

Adams – Oliver Syndrome

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Abstract

Aplasia cutis congenita is characterized by congenital absence of portion of skin over a localized or widespread area. Adams-Oliver syndrome (subtype-II of ACC) is associated with distal limb reduction anomalies. We describe an infant with this uncommon disease associated with multiple midline lesions, which is a rare occurrence.

Key words: Adams-Oliver Syndrome, Aplasia cutis congenita, Hypoplastic toes

Introduction

Aplasia cutis congenita (ACC) is characterized by congenital absence of a portion of skin over a localized or widespread area. Frieden classified ACC into 9 subtypes. (Table 1) Adams-Oliver syndrome (AOS) is subtype- II of ACC which is associated with distal limb reduction anomalies. Multiple systems may be involved including central nervous system, cardiovascular system, gastrointestinal and genitourinary systems. Internal anomalies may be severe and lethal.^{1,2}We present a case of AOS with isolated terminal limb defects and aplasia cutis congenita involving the scalp and the back without any systemic involvement, which is a rare occurrence.

The Case

A five month old male child presented with bald scars over the scalp and back since birth. He was born at term by normal vaginal delivery to primigravida mother with uneventful antenatal period. Parents were non-consanguineous. There was no history of maternal drug intake, infection or radiation exposure during pregnancy. There was no family history of AOS, mental retardation or central nervous system anomalies. Physical growth and psychomotor development were within normal limits. On examination he had a smooth, round bald scar of 9 cm maximum diameter with dilated veins and a rim of hypertrichosis surrounding it over mid scalp. Similarly a linear, stellate bald scar of 21 cm maximum diameter with glistening surface, surrounded by rim of hyper pigmented skin was present over lower mid-back (Fig.1). He had hypoplasia of toes in both feet, more marked in little toes (Fig.2). Radiograph of feet showed hypoplastic phalanges in all toes with absence of distal phalanges in little toes (Fig.3). Radiograph of spine and USG cranium were normal.

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Systemic examination including funduscopy was normal.

Discussion

Aplasia cutis congenita (ACC) is a rare inherited developmental defect characterized by absence of skin in small patches. The etiology is not certain but suggested causes include ectodermal arrest during embryogenesis, compromised vasculature to the skin during embryogenesis, trauma, teratogens (methimazole), amniotic bands and genetic factors. Pulmonary hypertension, periventricular leukomalacia and retinal folds were also proposed as causative mechanisms. More recently abnormal pericyte recruitment to blood vessels was postulated as a possible etiology^{3,4,5}.

Table 1: Classification and Salient features of Aplasia Cutis Congenita¹

Type	Sites	Transmission	Associated Malformations
Type I	Scalp (usually vertex)	Autosomal dominant	Nil
Type II (Adams Oliver syndrome)	Scalp (usually midline)	Autosomal dominant/ sporadic	Limb reduction anomalies
Type III	Scalp (asymmetrical)	Sporadic	Epidermal naevi
Type IV	Any	Sporadic/multifactorial	Omphalocele, gastroschisis, encephalocele, spinal dysraphism, prosencephaly
Type V	Limbs/ trunk	Unknown	Twins with one sibling usually fetus papyraceous
Type VI	Extremities	Autosomal recessive or dominant	Pyloric /duodenal atresia, narrow palpebral fissures, nasal hypoplasia, low set ears, micrognathia, syndactyly, hypoplastic nails, overlapping of fingers, Simian crease
Type VII	Scalp/limb/trunk	Sporadic	Methimazole induced
Type VIII	Linear unilateral	Sporadic	Varicella/Herpes infection
Type IX	Scalp	Sporadic/ Autosomal	Trisomy 13, 4 p-syndrome, Johanson-Blizzard Syndrome, Delleman-Orthuys Syndrome.



Fig 1: A mid-line bald scar on back



Fig 2: Showing hypoplasia of toes in both feet



Fig 3: X-ray feet showing hypoplastic phalanges and absent distal phalanges in little toes

Adams Oliver syndrome (ACC type II) is characterized by distal limb reduction anomalies in association with the absence of skin. It is most commonly transmitted as an autosomal dominant trait with variable penetrance and expression, but autosomal recessive (AR) mode of inheritance and sporadic cases are also reported⁴. There are no differences in clinical manifestations based on the mode of inheritance. In our case, there was no family history of AOS, so it is most likely a sporadic one.

The skin lesions are characteristically solitary or multiple bald scars over the parietal area of skull, near the site of posterior fontanelle accompanied by dilated scalp veins. At birth the lesions may have already healed with scarring as an atrophic, parchment like scar with associated alopecia or may remain superficially eroded to deeply ulcerated.

The commonest limb anomaly is in the form of hypoplastic or absent distal phalanges. Other anomalies include syndactyly, brachydactyly, polydactyly and oligodactyly but some cases may have amelia of upper and lower limbs^{2,6}. AOS usually affects lower limbs more severely than the upper limbs.

Various intracranial anomalies described in these patients include encephalocele, microcephaly, cortical dysplasia, pachygyria, hypoplastic corpus callosum, parenchymal calcifications, abnormal cerebral vasculature and ventriculomegaly. Due to these abnormalities, symptoms like spastic hemiplegia, epilepsy and mental retardation are frequently found in AOS patients⁷.

Cardiovascular malformations include obstructive defects in left heart, valvular anomalies, pulmonary vascular malformation and pulmonary hypertension⁵.

Other associated anomalies reported are cutis marmorata telangiectasia congenita, gastrointestinal and hepatic malformations, genitourinary malformations, skin tags, super-numerary nipples, microphthalmia, hereditary hemorrhagic telangiectasia, cleft lip and wooly hair^{8,9}.

Apart from classical scalp lesion and limb anomalies, the present case had a stellate large bald scar over back which is an unusual occurrence. In fact, about 80% of all skin lesions reported are on the scalp. Lesions are single in about 70% of cases, double in about 20% and triple in about 5 %¹. Major complications are rare but include secondary local infection, hemorrhage, meningitis or sagittal sinus thrombosis. Our patient received symptomatic treatment and the hospital stay was uneventful.

The differential diagnoses include obstetric trauma, focal dermal hypoplasia, trisomy 13 and amniotic band sequence². Iatrogenic scalp defects following the use of forceps or scalp electrodes during labour can be mistaken for congenital absence of skin, a confusion which may have significant medico-legal repercussions. History of use of such instruments during delivery and absence of any other associated congenital anomaly may help to distinguish between the two. Focal dermal hypoplasia is characterized by linear streaks of atrophied skin, telangiectasia with soft, fatty nodules and digital malformations. Trisomy 13 is distinguished by associated characteristic findings including holoprosencephaly, microphthalmia, iris colobomas, cleft lip and/or palate and port wine stain on the forehead and rare survival beyond infancy. In amniotic band sequence, lesions may occur at any site

and are unlikely to be symmetrical.

Prognosis of the isolated or smaller skin lesion(s) is good if care is taken to prevent secondary infection and trauma using gentle cleaning, bland ointment and appropriate antibiotics if infection occurs². They tend to heal spontaneously from the margins in due course of time leaving behind a smooth, yellowish, hairless and papery scar. Larger defects may need grafting or flap rotation¹⁰.

Genetic counseling is difficult because of possible heterogeneity and broad spectrum of symptoms but should be considered if associated anomalies and family history are noted⁴. Prenatal diagnosis, as well as the assessment of severity is possible even in the first trimester¹¹.

Conclusion

Aplasia cutis congenita, so extensive, is rare. It is part of many syndromes and hence may be associated with multiple other major congenital anomalies. Hence, any baby with aplasia cutis should be evaluated for intracranial, cardiovascular, gastrointestinal, hepatic, genitourinary and limb anomalies. Moreover, as it may sometimes be misdiagnosed as an avulsion caused by vacuum application during delivery, correct diagnosis of aplasia cutis is of medico-legal significance. Hence this case is reported to highlight the importance of aplasia cutis congenita.

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