OLGU SUNUMU CASE REPORT

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Adams Oliver Syndrome in a Newborn: Case Report

Yenidoğanda Adams Oliver Sendromu

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ABSTRACT Adams Oliver syndrome (AOS) is characterized by a combination of congenital scalp defects (aplasia cutis congenita) and terminal transverse limb defects. Various expressions of AOS have been reported. It can also be associated with extensive lethal anomalies of central nervous, cardiopulmonary, gastrointestinal, and genitourinary systems. Here we report a newborn diagnosed AOS with severe scalp defect, terminal limb defects and cutis marmorata telangiectatica congenita without systemic involvement. During hospitalization menengitis developed as a major complication and treated with appropriate antibiotheraphy. Scalp defect was treated with local wound care. In conclusion it is important to be aware of different presentations of AOS. All newborn infants with aplasia cutis congenita should be evaluated carefully with physical examination and laboratory workup to rule out associated diseases.

Keywords: Adams Oliver syndrome; abnormalities

ÖZET Adams Oliver Sendromu (AOS) doğumsal saçlı deri defekti (aplazi kutis konjenita) ve çeşitli derecelerde (terminal transvers) ekstremite malformasyonları ile karakterize bir sendromdur. Hastalık, letal hemorajik kraniyal defektler, merkezi sinir sistemi, kardiyopulmoner, gastrointestinal ve genitoüriner sistem ve/veya şiddetli ekstremite malformasyonlarından, bulguların hafif olduğu vakalara kadar çok değişik klinik bulgular verebilir. Bu makalede kutis marmorata telenjiektatika konjenita ile birlikte ağır saçlı deri, ekstremite tutulumu olan yenidoğan olgu sunulmuştur. Sistemik tutulumu olmayan hastada hastane yatışı sırasında major komplikasyon olarak menenjit gelişmiş, bu antibiyotikle tedavi edilirken, skalp defekti ise lokal yara bakımı ile iyileşmiştir. Sonuç olarak, farklı prezentasyonla gelebilecek AOS vakaları konusunda duyarlı olunmalı, aplazi kutis konjenita'lı tüm yenidoğanlar fiziksel ve laboratuvar açısından dikkatle değerlendirilmelidir. Eşlik eden anomaliler gözden kaçırılmamalıdır.

Anahtar Kelimeler: Adams Oliver sendromu; anormallikler

Aplasia cutis congenita (ACC) means congenital absence of skin at birth. It occurs most commonly on the scalp but can also affect any part of the body.[1] Frieden defined a classification of nine different groups of ACC based on the location and presence of other abnormalities in 1986.[2] According to this classification group 2 is defined as scalp ACC with associated limb abnormalities-Adams-Oliver syndrome.[1] Adams-Oliver syndrome (AOS) is characterized by a combination of congenital scalp defects (ACC), terminal transverse limb defects and often cutis marmorata telangiectatica congenita (CMTC).[3] The expression of scalp defects sometimes includes bone deformities, and limb defects can vary from nail dystrophy to complete absence of distal extremeties.[4] AOS can also be associated with extensive lethal anomalies in the central nervous, cardiopulmonary, gastrointestinal and genitourinary systems.[3,5]

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Here in we report a newborn diagnosed as AOS with ACC on the scalp, terminal limb defects and cutis marmorata telangiectatica congenita.

CASE REPORT

A male infant was born by vaginal delivery at 38 weeks of gestation to a 28-year-old mother, gravida 2, para 2. Birth weight was 3100 g (10-50 P), occipitofrontal head circumference (OFC) 34 cm (50 P) and height was 47cm (10-50 P). Apgar scores were 8 and 9 at 1 and 5 min respectively and no resuscitation was required. Parents were third degree relatives and they had a healthy, seven years old boy. The pregnancy was uncomplicated. There was no history of maternal medications. There was no maternal or other family history of skin, connective tissue or autoimmune disease.

On physical examination the infant had a scalp defect extending from anterior fontanel to occipital region with dilated scalp veins (Figure 1). Brachydactyly was noted on all fingers. Terminal phalanges of second, third, and fourth fingers and nails on all fingers were hypoplasic on the right foot (Figure 2). Generalized cutis marmorata was noted.

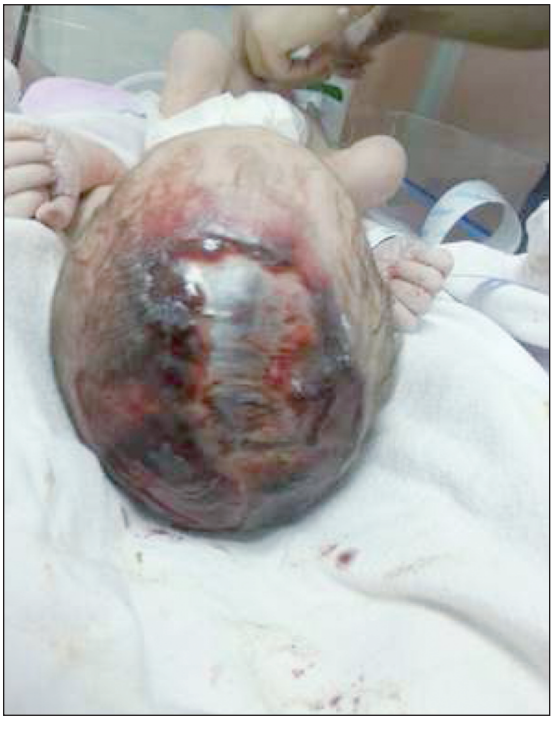

FIGURE 1: Severe aplasia cutis congenita with scalp defect.

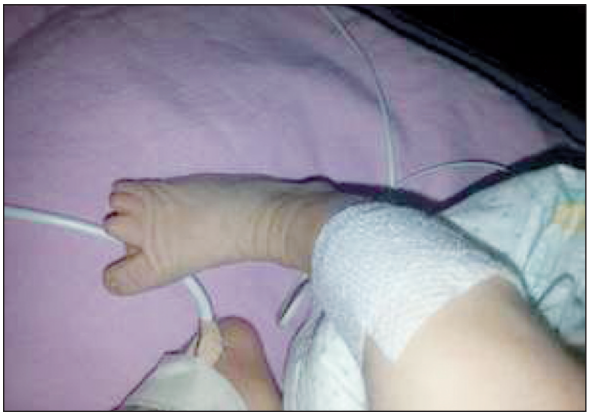

FIGURE 2: Brachydactyly and atrophic nails on toes.

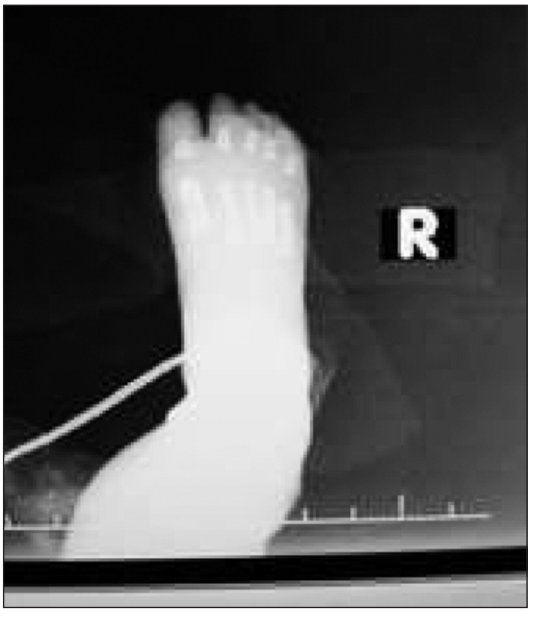

FIGURE 3: X-ray shows hypoplasia of terminal phalanges.

Laboratory tests upon admission included a complete blood count (CBC), C-reactive protein (CRP) and blood cultures. Complete blood count and CRP level were normal. Initial blood culture showed no growth at 48 hours. Serologic tests for TORCH were negative. The skull x-ray was normal. The chest roentgenogram, abdominal and cranial ultrasonography and echocardiography showed normal findings. Magnetic resonance imaging of the brain revealed no structural abnormality. X-ray of the right foot showed hypoplasia and complete absence of terminal phalanges (Figure 3). There was no additional muscu-

loskeletal anomaly. Ophthalmological and audiometric examinations were normal. Karyotype analysis revealed 46, XY, but we couldn't perform extended genetic analysis. Histopathological examination of the scalp lesion revealed flattening of rete ridges, thin dermal collagen in the dermis, and the loss of appendages that was compatable with ACC. The combination of ACC, brachydactyly and terminal dystrophy of the toes and nails led to the diagnosis of Adams-Oliver Syndrome.

Patient was consulted with dermatology and plastic surgery consultants. They recommended to allow the area to heal spontaneously by using conservative wound care such as topical bacitracin and petrolatum two to three times a day. Skin graft was also recommended at next mounths because of large skin defect.

On the 3rd day of hospitalization he became febril, lethargic and hypoactive. CBC, CRP, peripheral blood smear and blood culture were obtained. CBC revealed white blood cell count of $12x10^3$/mcL ($12x10^9$/L), with 68% neutrophils, 12% bands, 4% monocytes, and 16% lymphocytes, CRP level of 15 mg/dL (N:<0.5). Lumbar puncture revealed turbid cerebrospinal fluid (CSF) with 350 cells (90% polymorphs, 10% lymphocytes). CSF culture showed growth of *Staphylococcus epidermidis.* Vancomycin and gentamicin were administered for 2 weeks. His wound began to heal by conservative therapy. He was fed with human milk and began to gain weight. The infant was discharged 3 weeks later to continue local wound care by his mother. He was seen twice a week for the evaluation of the wound healing for 2 months then followed up by plastic surgery department. Informed consent was obtained from the parents.

DISCUSSION

Adams-Oliver syndrome was initially described in 1945 by Adams and Oliver. It is defined by the combination of limb abnormalities and scalp defects, often accompanied by skull ossification defects.[1] AOS is thought to have an autosomal dominant mode of inheritance with variable expression, less commonly sporadic and autosomal recessive cases have been defined.[6] The present case had no family history of congenital deformities of the scalp and extremities.

Approximately 75% of the patients with AOS have ACC of the scalp.[1] Scalp lesions are most frequently found on the vertex of the skull that are variable in depth and in size.[1,6] Scalp defects can be mild or severe and often they are found underlying the scalp lesions. Ultrasound is helpful in evaluating the extent of skull involvement. Our patient had a severe scalp lesion but cranial ultrasound revealed no structural abnormality.

Central nervous system (CNS) involvement like microcephaly, polymicrogyria, hydrocephaly, cerebral calcification was described in patients with AOS.[5,7] Spasticity, epilepsy, motor and mental retardation are described as neurological abnormalities in these patients.[7-9] Our case had normal cranial magnetic resonance (MR) findings and his neurological examination was also normal.

The limb defects in patients are usually asymmetric and lower limbs are more commonly involved than the upper limbs. The most common limb defect in AOS is brachydactyly and limb defects can be subtle such as an absent nail or broad fingertip.[4,9] Our patient had asymmetric involvement at his right foot and brachydactyly and hypoplasic nail were observed at his toes.

Cutis marmorata telangiectatica congenita is a congenital capillary and venous vascular malformation. It has been specifically described in relation to Adams-Oliver syndrome.[10] In this syndrome CMTC is generalized and may be found in approximately 25% of patients.[11,12] Mild form of CMTC was present in our case.

Cardiopulmonary, gastrointestinal and genitourinary system involvement was also described in AOS, so evaluation of these systems should be performed.[1] Our patient didn't exhibit any involvement of cardiopulmonary, gastrointestinal and genitourinary system as proved by abdominal and pelvic ultrasonography and echocardiography.

Vascular impairment in utero has been proposed as a possible mechanism but the exact pathogenesis is unknown. The hypothesis that, impaired circulation in watershed areas during development can explain the cranial vertex and limb abnormalities.[1] A defect in angiogenesis is an another proposed mechanism.[13]

Several genes implicated in skull and limb development have been identified but no causative gene has been found in AOS.[1] Prenatal diagnosis has previously been reported a consanguineous Turkish couple.[14] Also, mutations in a recombination signal binding protein for immunoglobulin kappa J were identified through exome resequencing causing alterations in the signalling of the Notch pathway.[15] Genetic analysis was not performed at our patient.

Treatment of ACC of the scalp consists of allowing the area to heal spontaneously using conservative wound care. Larger defects may require a bone or skin graft.[16] Local wound care was reported in patients with AOS.[17] We also used conservative wound care, topical bacitrasin in our patient. At discharge his wound began to heal with forming a scab. He is also following by plastic surgery and skin graft is beeing considered by surgeons.

Consultation with an orthopedic surgeon may be helpful in more severe cases. Our patient had mild limb abnormality, so this will not probably cause skeletal disability and reduce the quality of his life.

Close follow-up for serious complications, such as hemorrhage, infection, sagittal sinus thrombosis are important in large scalp defects.[1] During hospitalization, menengitis developed as a major complication in our patient. He recovered by appropriate antibiotheraphy.

It is important to be aware of different presentations of AOS. All newborn infants with ACC should be evaluated carefully with physical examination and laboratory workup to rule out associated diseases.

Conflict of Interest

Authors declared no conflict of interest or financial support.

Authorship Contributions

Design and writing of the case with literature review: *Sumru Kavurt,* ***Literature review and revision of manuscript:*** *Fatma İyigün,* ***Literature review and revision of manuscript:*** *İstemi Han Çelik,* ***Revision of the manuscript:*** *Ahmet Yağmur Baş,* ***Revision of the manuscript:*** *Nihal Demiral.*

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