ISSN No:-2456-2165

A Rare Case Series of Ankyloblepharon Filiforme Adnatum

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Abstract:- Ankyloblepharon filiforme adnatum (AFA) is a very rare congenital disease which usually present to ophthalmologists with only simply looking lid adhesion. This condition may be multisystem involving syndromic or may be isolated. We here reporting three cases of isolated and one case of systemic association of varying ages ranging from 3 days to 12 days and were well managed surgically. Updated knowledge of physicians about this condition can identify the syndromic association which can prevent complication and save life by timely intervention.

Keywords:- *Ankyloblepharon, Congenital Anomaly, Eyelid Band, Lid Surgery.*

I. INTRODUCTION

Ankyloblepharon filiforme adnatum (AFA) is a rare congenital anomaly where both eye lid are adhere partially or completely with thin bands of tissue. The difference from congenital ankyloblepharon is lid margins of both upper and lower lid are directly fused at birth due to failure of separation of both lids which usually separate during 5th months of gestation as a normal developmental process.[1] The incidence of this disease is about 4.4 in 100000 new born.[2] This condition was first described in 1881 by Josef von Hasner[3]. AFA may present as an isolated finding [4] or may be associated with multiple systemic association.[5] It may be sporadic[6] or autosomal dominant with variable expression of phenotype.[7] The systemic association may be even life threatening, that's why complete systemic examination and ancillary tests are necessary to find out the abnormalities. Here we report a series of 4 cases with surgical outcome.

➢ *Case -1*

Parents presented with a 10 days old boy with the complaints of inability to open both eyes since birth. Ocular examination revealed adhesion of both upper and lower lids with thin bands of tissue about 3-4 mm in breadth at middle portion of the both lids which did not disrupting the lash line without other abnormalities. Systemic examination done by pediatrician and no abnormality was detected. He was born by cesarean section with a weight of 3000 grams and had normal prenatal, natal and postnatal history. He was the first baby and there was no positive family history of this kind of abnormality. He was managed by simple excision of bands with topical anesthesia by holding the head firmly in sterile method at the operation theater. He was fine in subsequent follow up of 1 year.

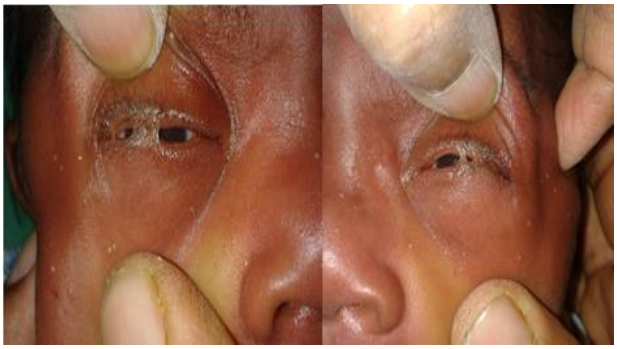

Fig 1: Bilateral AFA at middle portion of lids.

➢ *Case -2*

A 7 days old male child presented with bilateral adhesion of both lids at lateral part of palpebral fissure with thin tissue tags. Rest of ocular and systemic examination revealed no abnormality. His birth weight was 3500 grams and delivered by normal vaginal delivery with normal prenatal, natal and postnatal history. He was the third baby of his parent with no family history of same condition. His lid adhesion was separated at the operation theater by westcott scissors and there was only mild bleeding which was easily managed by thermal cauterization. He was in regular follow up to 1.5 year and there was no other complication.

Fig 2: A) Bilateral AFA at lateral part of lids. B) Little bit bleeding immediate postoperatively.

➢ *Case -3*

A 12 days baby girl presented with the complaints of inability to open her left eye since birth. She was the second baby of her parent and delivered by normal vaginal delivery with a birth weight of 2900 grams at 39th week of pregnancy. Her prenatal, natal and postnatal history was normal and there was no positive family history. Examination revealed less hair in medial part of both brow, complete closure of left upper and lower lid with intact lash line, a hyper pigmented

ISSN No:-2456-2165

area over left upper and lower lid extending up to left sided check, widely spaced eyes (hypertelorism) and small jaw (micrognathia). After surgical separation of lids, both the lid margin showed more localized hyper pigmentation about 3-3.5 mm area at central portion of lid margin. Her other ocular structure was normal. She had no cleft lip and palate and no problem in sucking milk. Cardiac examination revealed irregular heart beat and cardiologist's finding was ventricular septal defect (VSD). After that she was lost in follow up.

Fig 3: Pre (3A) and post- operative (3B) picture of uniocular AFA with periocular hyper pigmentation.

➢ *Case -4*

A 3 days old boy presented with a single band adhering the both lids of right eye. He had a birthweight of 2800 grams and borne by cesarean section. His prenatal, natal and postnatal history was normal except history of O_2 inhalation after birth. He was the first baby and his elder cousin had a similar history of eye lid adhesion. His ocular and systemic examination was normal. He was managed surgically without anesthesia at the operation theater and was fine in six months follow up.

Fig 4: Uniocular AFA with single band

Table-1: Summery of reported children with AFA

SL No	Age in days	Sex	Unilateral/ Bilateral	Number of bands	Position of bands in lid	Associated findings
1.	10	M	Bil	5-6	Centrally	No
2.	07	M	Bil	3-4	Lateral part	No
3.	12	F	Uni	Multiple	Whole length	Periocular skin hyper pigmentation, hypertelorism, micrognathia, cardiac defect.
4.	03	M	Uni	Single	Lateral part	No

II. DISCUSSION

Though AFA is benign condition the associated congenital anomaly may be multiple and complex. These type of presentation is usual and suggestive of an association with autosomal dominant. Presentation may be unilateral or bilateral and no sex predilection is found.[8]

In 1980, Rosenman et al classified AFA into four subgroups like 1.Isolated, 2. Associated with cardiac or central nervous system anomalies, 3. Associated with ectodermal syndromes, 4.Associated with cleft lip and /or palate.[5] Bacal et al also suggested another fifth group where chromosomal abnormalities is associated with AFA.[9] Literature search showed that AFA can occur isolated or with other ocular abnormalities[10] like with infantile glaucoma and iridogoniodysgenesis, trisomy 18 and different multisystem syndromes like ankyloblepharon-ectodermal defects–cleft lip/palate (AEC) syndrome or Hay-Wells syndrome; EEC syndrome where ankyloblepharon associated with other related multisystemic disorders, such as ectrodactyly, ectodermal dysplasia, and cleft lip/palate; LMS or limb-mammary syndrome; ADULT or acro-dermatoungual-lacrimal-tooth syndrome; and RHS or Rapp-Hodgkin syndrome (RHS); CHANDS syndrome which is associated with curly hair, ankyloblepharon and nail dysplasia; popliteal syndrome.[11]This condition may also associated with cardiac abnormalities such as ventricular septal defects and patent ductus arteriosus; hydrocephaly, imperforate anus [6]. All these syndromes are characterized by abnormal development of ectodermal tissue such as skin, teeth, hair, nails and exocrine glands embryonically and share p63 gene mutation which is a transcription factor related to tumor suppressor gene p53 and p73. TP 63 gene plays an important role in ectodermal, craniofacial, and limb development during fetal life[7].

In our case series among four cases, three were isolated. One case presented with periocular skin hyper pigmentation, hypertelorism, micrognathia, cardiac defect (VSD). Her physical features like AFA, VSD, micrognathia and hypertelorism were in favour of Edward syndrome (trisomy 18). But as there were no other features and may be a case of mosaic trisomy. As genetic testing is costly in our country, parent denied to do it. Literature search showed that female baby and mosaic trisomy has better survival rate than male baby who usually die before birth.[12] Literature review also

ISSN No:-2456-2165

showed cases with hypopigmented patches in skin in ACE syndrome but in this case it was periocular hyper pigmentation which may be an isolated association or may be associated with syndrome which needed further investigations. In this case series only one baby had positive family history and was isolated case of AFA.

The adhesive bands of AFA are thin extensile band with a variable length of 1 to 10 mm and breadth of 0.3 to 0.5 mm which is composed of a central vascular and highly cellular connective tissue strand surrounded by squamous epithelium lying between the cilia and orifices of the tarsal gland. Pathogenesis of this condition stated various theories like simple defect in lid separation, pathological growth of skin due to inflammation, abnormal connective tissue growth through an epithelial defect in fetal life due to trauma etc. The most accepted theory is the aberrant development of lid in fetal life due to either a temporary arrest of epithelial growth or abnormally rapid mesodermal proliferation which unite at certain points of the mesenchyme of the lid folds without epithelial interposition.[13]

The ocular risk of AFA is amblyopia due to stimulation deprivation if the adhesion remains. The management is very simple by cutting the band with or without anesthesia under sterile condition. Syndromic cases needs appropriate surgical correction and timely attention to different physical deformities in planned way.

III. CONCLUSION

Though ankyloblepharon filiforme adnatum is a rare disease, can be easily missed if meticulous examination is not done as eyelid swelling at birth may obscure that. Physician also have to be aware about the different multisystem syndromic association of simply looking eyelid adhesion to screen and manage these condition appropriately. Early surgical intervention which is very easy can prevent amblyopia also.

Conflicts of interest - The authors declared no conflicts of interest.

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