

Ankyloblepharon filiforme adnatum (AFA) associated with trisomy 18

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SUMMARY: Tüysüz B, Ilıkkın B, Vural M, Perk Y, Ankyloblepharon filiforme adnatum (AFA) associated with trisomy 18. Turk J Pediatr 2002; 44: 360-362.

Ankyloblepharon filiforme adnatum (AFA) is a rare congenital anomaly which is characterized by single or multiple strands joining the upper and lower lids. Its etiology is unknown. A girl with a gestational age of 38 weeks is presented. It was interesting to find an association of AFA with hypoplasia of hair, eyelashes, eyebrows and nails and a karyotype of trisomy 18 in this case.

Key words: ankyloblepharon filiforme adnatum, trisomy 18.

Ankyloblepharon filiforme adnatum (AFA) is a rare congenital anomaly which is represented by single or multiple strands of tissue, joining the upper and lower eyelids¹. Although this anomaly has been reported as an isolated malformation, it may also be found in association with other anomalies or as a part of a defined syndrome². AFA associated with trisomy 18 was first reported by Clark and Patterson³, and six other cases have been published^{4,5}. We present a case with AFA, whose karyotype revealed a trisomy 18 syndrome and in whom we found clinical signs of both trisomy 18 and Hay-Wells syndrome of ectodermal dysplasia.

Case Report

A girl was born by cesarean section at 38 weeks gestation to a 42-year-old gravida 7 para 6 mother as a first child from the second marriage of the mother. Two of her brothers from the first marriage had died, and their etiologies are still unknown. Her mother had four healthy children from the first marriage. The pregnancy was uncomplicated but the baby had Apgar scores of 0 at 1 minute and 6 at 5 minutes. After a thick meconium had been aspirated from the larynx she was intubated and ventilated by airbag. Birth weight was 2650 g (10th percentile), length was 45 cm (3rd percentile), and head circumference was 34 (50th percentile).

Sparse hair, absent eyebrow and eyelash, broad nasal bridge, short palpebral fissure, two narrow bands on the right and one thicker band on the

left eye joining the upper and lower eyelids (Fig. 1), low-set ears, micrognathia, hypoplastic nails (Fig. 2), hypoplasia of labia majora, erosion between anus and external genitalia, hyperpigmentation of anal and perineal regions (Fig. 3), short and dorsiflexed hallux, rocker-bottom feet (Fig. 4) and 3/6 systolic murmur were detected on physical examination.

Cranial ultrasonography showed a cystic expansion of the posterior fossa and renal ultrasonography was normal except for a minimal degree of ectasia of the right kidney. On echocardiography ventricular septal defect (VSD), atrial septal defect (ASD), and patent ductus arteriosus (PDA) were detected. Peripheral lymphocyte karyotype in 20 different metaphases showed 47, XX, +18 (Fig. 5). The infant died on day 10 after a cardiac arrest.



Fig. 1. Facial anomalies including absent eyebrows and eyelashes, low-set ears and strands of tissue joining the upper and lower eyelids.



Fig. 2. Hypoplastic nails.

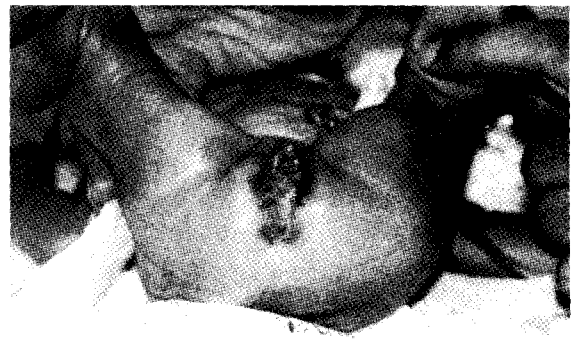


Fig. 3. Erosion between anus and external genitalia, hyperpigmentation of anal and perineal regions.



Fig. 4. Short, dorsiflexed hallux and rocker-bottom feet.

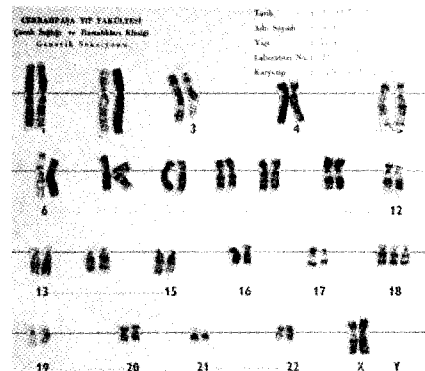


Fig. 5. Karyotype of the patient.

Discussion

Classification of AFA into four subgroups by Rosenman et al.² is as follows: Group I includes sporadic cases without any associated defects. Group II is also sporadic, but AFA is found in association with cardiac, gastrointestinal and central nervous system (CNS) defects. Group III represents cases of AFA in association with ectodermal syndromes (ankyloblepharon-ectodermal dysplasia-clefting-AEC syndrome). Group IV represents cases of AFA with cleft lip and palate defects in the patient or in the extended family. The seven previous reports describing the association with trisomy 18 suggested a fifth subgroup of AFA⁵, which is also how we classify our case. AFA may also be associated with Fraser's syndrome, Van der Woude syndrome and popliteal pterygial syndrome^{6,7}.

In our patient, association of AFA with hypoplasia of hair, eyelashes, eyebrows and nails suggested Hay-Wells syndrome of ectodermal dysplasia. Presence of perineal erosion and

hyperpigmentation in the same region supported our diagnosis. When we reviewed other syndromes in association with AFA, we found that clinical signs of our patient, such as short palpebral fissure, nail hypoplasia, severe cardiac malformations, short and dorsiflexed hallux and rocker-bottom feet had also been reported in trisomy 18, but typical cranio-facial signs (narrow bifrontal diameter, prominent occiput) and hand malformations (clenched hand, tendency for overlapping of index finger over third, and fifth finger over fourth) of trisomy 18 were absent in our patient. Ocular abnormalities reported in trisomy 18 are numerous^{8,9}. The most commonly found are short palpebral fissures, ptosis, epicanthus, mongoloid and anti-mongoloid palpebral fissure, abnormally long or sparse eyelashes and thick lids. But AFA has been described in only seven trisomy 18 syndromes.

The etiology of AFA is unknown. A number of theories have been proposed. The currently

accepted theory is that the condition is due to an interplay of temporary epithelial arrest and rapid mesenchymal proliferation, allowing union of the lids at certain points. The association of AFA with the cleft lip and palate, popliteal pterygium, ectodermal dysplasia and vaginal erosion may be explained by a delay in temporary overlapping and apoptosis during the development of mesenchymal and ectodermal tissues¹⁰. This suggests a common defect in the mechanism that regulates tissue fusion at multiple sites during development. It has also been postulated that in trisomy 18, there is an abnormal cellular proliferation and hyperplasia¹¹. Interestingly, since in our patient we found the clinical signs of both Hay-Wells syndrome of ectodermal dysplasia and trisomy 18 syndromes, and since most of the associated anomalies (congenital heart abnormality, cleft palate, nail hypoplasia) of AFA are found in trisomy 18, we thought that the common etiology of these anomalies could be related to the 18th chromosome.

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