

Ankyloblepharon filiforme adnatum

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SUMMARY Two cases of ankyloblepharon filiforme adnatum in siblings are presented. One sib had associated cleft lip and palate, the other had bilateral syndactyly of the second and third toes. Father, paternal grandmother, and great-grandmother all had bilateral syndactyly of the second and third toes. Cases of ankyloblepharon filiforme adnatum reported in the ophthalmic literature are reviewed as are cases of ankyloblepharon occurring as part of the popliteal pterygium syndrome. It is postulated that the association of ankyloblepharon with other abnormalities may be part of the spectrum of the popliteal pterygium syndrome with autosomal dominant inheritance, incomplete penetrance, and variable expressivity.

Ankyloblepharon filiforme adnatum (AFA) is a rare benign congenital abnormality. A more than chance association with other congenital abnormalities and occurrence in siblings make the study of pedigrees of affected individuals of interest in trying to ascertain the heritability of the condition.

Case report

This girl was born on August 6 1977, the result of a normal full-term pregnancy. Her mother denied taking any drugs during the first trimester. At birth the baby was noted to have 'fused' eyelids as well as bilateral cleft lip and palate (Fig. 1*a, b*). Cardiac auscultation revealed a systolic murmur which disappeared by the second day and was thought to represent a closing ductus arteriosus.

More detailed ophthalmic examination showed that there were 2 asymmetrical adhesions between each pair of eyelids; these were easily parted with a strabismus hook. The manoeuvre was accompanied by minimal bleeding along the grey line of each eyelid (Fig. 2*a, b*). The vestigial remnants on the eyelid margins disappeared completely within 48 hours. The cilia and posterior surface of each eyelid were normal, as were the eyes to external examination, ophthalmoscopy, and retinoscopy. Chromosome analysis revealed a normal female karyotype.

The family history had no records of consanguinity. An older brother born on August 19 1973 had bilateral AFA as well as bilateral syndactyly of

the second and third toes. The eyelid adhesions were freed by the family physician. There were no other ocular abnormalities. The father has no ocular abnormalities but has similar bilateral syndactyly of the second and third toes. The mother is myopic but is otherwise normal. The paternal grandmother and great grandmother both had bilateral syndactyly of the second and third toes.

Discussion

Ankyloblepharon filiforme adnatum was first described by von Hasner in 1881 (Judge *et al.*, 1929). It occurs in many cases as an isolated anomaly, and the bands may be single or multiple (Duke-Elder, 1964). Associated anomalies have included cleft lip and palate (Khanna, 1957; Long and Blandford, 1962; Sood *et al.*, 1968; Ehlers and Jensen, 1970). Neuman and Shulman (1961) described a 3-day-old boy with AFA who had in addition adhesions between the alveolar processes of both upper and lower jaws, a papilla and a sinus on the left side of the lower lip, and a cleft palate. Bilateral syndactyly of the fourth and fifth toes, partial adherence of the left ear to the skin of the head, and a ventricular septal defect of the membranous type with a patent ductus arteriosus were associated abnormalities reported by Rogers (1961) in a premature baby. More recently Kazarian and Goldstein (1977) have reported a single case of AFA associated with hydrocephalus, meningomyelocele, and imperforate anus.

The histological changes of AFA have been described by many authors; the cause, however, has not been elucidated. A 'familial' tendency was invoked by Howe and Harcourt (1974), who

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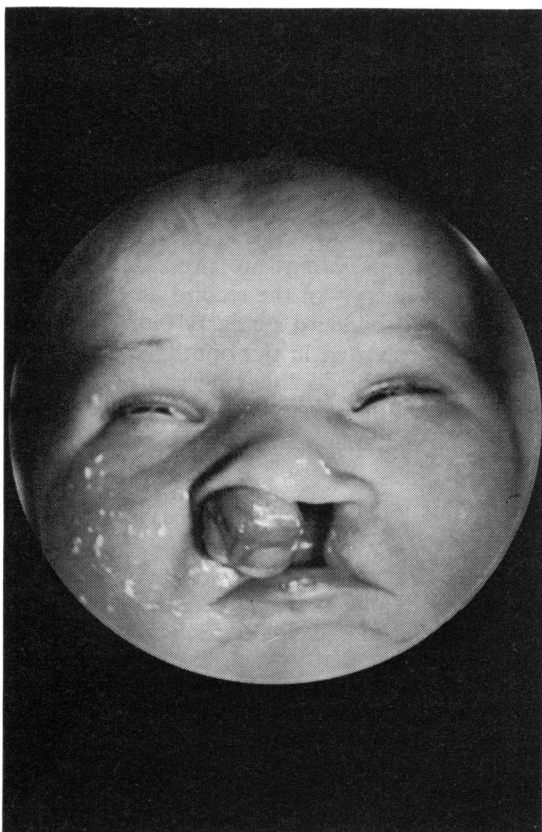


Fig. 1a *Bilateral AFA and cleft lip*

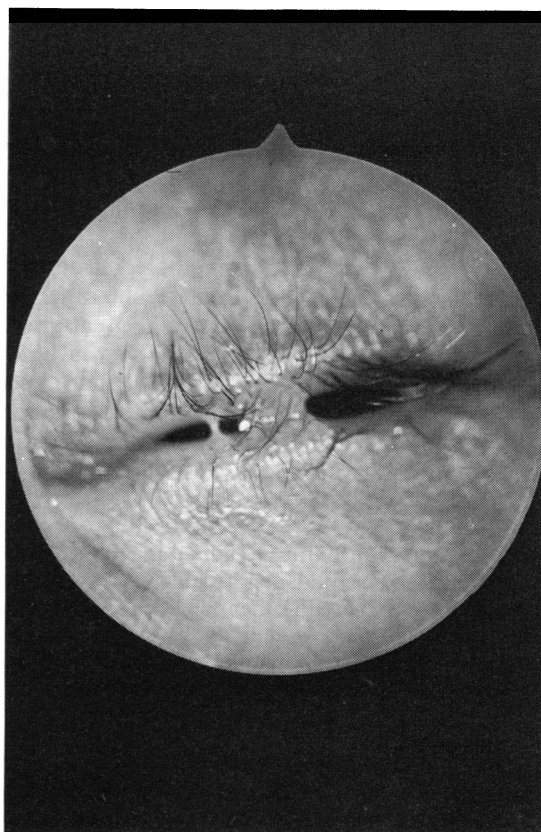


Fig. 1b *Close-up of adhesions of left eyelids*

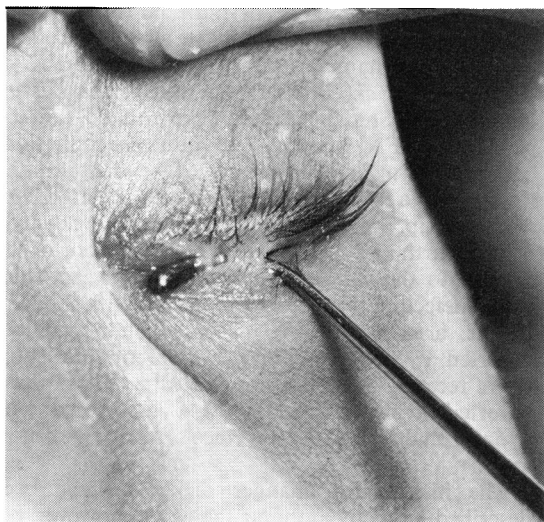


Fig. 2a *Strabismus hook in place prior to separation of left eyelid adhesions*



Fig. 2b *Left eyelids showing minimal bleeding along the grey line following separation*

described the condition in identical twins, and by Khanna (1957), who reported the condition in sisters, one of whom had cleft lip and palate. Gorlin *et al.* (1971) stated that 'the association of cleft lip or cleft palate or both and congenital filiforme fusion of the eyelids appears to be inherited as an autosomal dominant trait'.

AFA may occur as part of the popliteal pterygium syndrome. Other features of this syndrome include popliteal pterygia and oral, skeletal, digital, and genitourinary anomalies. Hecht and Jarvinen (1967) described 2 families in whom a mother and 2 of her children were affected. They noted wide variability of expression of the syndrome and postulated autosomal dominant inheritance. Gorlin *et al.* (1968) also suggested that the syndrome was inherited as an autosomal dominant trait with incomplete penetrance and variable expressivity. The same view was expressed in a recent phenotypic and genetic analysis of the popliteal pterygium syndrome by Escobar and Weaver (1978). They reviewed a number of pedigrees and stressed that the few documented familial cases showed a wide variation in gene expression in affected relatives.

We suggest that virtually all the cases of AFA reported in the ophthalmic literature as well as our own 2 cases may be part of the spectrum of the popliteal pterygium syndrome. Like other authors we suggest an autosomal dominant inheritance with incomplete penetrance and variable expressivity. This would have important implications in genetic counselling of the parents of affected offspring.

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