Adams–Oliver Syndrome – A Case Report

Dear Editor,

Adams–Oliver syndrome (AOS) initially described in 1945 by Adams and Oliver, is a rare congenital disorder.[1] Congenital scalp defects and terminal limb anomalies are the most characteristic features of this syndrome.[2-5] Life expectancy of the patient is influenced by internal organ involvement and may be lethal.[4] We describe a case of Adams–Oliver Syndrome with aplasia cutis congenita (ACC) over the scalp with digital deformities, atrial septal defect and hypoxic ischemic encephalopathy with frontal encephalocele.

An 8-day-old female child presented to us with a scalp defect since birth. The neonate had non-consanguineous parentage, and parents with no known medical problems. She was the third child with no similar complaints in the siblings. Mother's antenatal history was uneventful with the baby born via normal delivery at 32 weeks with a birth weight of 1.96 kg. Dermatological examination revealed an 8 × 9 cm diameter defect in layers of scalp, appendages and subcutaneous tissue in vertex of scalp extending from frontal hairline to parieto-occipital areas of scalp. Lesions are associated with seropurulent discharge, crusts and erosions [Figure 1a and b]. Syndactyly was present over 2nd and 3rd toes of both feet [Figure 2a]. There were absent and few dystrophic nails over toes and fingers [Figure 2b]. Clinical diagnosis of aplasia cutis congenita was made and further investigated to rule out syndromic association. Magnetic Resonance Imaging (MRI) of the brain revealed hypoxic ischemic encephalopathy changes in bilateral parieto-occipital lobes with left frontal encephalocele [Figure 3]. 2D echocardiography showed atrial septal defect. Ultrasonography of the abdomen did not reveal any abnormalities. Neonate was started on intravenous antibiotics, analgesics, calcium gluconate and vitamin K. Daily paraffin dressings were done for the wound. Opinion of the Pediatric Surgery department was sought for but due to low weight, surgical defect closure could be not be done until the child gained weight. Scalp biopsy could not be done on account of the infected nature of the lesions and the critical situation of the child. Neonate was managed with IV antibiotics and daily dressings. One week later, she developed convulsions and was shifted to the Neonatal Intensive Care unit. Inspite of anticonvulsant therapy, child developed multiple convulsion episodes and succumbed to death.

AOS (MIM 100300) is a rare disorder with 1 in 225,000 as an estimated incidence.[1] Autosomal dominance is the main inheritance pattern but, autosomal recessive and sporadic cases have also been reported with similar clinical presentations.[4] AOS is divided into three types phenotypically.[5] Autosomal dominant form with variable expression is type 1 caused by heterozygous mutations in the ARHGAP31 gene, a Cdc42/Rac1 GTPase regulator. Type 2 AOS is an autosomal recessive form, which can be caused by compound heterozygous or loss-of-function homozygous mutations in the DOCK6 gene, an atypical guanidine exchange factor known to activate Cdc42 and Rac1. Type 3 AOS is an autosomal dominant form, which can be caused by heterozygous mutations in the RBPJ gene, a primary transcriptional regulator for the Notch signaling pathway.[5] Several authors propose vascular impairment during embryogenesis as a possible mechanism. Terminal transverse limb defects, aplasia cutis congenita, and family history of AOS form the major criteria for diagnosis. Minor criteria include cutis marmorata, vascular anomaly as well as congenital heart defects. AOS has the most frequent clinical manifestation as limb anomaly which is commonly asymmetric and presents in different forms such as brachydactyly, oligodactyly, syndactyly, polydactyly, and hypoplastic nails.[3] The second most common presentation is scalp defect, generally in vertex area with or without underlying skull defect.[2,5] Our case presented with limb anomalies as well as scalp and skull defects with CVS involvement in the form of atrial septal defect.

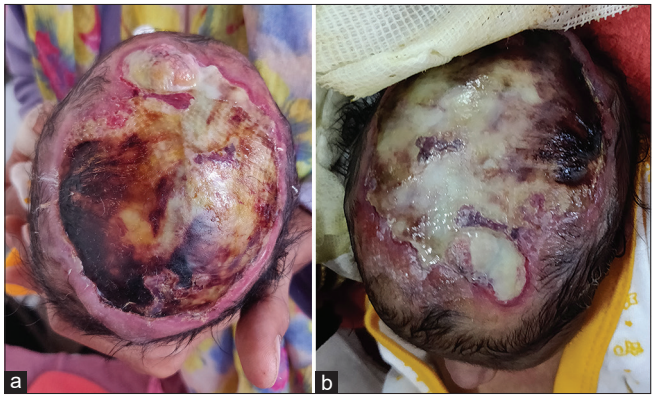

Figure 1: (a) 8x9 cm defect in layers of scalp. (b) Lesions are associated with pus and serous discharge, crusts and erosions

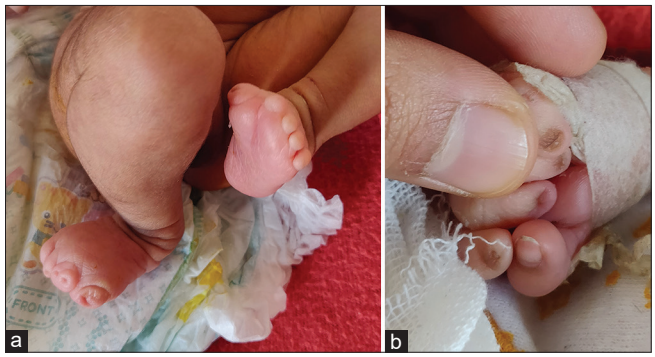

Figure 2: (a) Syndactyly was present over 2nd and 3rd toes of both feet and absent and few dystrophic nails over toes. (b) Absent and few dystrophic nails over fingers

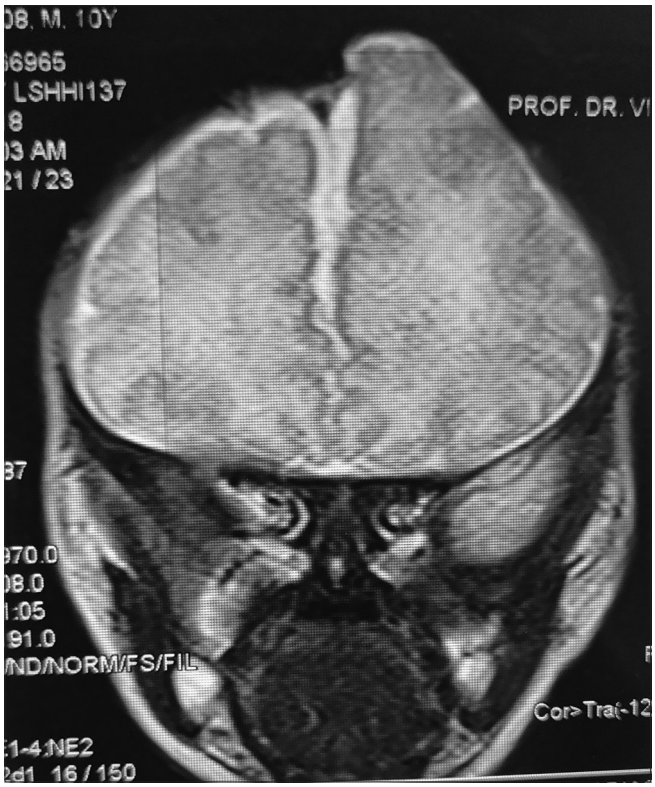

Figure 3: MRI of the brain revealed hypoxic ischaemic encephalopathy changes in bilateral parieto-occipital lobes with left frontal encephalocele

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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