# Case Report

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# Aplasia cutis congenita

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### **ABSTRACT**

Aplasia cutis congenita (ACC) is a rare condition in which there is congenital focal absence of skin with or without absence of underlying structures such as bone. Consanguinity may play a role. The cause of this condition is unclear and appears to be multifactorial; contributory factors may include teratogens, genes, trauma and compromised blood flow to the skin. Various expressions of Adams Oliver syndrome (AOS) have also been reported which is a rare autosomal dominant congenital disorder characterized by absence of skin and or underlying structure over scalp along with transverse limb defect. It was first described by Adam and Oliver in 1945.

Keywords: Aplasia cutis congenita, Adams Oliver syndrome, Denuded areas over scalp and trunk

# INTRODUCTION

Aplasia cutis congenita is characterized by a localized or widespread, complete or partial absence or scarcity of skin at birth. The disease may be result of disrupted development or degeneration of skin in uterus.<sup>1</sup>

The impact of consanguinity on the incidence of congenital malformations was described by Mosayebi and Movahedian, who reported that the rate of congenital malformation was 2% among neonates from nonconsanguineous marriages compared to 7% from consanguineous marriages.<sup>2</sup>

ACC is most commonly found on the scalp. Other areas of involvement include trunk, extremities or both and are usually bilateral and symmetrical.

The disease has been classified in to several groups according to its pattern, area involved, underlying etiology and associated anomalies.

Adams-Oliver syndrome (AOS) is a rare developmental disorder which constitutes aplasia cutis congenita of scalp and terminal transverse limb defects like syndactyly, brachydactyly, oligodactyly and amputations. AOS is classified as type 2 ACC under Friedens classification that includes both ACC of scalp and distal limb defects.<sup>3</sup> Congenital heart defects are present in 20% of these patients and reported malformations include ventricular septal defects, anamolies of the great arteries and their valves, and tetrology of Fallot. It is an autosomal dominant trait with highly variable penetrance and expression. It was subsequently reported that some cases have autosomal recessive inheritance with severe phenotypic effects.

Snape et al, suggested criteria for diagnosis of AOS. Presence of two major criteria confirms the diagnosis. Presence of one major and one minor criteria should place AOS as differential diagnosis.

### Major criteria

- Terminal transverse limb defects.
- Aplasia cutis congenita.
- Family history of AOS.

# Minor criteria

- Cutis marmorata telangectasia congenita (CMTC)
- Congenital cardiac defect.

# **CASE REPORTS**

#### Case 1

A term male baby of weight 2.3 kg born to a P3L2 mother through consanguineous marriage of third degree was brought to the hospital with areas of denuded skin over upper limbs and lower limbs (Figures 1 A-C). History of similar skin lesions in the elder siblings who

died on 4<sup>th</sup> and 7<sup>th</sup> day of post natal life was present. History of drug intake of drug (propylthiouracil) during her pregnancy was present. On examination, denuded areas were seen over all the four limbs and intact bulla was seen over the right shoulder.

## Case 2

A term female baby of weight 2.9 kg born to P2L2 mother through non-consanguineous marriage was brought to the hospital with lesions over right thigh, scalp and limb deformities (Figure 2A and 2B). On examination, multiple small hypopigmented atrophic macules were present over the right thigh and temporal areas of the scalp. Hemorrhagic linear lesion was present over the right thigh surrounded by atrophy. Phocomelia was present with absence of right hand and absence of 2<sup>nd</sup> 3<sup>rd</sup> 4<sup>th</sup> fingers of left hand. ECHO showed mild PDA with left to right shunt with small ostium secundum atrial septal defects (OS ASD).



Figures 1 (A-C): A case of aplasia cutis with areas of denuded skin over lower limbs.



Figure 2: (A) Phocomelia with absence of right hand and absence of 2<sup>nd</sup> 3<sup>rd</sup> 4<sup>th</sup> fingers of left hand; (B) Absence of skin over right thigh.

## **DISCUSSION**

Aplasia cutis congenita (ACC) is a rare condition in which there is congenital focal absence of skin with or without absence of underlying structures such as bone. Consanguinity may play a role. The cause of this condition is unclear and appears to be multifactorial; contributory factors may include teratogens, genes, trauma and compromised blood flow to the skin, amniotic defects, and intrauterine problems. In majority of the cases ACC manifests as solitary defect of the scalp but may also involve trunk and extremities.

In our first case, mother gave history of intake of propylthiouracil during her pregnancy. Hence we postulate that repeated intake of drug by the mother have caused enough concentration of the drug in the fetal tissues over time and have possibly contributed to this condition.

At birth, the lesions can either be healed with scarring or they can be open with ulceration. After healing, the epidermis may appear flattened with a proliferation of fibroblasts within the connective-tissue stroma and absence of adnexal structures. Although the lesions are non-inflammatory and well demarcated, there is a controversy concerning treatment of ACC and there has been a great scientific interest due to extremely high mortality figures that range from 20% to 50%. High mortality/morbidity rates are due to saggital sinus bleeding, secondary local infection, meningitis, saggital sinus thrombosis, or the direct result of other serious congenital defects that are associated with ACC.

AOS is a rare congenital disorder whose pathogenesis remains obscure. The original description of Adam and Oliver suggested that there is an arrested development or agenesis of certain parts of the skeleton and the soft tissue.<sup>5</sup> Teratogenic factors, intrauterine infections, fetal exposure to cocaine, heroin, alcohol or anti thyroid drugs have all been implicated.<sup>6</sup>

Clinically AOS is characterized by presence of variable combinations of ACC of the scalp, transverse limb defects and cutis marmorata telangectasia congenita (CMTC). The ACC typically involves vertex of scalp and less commonly the parietal scalp, trunk and limbs. CMTC is a variable feature seen in 20-25% of the children and may involve the entire skin.

Congenital cardiac defects are seen approximately in 20% of AOS. These include ventricular septal defect, tetralogy of fallot, pulmonary atresia, pulmonary arterial hypertension. Other less commonly described associations include brain anamolies such as polymicrogyria, encephaly, hydrocephaly, cerebral

cortical dysplasia, or even acrania, spina bifida, spina occulta, accessory nipples and cryptorchidism.

#### **CONCLUSION**

ACC is a rare condition of uncertain etiology, but consanguinity may play a role. Various expressions of AOS have been reported. A case of Aplasia cutis congentia and Adams-Oliver syndrome are reported here without major organic abnormalities whose management depends on several factors, i.e., its pattern, location, underlying causes and associated anomalies.

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