Aplasia cutis congenita: a case report

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Conflict of interest: none to declare

Received: 12 June 2012
Accepted: 20 October 2012

Aplasia cutis congenita is the congenital absence of skin most commonly affecting the scalp. No definite etiology is available but multiple causes such as intrauterine infection, fetal exposure to cocaine, heroin, alcohol or antithyroid drugs, vascular disruption, genetic causes, syndromes and teratogens have been suggested. We present an infant with extensive aplasia cutis of the trunk and thigh. He was the third child of his parents while the outcome of the first and the second pregnancy was intrauterine death; the dead fetuses however had no skin anomaly.

Keywords: aplasia cutis congenita, scalp, skin defect, trunk

Iran J Dermatol 2013; 16: 36-38

INTRODUCTION

Aplasia Cutis Congenita (ACC), congenital absence of the skin, is an uncommon anomaly. It most commonly presents as a solitary defect of the scalp but may also involve the trunk and extremities. The lesions are non inflammatory, well demarcated and have variable extents, ranging from 0.5 to 10 cm or more [1,2]. It is present at birth. The cause is not clear but genetic factors, compromised vasculature to the skin, infection, teratogens, fetus papyraceous and trauma are all implicated [1-5]. Truncal aplasia cutis congenita has been reported with biliary atresia, distal duodenal atresia, intestine infarction and multiple hepatic hematomas [3,6]. Syndromes such as Adams Oliver syndrome, SCALP syndrome (nevus sebaceus, CNS malformations, aplasia cutis congenita, limbal dermoid, pigmented nevus), Opitz syndrome, and chromosomal disorders are associated with this lesion [2,7-10]. The main complications of larger defects include infection, bleeding and thrombosis that may be fatal. Therefore, prompt diagnosis and appropriate treatment are critical for avoiding the adverse outcomes. Management is conservative and surgical. Allogenic dermal graft and cultured epithelial autografts have also been used to reconstruct the defects [2,4,11-13].

Histological details are available in very few reports. Histological features vary depending on the depth and duration of aplasia. Ulcers are seen at birth. After healing, the epidermis appears flattened with proliferation of fibroblasts within a connective tissue stroma. The total absence of the epidermal appendages remains a characteristic feature [14]. We describe a new case of ACC of the trunk and thigh in a neonate.

CASE REPORT

A 1-day-old male, term neonate with appropriate growth for gestational age (birth weight 2850 gr, length 50 cm and head circumstance 34 cm), was noted to have bilateral skin defects on the trunk and thigh at birth. These lesions were symmetrical in appearance with a diameter of about 10-12 cm (Figure 1). A healed fibrous band was found on the abdomen and thigh. He was the third child of his parents. Prenatal history was significant for one fetus intrauterine death at 24th week of gestation and another fetus was stillbirth. However, there was no skin anomaly or defect in the dead fetuses. There were no other organ abnormalities on clinical examination. Radiological examination and ultrasonography of the abdomen revealed no abnormalities. Liver and gallbladder were normal

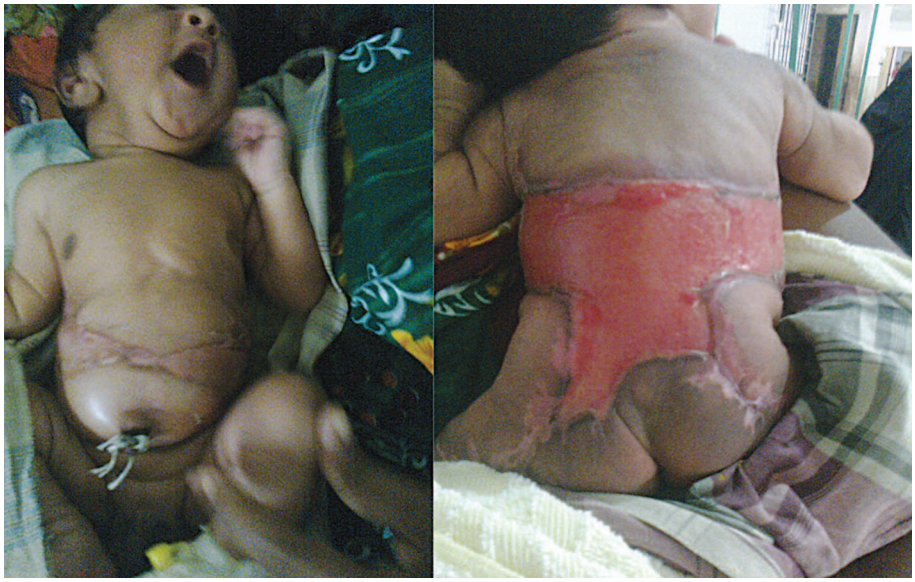

Figure 1. Aplasia cutis congenita in different body areas.

in size and echo. Routine laboratory data and liver function test were normal. The affected area was treated by systemic and topical antibiotics and local daily dressing. Neurologically, the infant had no obvious deficit. The skin lesion gradually healed without any surgical procedures. Muscular tone and hearing were normal. All laboratory findings were normal. Histopathological report showed loss of epidermis and fibrosis. No epidermal appendage was found in the lesion (Figure 2). Unfortunately, his parents were very much poor and unable to continue hospital based treatment and left the hospital with appropriate recommendations.

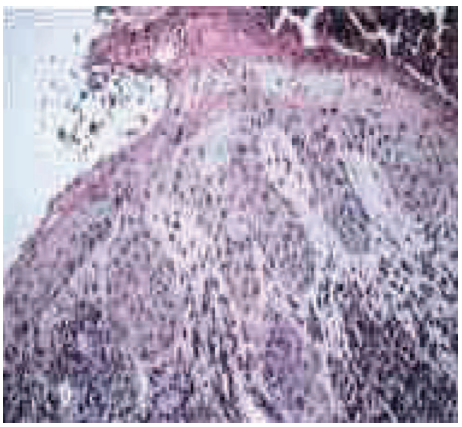

Figure 2. Loss of epidermis and fibrosis with no epidermal appendage in the lesion (H&E*100)

DISCUSSION

ACC is an uncommon disorder presented at birth. The most common presentation is a solitary lesion on the scalp but in our case, the lesion was on the trunk and extremities. The significant factor of this patient was history of two intrauterine deaths of first two children of his parents. Truncal aplasia cutis with fetus papyraceous has been reported in other reports [3,9,14]. This disorder occurs sporadically with no familial history. Affected patients show large areas of absence of skin that have a bilateral pattern of distribution along the flanks, back, abdomen and the lateral aspect of the limbs [15,16]. Therefore, prenatal ultrasound has been a great help in better understanding of this disorder. The cause of the symmetrical type of aplasia cutis is a vascular disruption inducing abnormal dermoepidermal development or cutaneous defects through ischemic and thrombotic events. Other abnormalities such as hepatic hematoma, dudonal atresia, and biliary atresia may be observed. These findings prove the hypothesis of the vascular origin of the disorder [3,6,16]. Our patient did not have any other organ abnormalities. However, in other cases, no relationship has been found between the extension and localization of the disorder in the viable neonate [3,17]. Wu reported a 1-year-old boy with scalp aplasia cutis associated with the clinical manifestations of intractable seizures and developmental delay [18], but mental deficit is not

reported in the literature. The relationship between aplasia cutis congenita and developmental delay needs more investigation. However,different clinical presentations may be observed in infants with aplasia cutis congenita born from twin or triple pregnancies associated with early death of one or two fetuses.

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