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Aplasia cutis congenita and limb anomaly: A case of non-scalp lesion

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SUMMARY

Aplasia cutis congenita is a rarely seen disease in which dermis, subdermal tissue and extremely rarely bone are absent. Significant number of cases are lost due to infections, electrolyte imbalance, and massive hemorrhage. In this report, we present a 2-day-old infant who had a wide aplasia cutis area on the right lower limb.

Key words: *Aplasia cutis congenita, limb anomaly*

ÖZET

Aplazia kutis kongenita ve ekstremite anomalisi: Non-skalp lezyonlu bir olgu

Aplazia kutis kongenita ender görülen deri tabakalarının, subkutan dokunun daha ender olarak da kemiğin olmadığı bir hastalıktır. Olguların önemli bir kısmı infeksiyonlar, elektrolit kaybı ve masif hemoraji nedeniyle kaybedilir. Biz bu çalışmada sağ bacağında geniş deri aplazisi alanına sahip 2 günlük bir yenidoğanı sunduk.

Anahtar kelimeler: *Aplazia kutis kongenita, ekstremite anomalisi*

Aplasia cutis congenita (ACC) is a rare congenital disorder characterized by a localized absence of skin, dermal appendages, and in some cases, the subcutaneous tissues. It was first described by Cordon in 1767 [1,2]. ACC may occur anywhere in the body; however, in 84 % of the cases, the defect is found in the scalp [3], where it is often solitary and located predominately in the midline vertex. Non-scalp lesions may involve the trunk and/or extremities and are usually bilaterally symmetric [4].

ACC was reported to affect 1 in every 10.000 live births [5]. Many theories have been postulated to explain the occurrence of ACC; however, neither the pathogenesis nor the aetiology has been clarified yet. Factors like intrauterine trauma, amniotic bands, and some drugs have been implicated [5,6]. Majority of the published cases of ACC are sporadic; with a few reports describing a familial occurrence in the form of autosomal dominant [7], as well as autosomal recessive [8] pattern of inheritance.

In this article, we present a rare case of ACC in a patient who had a wide aplasia cutis area on the right lower limb.

CASE

A full term 2-day-old girl was referred to our hospital with a subdermal tissue , skin, and full thickness dermal defect on the right lower limb. The child's birth weight was 2410 g, head circumference was 31 cm and length 41 cm. Nonconsanguineous father, and mother were 25 and the 23 years old, respectively.

On physical examination, the patient had pes equinovarus, and a widespread skin defect on the right lower limb (Figure 1). In addition, she had microcephalia, hirsutism on the forehead region,

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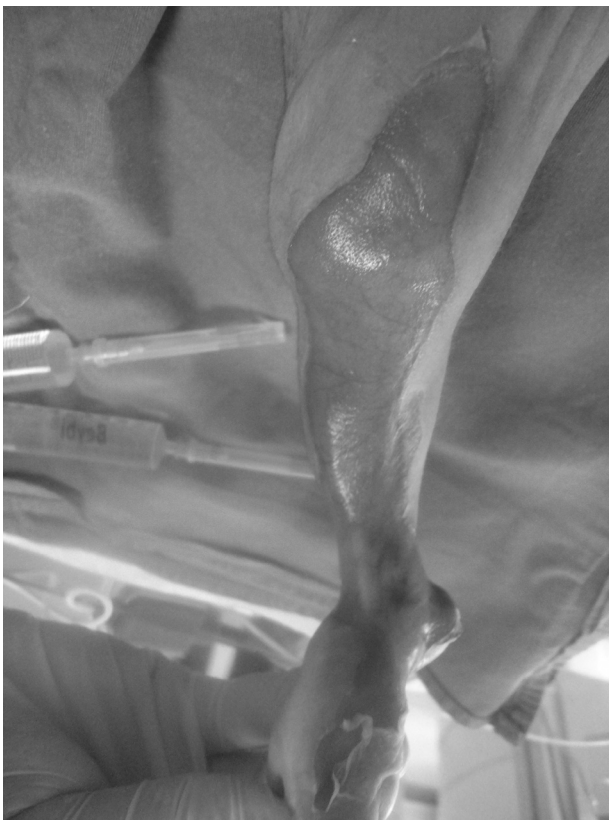
synophrys, arched eyebrow, swollen eyelid, wide nose and a thick inferior lip (Figure 2). Radiological examination showed right distal tibia lying on the left side (Figure 3). Laboratory examinations and karyotype analysis were normal. Ultrasonographic, echocardiographic examinations, and family history were unremarkable.



Figures 1. Appearance of the right lower limb of the patient.

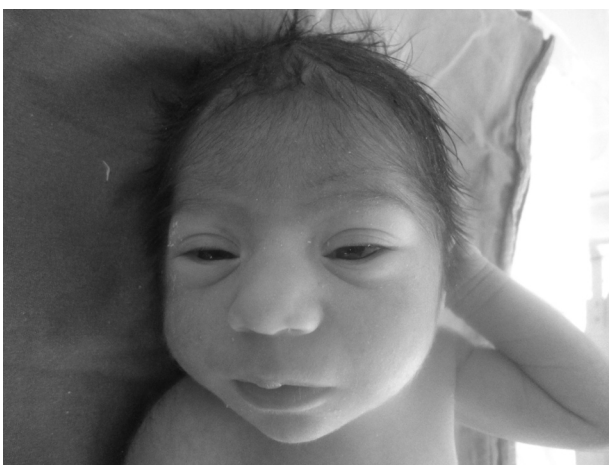


Figure 2. Facial appearance of the patient.

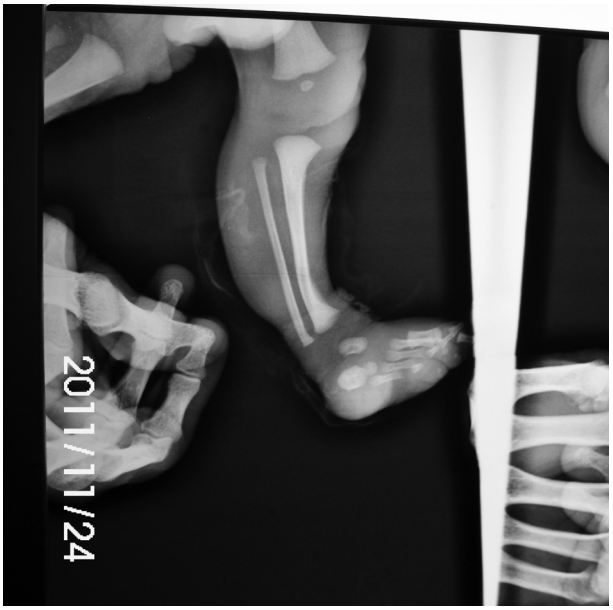


Figure 3. Radiographs of case's lower limbs: right distal tibia lying on the left side is present.

DISCUSSION

Aplasia cutis congenita is an uncommon disorder with focal absence of epidermis, subcutaneous tissue, galea and calvarial bone in rare cases. In addition to scalp and bony defects, our patient had a dura defect with herniation of brain tissue. Characteristically terminal transverse limb defects affect the distal phalanges or entire digits. Both lower and upper limb defects can be seen, but lower limb defects are more common. Shortening of the fingers with loss of terminal phalanges are the most common defects but clubfoot, syndactyly, nail hypoplasia, absence of fingers can be seen less commonly. Limb involvement is usually asymmetrical [9]. Our case had pes equinovarus, and skin defects on the right lower limb.

Few conditions may be associated with ulceration in the newborn. The most common lesion of them is the ACC either alone or with epidermolysis bullosa (EB). In transient bullous dermolysis, which is a form of dystrophic EB, the baby may have blisters on the limbs [10]. Congenital herpes may rarely be the cause of congenital abrasions [11]. Neonate with Setleis syndrome may have depressed scarred areas

on the temporal scalp resembling healed ACC. However, it can be differentiated easily by its characteristic facial features especially periorbital puffiness and inverted V-shaped mouth [12]. Other causes of ulcerations in neonates that should be differentiated from ACC include ulcerations caused by scalp electrodes [13], and pyoderma gangrenosum [14].

The management of non-scalp ACC is still controversial. Most lesions heal spontaneously with conservative dressing, but large lesions may necessitate surgical interference with skin grafts or local skin flaps [2]. Fresh allograft has been used as temporary biological dressing to enhance epithelization of the defects [15]. Cultured epithelial autografts have been used together with acellular allogenic dermal grafts [16]. Skin grafting is limited by donor-site availability, potential morbidity and the technical difficulties associated with handling the thin neonatal skin. Flap reconstruction involves subjecting a neonate to anaesthesia and a major surgical procedure, with the risk of significant blood loss. Although the use of cultured keratinocytes is promising, it is still restricted to centres having tissue culture laboratory.

ACC is a rare disorder that is present at birth. The most common presentation is the solitary lesion on the scalp. The peculiarity of a patient is that his mother had a similar lesion, possible evidence for a genetic influence. The presence of ACC in both mother and child is rare, but the condition has been noted in siblings [17,18]. Localized congenital absence of skin is also seen in Bart syndrome [18] which is now considered to be a variant of EB. There appears to be a clear genetic influence in many cases, but the same mechanism is un likely to be associated with each case. Friedan proposed a classification of ACC [19]. The clinical description of our patient points to type 7 in Frieden's classification.

In conclusion, we present a rare case who had a wide aplasia cutis area on the right lower limb, and the clinical features were discussed in the light of the literature.

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