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Ankyloblepharon Filiforme Adnatum

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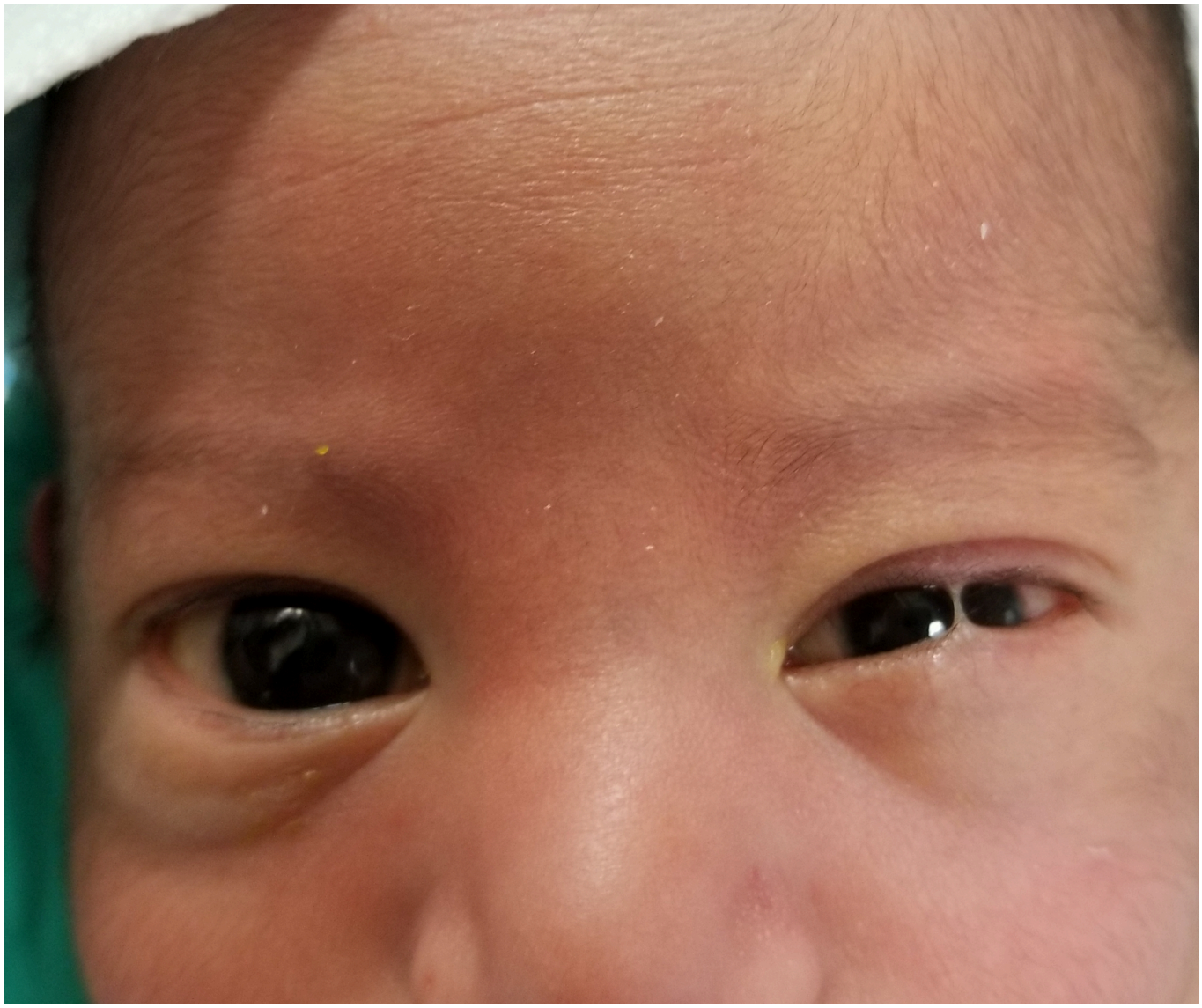
A boy was born to a 31-year-old gravida 2, para 1 woman at 40 weeks of gestation. The mother had received good prenatal care. During the pregnancy, maternal serology test results and prenatal test results were unremarkable. The maternal history was significant for hypothyroidism, which was being treated with levothyroxine. The pregnancy had been complicated by oligohydramnios.

The infant had been born via normal vaginal delivery. The Apgar score was 9/9 at 1 and 5 minutes.

Physical examination. The newborn's weight was 2610 g (5th percentile), head circumference was 31.8 cm (1st percentile), and length was 45.7 cm (2nd percentile). Physical examination findings were remarkable for a 2-mm band present in the left eye (**Figure**), completely connecting the ciliary edges of the upper and lower eyelids. A detailed physical examination did not reveal any other congenital abnormalities.



The patient received a diagnosis of ankyloblepharon filiforme adnatum (AFA).

Discussion. AFA, first described by von Hasner in 1881, is a rare congenital abnormality defined by adhesion of the superior and inferior eyelids through extensible connective tissue band(s) varying in length from 1 to 10 mm.1 The condition can lead to occlusion amblyopia.

During normal fetal development, the eyelid margins remain fused until the fifth gestational month but may take up to 7 months to become completely separated. Fusion seen at birth such as in AFA is abnormal.2

AFA is usually a sporadic congenital abnormality, but it can be associated with severe abnormalities in ectoderm-derived tissues due to mutations in the tumor protein p63 gene (*TP63*), which encodes the regulation of epidermal development. This syndrome is known as ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome, which had previously been divided into Rapp-Hodgkin syndrome and Hay-Wells ectodermal dysplasia syndrome, which are now considered to be part of the same disease spectrum.3 These encompass ankyloblepharon, ectodermal dysplasia, and cleft palate or cleft lip.3 The hallmark of AEC is the presence of

severe scalp erosions. However, erosions can be present anywhere on the skin, including palms and soles.4

Other associations are the popliteal pterygium syndrome (characterized by intercrural webbing of the lower limbs), curly hair-ankyloblepharon-nail dysplasia syndrome (CHANDS), trisomy 18 (Edwards syndrome), hydrocephalus, meningocele, imperforate anus, bilateral syndactyly, infantile glaucoma, and cardiac problems such as patent ductus arteriosus and ventricular septal defect.5,6

AFA is classified in 4 groups (**Table**), with groups I and II occurring sporadically and groups III and IV being autosomal dominant with variable expressivity.7 A fifth group also has been suggested for cases of AFA in association with chromosomal abnormalities.5

Table. Classification of AFA

Group	Associated Abnormalities
I	None
II	Cardiac or central nervous system
III	Ectodermal syndrome
IV	Cleft lip and/or palate

In cases of AFA, early ophthalmologic consult and treatment are required to minimize the risk of occlusion amblyopia. its presence also should alert the clinician to perform a thorough physical examination, since AFA can be associated with other significant disorders.

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