A rare variant of ankyloblepharon filiforme adnatum associated with skin hypopigmentation: A case report from South India

Shilpa Elizabeth Kuruvilla, Arathi Roddam Simha

We report ankyloblepharon filiforme adnatum (AFA) along with associated skin lesions in a 7-day-old child from South India. It could be a variant of the well described ankyloblepharon-ectodermal defects-cleft lip and palate syndrome also called Hay–Wells syndrome wherein AFA, skin lesions, and clefting are characteristic. The ocular features, genetic inheritance, and possible systemic associations, along with the options for management, are discussed. The need for awareness among ophthalmologists of its systemic associations is discussed to ensure that proper multidisciplinary care is offered to the individuals affected by this rare disorder. This article also highlights the unusual hypopigmented skin lesions found in this infant, which has been scantily reported in the literature, as a possible variation in patients of Indian ethnicity.

Key words: Ankyloblepharon, ankyloblepharon filiforme adnatum, hypopigmentation

Ankyloblepharon filiforme adnatum (AFA) is a rare congenital abnormality of the eyelids, wherein thin bands of tissue bridge the upper and lower eyelids. It can occur as an isolated finding or associated with other systemic abnormalities. Ankyloblepharon-ectodermal defects-cleft lip and palate (AEC) syndrome or Hay–Wells syndrome – triad of AFA, skin lesions, and orofacial clefting is a well-recognized syndrome. We report a rare variant of AFA associated with skin hypopigmentation in the absence of cleft lip/palate, and discuss the clinical implications in the management of this rare disorder.

Case Report

A 7-day-old male infant born of full-term normal vaginal delivery was referred to our Tertiary Care Eye Hospital in South India for an eye check-up. The infant was the second born of a third degree consanguineous marriage. There was no family history of anyone born with similar eye and skin problems or cleft lip/palate.

On examination, the right eye had a thin band of tissue 1 mm wide and 6 mm long, stretching from the gray line of the medial part of the upper lid and attached to the medial aspect of the lower eyelid inferiorly. The lower eyelid also

Department of Ophthalmology, Flinders Medical Centre, Adelaide, South Australia, Department of Ophthalmology, Christian Medical College, Vellore, Tamil Nadu, India

Correspondence to: Dr. Shilpa Elizabeth Kuruvilla, 6 Royal Close, Findon, SA 5023, Australia. E-mail: shilpaethomas@gmail.com

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had notches along its margin, which appeared as freshly de-epithelized lesions [Fig. 1]. The band did not restrict the eye from opening [Fig. 2]. The palpebral fissure was equally wide bilaterally. Meibomian gland orifices were present. There was no swelling over the lacrimal sac area and no regurgitation of fluid on pressure over the lacrimal sac, suggesting that there was no nasolacrimal duct obstruction. The anterior and posterior segments of the eye were well visualized and did not show any abnormalities. The left eye did not show any band joining the lids, but both upper and lower lid margins showed notching similar to the right eye [Fig. 3]. The anterior and posterior segments of the left eye were normal.

The infant had multiple patches of hypopigmentation of the skin over all the four limbs [Figs. 4 and 5]. The skin over the face, however, appeared normal. There were no skin erosions over the scalp or elsewhere. There was no cleft lip or palate. There were no structural limb abnormalities and no syndactyly. There were no obvious nail lesions. The child was feeding and thriving well.

Although an initial neonatal checkup by the referring doctor did not reveal any systemic abnormalities, the infant was referred to the pediatrician to specifically rule out cardiac, genitourinary, neurological, and odontological abnormalities and advised to review with us. A simple division of the band as an outpatient procedure was advised. The parents were not willing for the procedure immediately and wished to review back for the same. However, the patient was lost to follow-up. Division of the band and genetic testing were therefore not possible.

**Discussion**

AFA has been associated with a wide range of systemic abnormalities. These include the AEC syndrome, also called the Hay–Wells syndrome, Edwards syndrome (Trisomy 18), Chands (curly hair-ankylolobpharon-nail dysplasia), and popliteal pterygium syndrome (popliteal webbing with cleft lip/palate), cardiac abnormalities such as ventricular septal defects and patent ductus arteriosus, hydrocephaly, imperforate anus, iridogoniodysgenesis, infantile glaucoma, and ectodermal syndromes.

The AEC syndrome is a rare autosomal dominant congenital ectodermal dysplasia linked to abnormalities in the TP63 gene. The diagnosis is usually made clinically by noting features of AFA accompanied by ectodermal defects and cleft lip/palate. The absence of cleft lip/palate in this syndrome is a rare variant,
which has been reported previously. The ectodermal defects characteristically include scalp erosions (reportedly seen in 100% of patients), but also sparse, wiry hair, skin erosions, and unique pigmentary changes; nail changes, dental changes, and a decrease in sweating. Limb anomalies can occur, including syndactyly, camptodactyly, and ectrodactyly. The nail and dental changes are usually noted at a later age. The reported pigmentary changes include a hyperpigmentation in fair-skinned affected individuals and a mask-like hypopigmentation over the face noted in infants of African ethnicity.

The ocular manifestation of this syndrome carries the risk of amblyopia. The management of AFA thus involves a simple separation of the tissue bands, which can be done even without local anesthesia.

Although absent in our patient, it is important to be aware of the associated systemic complications to ensure proper management of the infant. Eroding skin lesions prone to recurrent infections are problematic which needs appropriate treatment under the supervision of dermatologists. Surgical correction of associated cleft lip/palate and other physical deformities at the appropriate time has to be planned. As reported by Mohan et al., one patient with Hay–Wells syndrome developed end-stage renal failure secondary to reflux nephropathy related to urinary tract abnormality. Cardiac and neurological abnormalities need to be identified and treated promptly. Thus, it is important to be aware of reported associated systemic abnormalities so that patients can be screened for these, followed up, and managed appropriately to improve the quality of life and prevent life-threatening complications. Identifying the pattern of inheritance and appropriate genetic counseling is necessary.

We report a rare variant of the AEC syndrome, with AFA and skin hypopigmentation in the absence of cleft lip or palate. This clinical presentation of AFA with atypical hypopigmented nonerosive skin lesions in the absence of orofacial clefting has not been widely reported or described in existing literature. Similar hypopigmented skin lesions in another Indian patient with AFA suggest that this presentation could be a unique variation of AEC in patients of Indian ethnicity, which warrants further genetic studies. It is important to be aware of this presentation of skin lesions along with AFA in patients of Indian ethnicity so that they are diagnosed and managed appropriately.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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