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Case Report

Paediatrics Section

# Adams Oliver Syndrome- A Rare Entity

JYOTI RANI THAKUR[1], NANCY PHERLIN KHARKONGOR[2], JAGJIT SINGH DALAL[3]



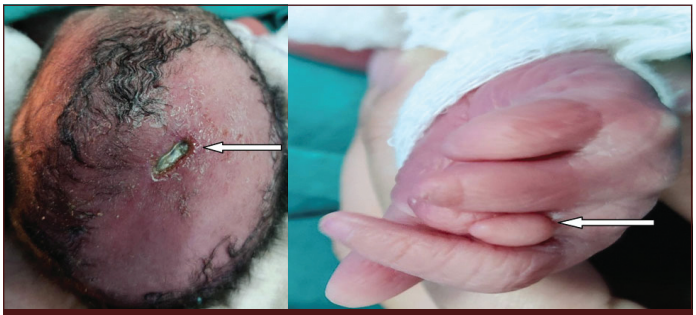

## ABSTRACT

Adams-Oliver syndrome (AOS) is a rare congenital disorder with an incidence of 1 in 225,000 individuals. It is characterised by the presence of Aplasia Cutis Congenita (ACC) of the scalp and transverse limb defects. The authors herein describes a rare case of late preterm (36-weeks) male neonate born with cranial vault defect and transverse limb defect in the form of constricted short fingers of bilateral hand with Atrial Septal Defect (ASD) diagnosed on Echocardiography (ECHO) while screening. No other associated anomalies were found. Baby was diagnosed as AOS based on these features. Patient was clinically stable with no signs of complications due to skull defect. Multidisciplinary team was involved in management and closure of skull defect was planned if not closed spontaneously in follow-up and early physical therapy or prosthetic application for the limb defects was planned as the child grows. Since the ASD was small, parents were reassured about the spontaneous closure with regular two dimensional (2D) ECHO monitoring. On follow-up at nine months of age, scalp defect closed spontaneously and child is growing well. Clinicians should be aware and all the babies born with ACC and scalp defect should be evaluated for associated anomalies as it can be a part of AOS. The early evaluation and management can be planned accordingly for better outcome.


**Keywords:** Aplasia cutis congenita, Atrial septal defect, Transverse limb defects

## CASE REPORT

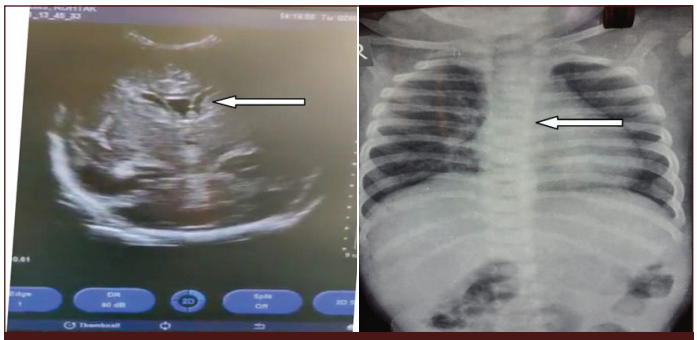
A late preterm (36 weeks) male neonate, small for gestational age, weighing 1.6 kg was born out of a non-consanguineous marriage to a third gravida severely anaemic mother by normal vaginal delivery in labour room of Obstetrics and Gynaecology Department of the hospital. Patient was born with multiple congenital anomalies i.e., aplasia cutis [Table/Fig-1] over the scalp and transverse limb anomalies in the form of constricted short finger of bilateral upper limb [Table/Fig-2]. There was no history of smoking, drinking, fever, drug intake, intrauterine infection or rashes, also no history of any medical illness such as hypothyroidism, diabetes, hypertension during antenatal period. There was no history of such anomalies in siblings and family.



**[Table/Fig-1]:** Aplasia cutis: slit like defect of top of the scalp with loss of hair with visible brain covering. **[Table/Fig-2]:** Transverse limb defect: short middle phalanx with horizontal defect consequence of amniotic band sequence. (Image from left to right)

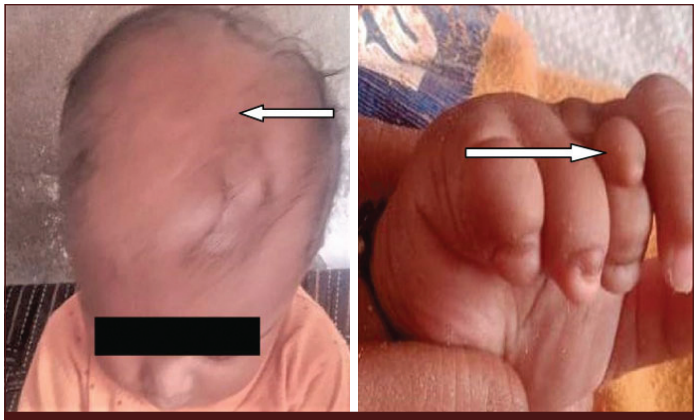
Mother was not in regular antenatal visit, however, Ultrasonography (USG) done on first and last trimester was normal. No anomaly scan was done during antenatal period.

Patient was admitted in Neonatal Intensive Care Unit (NICU) for evaluation and was clinically asymptomatic with normal vital parameters (Heart Rate-136/min, Respiratory rate-48/min) and no systemic signs of infections such as lethargy or poor feeding. Baby was started on bowl spoon feeding from day one. Complete blood count done on day second of life was normal (Total leucocyte count 11000/mm$^3$, Absolute neutrophil count 6380/mm$^3$). ECHO, ultrasound skull [Table/Fig-3] along with X-ray chest [Table/Fig-4], abdomen, and limbs were done on day third of life to look for any associated anomalies. All investigations were normal except small ASD on ECHO. Since the ASD was small, parents were reassured about the spontaneous closure with yearly 2D ECHO monitoring.



**[Table/Fig-3]:** Ultrasound skull, showing bilateral normal size ventricle with normal surrounding parenchyma. **[Table/Fig-4]:** Chest X-ray with bilateral normal lung field and normal cardiac silhouette. (Images from left to right)

Initially differential diagnosis of focal dermal hypoplasia, Scalp-Ear-Nipple (SEN) syndrome, dominant dystrophic epidermolysis bullosa was considered but all were ruled after investigations and clinical examinations and the diagnosis of AOS was made. Child was discharged on breast feeding and parents were advised for prevention of infection, trauma to skull, regular dressing, and follow-up advised with pediatrician and plastic surgeon. Parents were advised to wait for spontaneous closure of defect on follow-up, as defect was smaller. If defect does not get close spontaneously or any complications such as recurrent infection at site or meningitis occurs, surgical closure may be required. Early physical therapy and artificial prosthesis was planned for upper limb defect as child grows up. The baby was on exclusive breast feeding and weight of 2.4 kg at 1.5 month of age. At age of nine months, skull defect completely closed on follow-up [Table/Fig-5,6].



**[Table/Fig-5]:** Follow-up image of closed scalp defect: showing closed skull defect with no residual defect remaining although baldness present; **[Table/Fig-6]:** Limb anomaly on follow-up: showing normal growth of phalanx with extra abnormal tissue. (Images from left to right)

## DISCUSSION

AOS is a rare congenital disorder with features of ACC of the scalp and transverse limb defects [1]. First described by Adams FH and Oliver CP in 1945 as an autosomal dominant disorder [1]. The incidence of AOS is approximately 1 in 225,000 individuals [2]. Multiple hereditary patterns including autosomal dominant, autosomal recessive and even some sporadic cases have been reported [3]. There are three types of AOS. Type 1 have autosomal dominant inheritance due to heterozygous mutations in the ARHGAP31 gene, type 2 AOS have autosomal recessive pattern caused by mutation in DOCK6 gene and type 3 due to mutation in RBPJ gene which is inherited in an autosomal dominant manner [4].

The pathogenesis of AOS remains unclear, but various hypothesis has been suggested by different authors [1,5-7] as a causal role in its occurrence. The first description for its cause put forward by Adams FH and Oliver CP suggested that there is an agenesis or arrested growth of certain parts of the skeleton and soft tissue [1]. Later on, amniotic band sequelae and intrauterine compression were suggested as pathogenic factor [5]. Impairment in vascular development during embryogenesis as a possible mechanism was also proposed [6]. Intrauterine infections toxoplasmosis, rubella cytomegalovirus, herpes simplex, and HIV i.e. TORCH, teratogenic factors, foetal exposure to heroin, cocaine, antithyroid drugs or alcohol have been implicated in the aetiopathogenesis [7].

Clinically, the syndrome is characterised by the presence of limb and scalp defects, including transverse limb defects, ACC of the scalp and Cutis Marmorata Telangiectasia Congenita (CMTC) [8-10]. In ACC, the vertex of the scalp is usually involved and in rare cases, parietal scalp, limbs and trunk can be involved. ACC involving the scalp may involve the underlying bony structures causing bony defect which can lead to serious complications such as meningitis, encephalocoele and haemorrhage [11]. The associated transverse limb defects are usually asymmetrical and the lower limbs are more affected than the upper limbs [11]. Among the different limb defects described, brachydactyly is the most consistent feature [12]. Other limb defects such as loss of terminal phalanges, syndactyly or absence of toes and fingers of hand, toenails or fingernails agenesis have also been described but they are less common. Cardiovascular anomalies (23%), brain malformations, liver, renal and ophthalmic anomalies [13,14] are although less frequent but can also be seen [15].

AOS is diagnosed based on clinical criteria. Presence of two major criteria out of three (ACC, terminal transverse limb defect and history in family of AOS) is sufficient for diagnosis, while the combination of one major and one minor criteria (such as CMTC, congenital heart defect like ASD, Ventricular Septal Defect (VSD), Tetralogy Of Fallot (TOF) and vascular anomaly like arterial hypoplasia, bronchopulmonary haemangioma, hepatoportal sclerosis) if present there is high likelihood of AOS [11]. In present case, two out of three major criteria (ACC, terminal transverse limb defect), along with one minor criteria (ASD) were present and meet the diagnostic criteria for AOS.

Magement of AOS is only symptomatic and patient may require sugical correction of skull defect if it is large and calvarial involvement is present. Many limb anomalies that are not severe, require early physiotherapy and a multidisciplinary team of orthopaedician, plastic surgeon, rehabilitation medicine and paediatrician. Surgery or artificial limbs (prostheses) may be recommended for children who have partial or complete absence of fingers, feet, toes, upper limbs or lower legs [11]. Overall prognosis is good in AOS and likely to have a normal life span [12].

## CONCLUSION(S)

A late preterm born out of non consanguineous marriage with no significant antenatal history with congenital aplasia cutis and transverse limb defect, was diagnoised to have AOS. Clinicians should be aware of this rare entity and all babies born with ACC and scalp defect should be evaluated for associated anomalies as it can be a part of AOS. The early evaluation and management can be planned accordingly for better outcome.

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**PARTICULARS OF CONTRIBUTORS:**
1. Postgraduate, Department of Paediatrics, Pandit Bhagwat Dayal Sharma, Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.
2. Postgraduate, Department of Paediatrics, Pandit Bhagwat Dayal Sharma, Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.
3. Associate Professor, Department of Neonatology, Pandit Bhagwat Dayal Sharma, Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.

**NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:**
Jagjit Dalal,
PGIMS, Rohtak, Haryana, India.
E-mail: jagjit.dr@gmail.com

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