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Case Report

Aplasia Cutis Congenita Associated with Multiple Congenital Anomalies: Case Report

Multipl Konjenital Anomaliyle İlişkili Aplazia Kutis Konjenita: Olgu Sunumu

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ABSTRACT

INTRODUCTION: Aplasia cutis congenita (ACC), is a rare anomaly presenting with a solitary scalp lesion of skin abrasion. Lesions can be multiple and on different surfaces of the body but are mostly seen on the scalp (%70) as a solitary lesion. As it is a rare disease, we aimed to describe our case with ACC.

CASE REPORT: Our case was a newborn infant with a large full thickness skin and skull defect on the scalp at birth. He also had hypoplasia on the fingers, omphalocele, ectopic anus, cleft lip and palate anomalies and multiple cardiac anomalies. We planned further evaluation but he died 23 hours after birth because of cardiac problems.

DISCUSSION: Multiple factors have been described as etiological factors for ACC but the etiology is not clear yet. ACC may also be associated with some syndromes. Management strategies are based on the size, presence of an underlying skull defect and the child's physical condition.

KEYWORDS: Aplasia cutis congenita, Skin anomalies, Neonatal skin disorders, Scalp lesions

ÖZ

GİRİŞ: ACC skalpte soliter lezyonun cilt abrazyonuyla prezente olan nadir bir anomalidir. Lezyon multipl ve vücudun farklı yüzeylerinde olabilir fakat çoğunlukla skalpte soliter bir lezyon olarak görülür (%70). Biz, nadir bir hastalık olması nedeniyle ACC'li vakamızı sunmayı amaçladık.

OLGU SUNUMU: Vakamız, doğum sırasında skalpte geniş bir tam kat cilt ve kafatası defekti olan yenidoğan infant idi. Aynı zamanda el parmaklarında hipoplazi, omfalosel, ektopik anüs, yarık damak ve dudak ve multipl kardiak anomalilere sahipti. Biz değerlendirmeyi planladık fakat kardiyak problemler nedeniyle doğumdan 23 saat sonra öldü.

TARTIŞMA: ACC için birçok etken etyolojik faktör olarak tanımlanmıştır fakat henüz etiyoloji tam olarak açık değildir. ACC aynı zamanda bazı sendromlarla ilişkili olabilmektedir. Hastalığa yaklaşım stratejilerinin temelinde büyüklük, altında kafatası defekti varlığı ve çocuğun fizik kondisyonu bulunmaktadır.

ANAHTAR SÖZCÜKLER: Aplazia kutis konjenita, Cilt anomalileri, Yenidoğan cilt hastalıkları, Skalp lezyonları

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INTRODUCTION

Aplasia cutis congenita (ACC) is a rare congenital absence of skin. It was first described as a lesion on an extremity by Cordon in 1767 and as a lesion on the scalp by Campbell in 1826 (6, 9). ACC can be familial or a part of a syndrome and the incidence is 1-3/2000-10000. More than 500 cases have been reported since (6) and 25% of the reported cases are familial with a vast majority (69%) showing an autosomal dominant inheritance. There is no sex predilection. Frieden classified ACC based on etiology and manifestation into nine groups and 86% of reported cases are known to be group 1. Lesions can be multiple and on different surfaces of the body but mostly seen on the scalp (70%) as a solitary lesion (6, 9). It may often occur on the parietal and occipital areas and rarely on extremities. It can also be associated with other physical anomalies such as defects of eyes, extremities, limbs, gastrointestinal system, genito-urinary system and central nervous system. Although most lesions are small, superficial, noninflammatory, and well demarcated, then can rarely be seen as large ulcers including the bone and meningial defects (2, 5). Approximately 20% of all cases have a defect (6) and Billard initially reported such a lesion in 1828 (9).

CASE REPORT

Our case was a 36 weeks gestational age newborn infant with a large full thickness skin and skull defect on the scalp at birth. There was no history of obstetrical trauma. His lesion was grey-black coloured, non-hemorrhagic, non-secretory and 5x4 cm in size (Figure 1A). There was no cerebrospinal fluid leakage. He also had hypoplasia of his fingers (Figure 1E), omphalocele, bladder anomalies (Figure 1D), ectopic anus (Figure 1C) and cleft lip and palate anomalies (Figure 1B) on physical examination. He was noted as group 4 according to Frieden Classification. We performed echocardiography (ECO) as the baby had respiratory and cardiac problems and detected atrial septal defect (ASD), ventricular septal defect (VSD), and transpositions of great arteries (TGA). Cranial computerized tomography (CT), cranial magnetic resonance (MR) and genetic evaluation were planned because of the full thickness defect with bone defect but he died 23 hours after birth because of cardiac problems.

Figure 1: ***A****. (Scalp defect including bone defect on vertex).* ***B****. (Cleft lip and palate anomalies).* ***C.*** *(Ectopic anus).* ***D****. (Omphalocele and bladder anomalies).* ***E.*** *(Finger hypoplasia).*

DISCUSSION

Genetic causes, syndromes and teratogens, intrauterine infection with the varicella zoster virus or herpes simplex virus, fetal exposure to cocaine, heroin, alcohol or anti-thyroid drugs, infarction of the placenta, and amniotic pathologies are some of the factors that have been described as etiological factors for ACC, but the certain etiology is not clear yet (1,3,5,6,10). There are lots of cases without any etiological factor in the literature (5,6). ACC may also be associated with the Adams-Oliver Syndrome, Barts Syndrome, Trisomy 13 (Patau's Syndrome), Ellis-van Creveld Syndrome, Wolf-Hirschhorn syndrome, and the Johanson-Blizzard syndrome. Cardiac pathologies are mostly seen in the Adams-Oliver and Ellis-van Cleverd Syndromes (7). The syndrome consisting of ACC, a mild blistering disorder of the hands and feet, oral mucosal lesions, and nail dystrophy is known as Bart's Syndrome (2). The association of ACC and Bart's Syndrome was defined in 1966 (2). In addition, a few cases have been reported with split cord malformation (SCM) and ACC (4). In our case, the mother was previously healthy and there was no drug intake history which could be considered as teratogenic. There was no evidence for an amniotic band in pregnancy. There was also no significant family history. Frieden classified ACC into nine groups according to the number and location of the lesions and the presence or absence of associated malformations (2) (Table I).

Table I: Frieden Classification.

1	Scalp ACC without multiple anomalies
2	Scalp ACC with associated limb anomalies
3	Scalp ACC with associated epidermal and organoid nevi
4	ACC overlying embryologic malformation
5	ACC with associated fetus papyraceous or placental infarct
6	ACC associated with epidermolysis bullosa
7	ACC localized to extremities without blistering
8	ACC caused by teratogens
9	ACC associated with malformation syndromes

ACC lesions may commonly occur on the parietal and occipital areas and they are often small and superficial. However, the lesion in our case was a large, full thickness defect including bone. ACC can be associated with other physical anomalies such as defects of eyes, extremities, limbs, gastrointestinal system, genito-urinary system and central nervous system so that skull and extremity radiographs, chromosome analysis, cerebral and abdominal sonography, echocardiography must be performed to look for concomitant anomalies. Similarly, we determined other pathologies accompanying the scalp lesion and we also planned further diagnostic evaluations but we could not because of the loss of the patient due to cardiac pathologies during follow-up.

Management strategies are based on the size, presence of an underlying skull defect and the child's physical condition. Small and superficial lesions may heal spontaneously. Large lesions with a bone defect require surgical closure to prevent massive hemorrhage, infection and thrombosis that might be fatal. Although surgical treatment is known to be adequate treatment for these lesions, conservative treatment has been proposed in some studies. Large lesions cause death secondary to infection or hemorrhage because of exposure of the brain and sagittal sinus. Nearly 22 cases of ACC with agenesis of the scalp and skull had been reported until 1956 and 59 % died from causes related to the lesion. With the development of new treatments since 1956, the rate of mortality is gradually decreasing from the previous figure of 22-55% (8). Early surgery has been reported to prevent vital risks, reduce local complications and make the final reconstruction easier. We covered the lesion with antibiotics but could not provide other treatment. More cases will have to be reported and studied in order to understand the etiology and management of this entity.

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