Since birth, he could not open his eyes due to upper and lower eyelid adhesions. There was also a coloboma in the one-third lateral upper eyelid in the right eye. The evaluation of other body systems was normal. The infant was diagnosed with type I AFA and underwent surgery under general anesthesia. After crushing the band with the clamp, the bands of tissue were excised with the blade. The coloboma was reconstructed by applying primary closure and pentagonal shape incision, then sutured with 8-0 vicryl. The eyelids were separated with an aligned lid margin. Conclusions: Simple surgical procedures are used to treat congenital ankyloblepharon. It depends on the nature and extent of the adhesions along the lid margin. To reduce the risk of amblyopia, early treatment should be initiated immediately.

Keywords: ankyloblepharon filiforme adnatum (AFA); coloboma of the upper lid; eyelid malformation

Introduction

The uncommon congenital disability known as ankyloblepharon filiforme adnatum (AFA) is characterized by partial or total adhesion of the ciliary edges of the upper and lower eyelids at the gray line by one or more thin bands of extensible tissue. This reduces the palpebral fissure by preventing the lids from moving.\[^1\] AFA was first described by Von Hasner in 1881.\[^2\] The incidence is 4.4 in one million births.\[^3\] The condition is typically associated with systemic malformations and is inherited autosomally dominantly with variable penetrance. Additionally, sporadic cases have been described in the medical literature.\[^4\]

Ankyloblepharon may be complete, partial or interrupted.\[^1\] The adhesions of the upper and lower eyelid margins may be congenital (present at birth) or acquired. Multiple phases are involved in the development of the eyelids, one of which is the fusion of the upper and lower eyelids. During the fifth month of pregnancy, the eyelids usually start to form, and by the seventh month, they have divided into an upper and lower lid.\[^5\] Nonetheless, if separation fails to occur, AFA is present at birth. This case study illustrates the ease of treating the condition that a general ophthalmologist could do. The patient was effectively treated by separating the eyelids.

Case presentation

A one-month-old male infant was brought to the reconstruction division of the ophthalmic department in Dr. Soetomo General Hospital, Surabaya, Indonesia by his parents after having been unable to open both eyes since birth fully. An initial physical examination of the neonate revealed the presence of connective
tissue bands linking the right and left upper and lower eyelids (Figure 1); impedes complete eye-opening. He was born unable to open his eyes due to upper and lower eyelid adhesions. He can still respond to the light, and his eyes seem to move with the subject. There was also a coloboma in the lateral one-third of the upper eyelid in the right eye. No other orbital anomalies were observed.

The results of his general physical examination were normal. The baby weighing 3.200 g, was born at 40 weeks gestation by spontaneous vaginal delivery to a 30-year-old primigravida. Prenatal screenings were unremarkable. The prenatal, intranatal, and postnatal stages were without incident. The mother denied using any medications aside from iron and mineral supplements. There was no history of exposure to X-rays. There was no history of a similar ailment in the patient’s family, nor was there any history of congenital abnormalities or consanguinity. Chest X-ray and echocardiography were performed to rule out congenital cardiac disease, with normal results. The evaluation of other body systems and laboratory findings was normal. Apart from this, he was perfectly healthy after a thorough pediatric assessment.

The infant was thoroughly evaluated for underlying syndromes, which all were normal. A diagnosis of AFA was made, and surgery was scheduled. After administering general anesthesia, his eyelids were meticulously cleansed and evaluated. The tissue band was examined by putting an iris spatula below to show no adhesions in the anterior segments. An artery clamp is then used to crush the bands to minimize bleeding. The eyelid adhesions were then liberated by a number 15 blade, and the left eye was trimmed of superfluous skin (Figure 2). The same procedure was repeated in the right eye (Figure 3).

After the band was released in the right eye, there was a coloboma in the lateral superior palpebra. The coloboma was reconstructed with a pentagonal incision and direct closure (Figure 4). Direct closure was done by suturing the tarsal plate, gray line, lash line, and skin with 8-0 vicryl. Subsequent ocular examinations following separation were done. Normal eyelid function, ocular motility, and fundus were observed.

Day one post-operation showed minimum lid edema in both eyes. The patient was given antibiotic oral and eye ointment for the wound. After a detailed ocular examination, the patient was discharged from the hospital. The patient visited the outpatient clinic one week after the surgery with no complaints or complications. Two weeks after the surgery, the patient revisited the outpatient clinic and the suture in the superior palpebra was excised. A month after the surgery, the patient eyelid wound already healed (Figure 5).

**Discussion and conclusions**

A one-month-old male infant was brought to the reconstruction division of the ophthalmic department in Dr. Soetomo General Hospital, Surabaya, Indonesia by his parents after having been unable to open both eyes since birth fully. An initial physical examination of the neonate revealed the presence of connective tissue bands linking the right and left upper and lower eyelids.

AFA is distinguished by tissue bands that unite the upper and lower eyelids.[1][4] Single or multiple bands of tissue may be evident in one or both eyes.[5] This is distinct from simple ankyloblepharon, in which the lid margins are fused directly. A central strand of vascular connective tissue is surrounded by squamous epithelium to form the tissue bands. Typically, the connective tissue is intensely cellular and embryogenic.[6]
After a detailed examination, the general physical examination of the infant was confirmed to be normal. General physical examination of the infant was confirmed to be normal. The result of an echocardiogram conducted to rule out congenital heart disease was normal. The evaluation of other body systems was normal. Apart from this, he was perfectly healthy after a thorough pediatric assessment.

AFA is also associated with multiple and complex malformations, which suggests an autosomal dominant inheritance with variable penetrance. To date, no sexual affinity has been discussed. Multiple phases are involved in the development of the eyelids, one of which is the fusion of the upper and lower eyelids. Within the fifth month of pregnancy, the eyelids typically begin to form, and by the seventh month, they begin to separate into an upper and lower lid completely. However, if there is a separation failure, it gives rise to AFA at the time of birth.

Rosenman et al. divided AFA into four subgroups. AFA type I manifests independently. Type II is associated with isolated central nervous system malformations (myelomeningocele), cardiac malformations (patent ductus arteriosus and septal defect), and gastrointestinal malformations (imperforated anus). Type III AFA is characterized by the presence of ectodermal malformation syndromes, such as pterygium-popliteal syndrome or Hay-Wells syndrome (ankyloblepharon, ectodermal defect, and cleft lip and palate). Type IV AFA is linked to labio-palatine fissures. A sixth type with the same characteristics as type I, but where there is also a family history of isolated congenital AFA transmitted by autosomal dominance, was proposed by Armes et al.

This case belongs to group one. Group one presents itself in isolation. In this instance, the prenatal, intranatal, and postnatal periods were without incident. The mother denied using any medications aside from iron and mineral supplements. There was no history of exposure to X-rays. There was no history of a similar ailment in the patient’s family, nor was there any history of congenital abnormalities or consanguinity. AFA could be a sign of multisystem disease. Therefore, thoroughly evaluating other body systems is strongly recommended to exclude associated malformations.

**Figure 3.** (A) Tissue bands showed above the iris spatula; (B) Using artery clamp to crush the bands; (C) After tissue bands excised; and (D) Excess skin was trimmed in the left eye.

**Figure 4.** (A) After the band was released in the right eye, coloboma was in the one-third upper eyelid; (B-C) The coloboma was reconstructed by applying primary closure and pentagonal shape incision; (D) After sutured with vicryl 8.0.; and (E) Both eyes after the tissue bands were excised.

**Figure 5.** (A-B) Day one post-operation showed minimal lid edema; and (C) One month post-operation.
Unknown is the pathogenesis of this anomaly and numerous hypotheses have been advanced. Many theories include a pathologic expansion of the skin of inflammatory origin, a simple separation defect, and an epithelial defect in fetal life through which connective tissue forms, most likely as a result of trauma, such as trauma to the fingernails during pregnancy. The most well-recognized explanation is a pure developmental aberration, which allows the union of lids in atypical positions because of a brief stop in epithelial growth and fast mesoderm proliferation.

AFA is usually a solitary malformation of sporadic occurrence but can occur in an autosomal dominant pattern associated with cleft lip and palate in most familial cases. Recent research demonstrates that AFA can occur both alone and in conjunction with other ocular anomalies, trisomy or other multisystemic syndromes, including Hay-Wells syndrome, also known as ankyloblepharon ectodermal defects cleft lip or palate (AEC) syndrome. A large number of congenital malformations and anomalies (ectodermal defects, cleft lip or palate, and limb anomalies) are associated with AFA.

The abnormal embryological development of ectodermally derived tissues, such as skin, hair, teeth, nails, and exocrine glands, is the hallmark of the ankyloblepharon-ectodermal defects-cleft lip or palate syndrome and other related multisystemic disorders, such as ectrodactyly, ectodermal dysplasia, and cleft lip or palate (EEC) syndrome; limbal-mammmary syndrome (LMS); acro-dermatoungual-lacrimal-tooth (ADULT) syndrome; and Rapp-Hodge-kin syndrome (RHS). All of these uncommon multisystemic illnesses are caused by mutations in the p63 gene, a transcription factor connected to the tumor suppressors p53 and p73. P63 is a gene on chromosome 3q27 that affects limb, craniofacial, and prenatal ectoderm development.

This anomaly’s cause is unknown, although several hypotheses have been proposed. According to the currently accepted theory, this condition is caused by temporary epithelial arrest and rapid mesenchymal proliferation, permitting abnormal lid union.

The infant was thoroughly evaluated for underlying syndromes, which all were normal. AFA was identified, and surgery was scheduled. After administering general anesthesia, his eyelids were extensively cleaned and evaluated. The bands are then crushed with an artery clamp to minimize hemorrhage. The left eyelid adhesions were separated promptly, as was done in this case. Generally, the cosmetic aspect of the separated bands was excellent. Complete ophthalmologic examinations and parental anxiety can be alleviated through early treatment.

AFA is a rare congenital condition easily neglected at birth due to ocular edema and swollen eyelids. Before discharging a neonate, an accurate examination of the eyelids is essential, as eyelid malformations may indicate the presence of a multisystem disease.

If not treated promptly, AFA can lead to vision impairment or amblyopia. The treatment of AFA is straightforward and consists of compressing the band with a clamp or forceps and then surgically removing it. In our case, the child was diagnosed with group I AFA, and surgery was successful. This report illustrates the ease of treatment and the significance of early intervention in preventing the development of amblyopia.

References


