Ankyloblepharon filiforme adnatum in trisomy 18 Edwards syndrome

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Abstract

Three cases of ankyloblepharon filiforme adnatum (AFA) in infants with Edwards syndrome are described. The case for a fifth subgroup of AFA is reinforced.

AFA is a rare congenital condition consisting of incomplete fusion of one or both eyelids by bands of elastic tissue. We describe three cases of AFA occurring in conjunction with trisomy 18. This brings the total number of cases with this association to four out of fewer than 30 published reports of AFA.

Case reports

CASE 1

A girl was born at 37 weeks' gestation by elective caesarian section for intrauterine growth retardation with unstressed deceleration on cardiotocogram. She was in poor condition at birth, requiring intubation and positive pressure ventilation for four minutes. Apgar scores were 2 at one minute and 9 at eight minutes. Birth weight was 1630 g and head circumference 30.3 cm, both well below the 3rd centile. She was noted at birth to have a 'bridge of skin' connecting her left eyelids. The baby also had features suggestive of trisomy 18, namely short sternum, overlapping fourth and fifth fingers, and absent distal interphalangeal joint creases. A systolic murmur was noted at 24 hours but this disappeared by day 10. Echocardiogram was unremarkable. Chromosome analysis showed a 47,XX,+18 karyotype.

Examination of the eyelids showed a thin elastic

strand joining the two lids across the visual axis. This was divided with minimal bleeding and all remnants had disappeared at later examination. Detailed ophthalmological examination indicated no other abnormality in either eye.

The baby was discharged from hospital after 19 days.

CASE 2

A girl (fig 1) was born at 39 weeks' gestation by forceps delivery weighing 2460 g. She was in poor condition at birth and required intubation. Apgar scores were 2 at one minute and 5 at five minutes. She continued to have respiratory problems requiring insertion of an airway. Her head circumference was 33 cm and she had many features of trisomy 18 including overlapping fingers on both hands, prominent occiput, narrow, high arched palate, choanal atresia, and bilateral short halluces. She also had a cardiac murmur and hepatosplenomegaly. In addition to this, she had small palpebral fissures and on the right at least two narrow bands attaching the upper and lower eyelids. Her condition steadily worsened and she died on day 10. Chromosomes from peripheral blood culture confirmed a diagnosis of trisomy 18.

Figure 1 Case 2: note two bands joining eyelids.

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Figure 2 Case 3: note single band connecting eyelids.

CASE 3

A macerated stillborn female (fig 2) weighing 2056 g was delivered at term to a 33 year old woman. The palpebral fissures were small and horizontal and in both eyes single bands connected the upper and lower lids. The nose was short and there was a right cleft lip and palate. The ears were small, posteriorly rotated, and crumpled. There was camptodactyly of all fingers with overlapping of the second and fifth digits. Valgus deformity of both feet was noted and the big toes were short and dorsiflexed. Necropsy showed a left diaphragmatic hernia, pulmonary stenosis, an atrial and ventricular septal defect, and horseshoe kidney. A clinical diagnosis of Edwards syndrome was made but tissue culture was unsuccessful. The parental karyotypes were normal.

Discussion

AFA was first described in 1881¹ and there has been sporadic reporting since. It has only once been described in association with trisomy 18; both eyes were affected, with one band on the right and two on the left.² Ocular manifestations of trisomy 18 are, however, well described³⁻⁶ and include short and slanting palpebral fissures, inner epicanthic folds, ptosis, sparse or abnormally long eyelashes, abnormally thick lids, and blepharophimosis. On histological examination⁷ the bands in AFA have been shown to consist of central vascular connective tissue which is highly cellular and embryonic in nature. The eyelids have been available on only two occasions,^{8,9} and in these the bands were found to be an extension of the normal connective tissue of the lids. The mechanism by which the condition occurs has been the subject of several theories over the years. It is now generally accepted that it results from (1) temporary arrest of the epithelial growth, or (2) an abnormally rapid proliferation of mesoderm, or (3) a combination of the

Classification of ankyloblepharon filiforme adnatum.

Group	Associated abnormalities
I	None
II	Cardiac or central nervous system
III	Ectodermal syndrome
IV	Cleft lip/palate
V	Chromosomal anomaly

two which allows union of unepithelialised mesenchyme at certain points. There appears to be some conflict as to which is the most common type of anomaly. Duke-Elder,⁷ reviewing the cases up to 1964, suggested that a single unilateral band was the most commonly found. This is at complete variance with the review of Clark and Patterson,² which showed only one reported case of unilateral AFA.¹⁰ However, a case of multiple unilateral bands has been reported.¹¹ Perhaps the discrepancy is the result of under-reporting of the unilateral single band since the early years. It would appear that the wider and more numerous the bands are on one side the more likely the condition is to be bilateral. This would seem to support the theory that the condition is essentially symmetrical but that owing to different interplay between the two mechanisms involved, AFA may present as just a single unilateral band.

Rosenman *et al*¹² proposed the classification of AFA into four subgroups (table), although a case has been made to include it as part of the popliteal pterygium syndrome.¹³ The fact that three of the four cases associated with trisomy 18 did not have a cleft palate would support the inclusion of trisomy 18 as a fifth subgroup, as previously suggested. We would therefore advocate chromosome studies on babies with AFA.

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