Case Report

Iran J Pediatr
Jun 2009; Vol 19 (No 2), Pp:185-188

Aplasia Cutis Congenita: a Case Report

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Received: *Aug 19, 2008*; Final Revision: *Jan 23, 2009*; Accepted: *Feb 18, 2009*

Abstract

Background: Aplasia cutis congenital (ACC) is a 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.

Case Presentation: We present an infant with symmetrical type of aplasia cutis on the trunk and proximal limbs. She was product of triple pregnancy with two fetuses papyraceous at 12th week of gestational age and at birth. She is treated by non surgical management despite remarkable extent of the lesion.

Conclusion: ACC of the trunk is less common than of scalp. Lesion often is symmetric and seen after fetus papyraceous in multiple pregnancies.

Iranian Journal of Pediatrics, Volume 19 (Number 2), June 2009, Pages: 185-188

Key Words: Aplasia cutis congenital; Fetus papiraceus; Skin defect; Twin pregnancy

Introduction

Aplasia cutis congenita (ACC), congenital absence of 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 noninflammatory, well demarcated and have variable extent range 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 cebaceus, CNS malformations, aplasia cutis congenita, limbal dermoid, pigmented nevus), Opitz

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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, 12,13].

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. Total absence of the epidermal appendages remains a characteristic feature[14]. We describe a new case of ACC of the trunk in a neonate with two papyraceous fetuses.

Case Presentation

A 1-day old female, term neonate with appropriate growth for gestational age (birth weight 3250 gr, length 50 cm and head circumstance 34 cm), bilateral skin defects on the trunk and lower extremities noted at birth. These lesions were gelatinous, symmetric, stellate appearance with about 8-10 cm in each radiation (Fig 1).

She was product of a triple pregnancy. Prenatal history was significant for one fetus aborted at 12th week of gestational age and another fetus papyraceous lost at birth.

There were no other organ abnormalities except systolic murmur on clinical examination. Echocardiography proved VSD. Radiological examination and ultrasonography of abdomen revealed no abnormalities. Liver and gallbladder were normal in size & echo. Routine laboratory data and liver function test were normal.

Affected area was treated by mupirocine and vitamin A+D ointment about two months. We visited her at 4 & 14 months of age. At 4 months growth was normal. Neurologically, the infant had no obvious deficit. The skin lesion healed without any surgical procedures (Fig 2A). Clinical examination at 14 months revealed minimal delay in growth and development. (weight 9200 gr, length 73 cm, head circumference 45 cm). She was able to sit but unable to stand or walk. Muscular tone and hearing were normal. Skin lesions were also flattened & cicatricle (Fig 2B). All laboratory findings were normal.

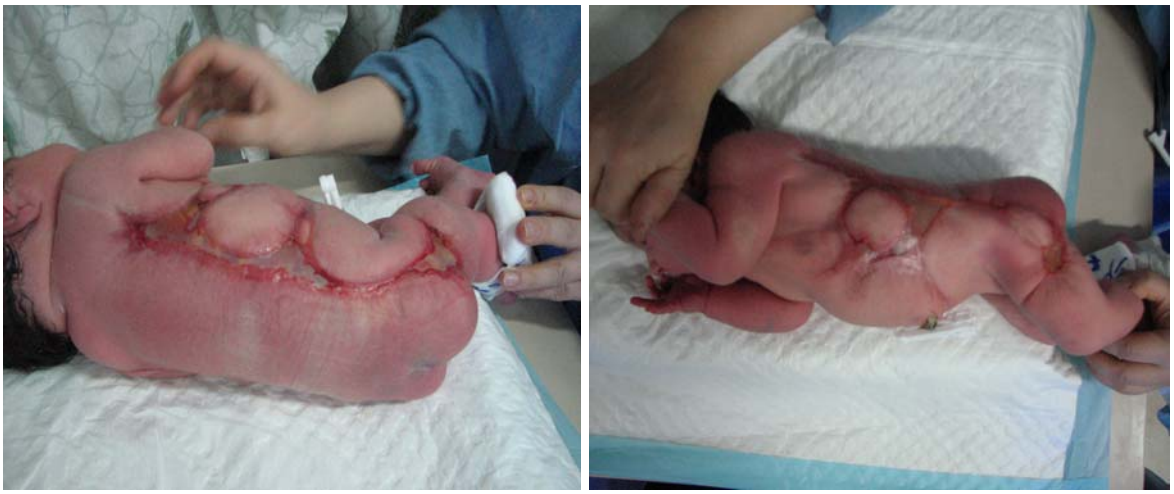

Fig 1: Aplasia cutis congenita in our patients at birth (before treatment)

Fig 2: Aplasia cutis congenita in our patients at 4 months (A) and at 14 months (B)

Discussion

ACC is an uncommon disorder presented at birth. The most common presentation is the 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 papyraceous fetuses simultaneously.

Truncal aplasia cutis with fetus papyraceous have also been reported in other reports[3,9,14]. During a twin pregnancy the death of one fetus occurring at the end of first trimester or at the beginning of the second may result in the persistence of a dead fetus (fetus papyraceous) in association with a live and viable twin [3]. Our patient is a severe example of aplasia cutis congenita associated with two fetus papyraceous. This disorder occurs sporadically with no familial history.

Affected patients show linear areas of absence of skin that have bilateral pattern of distribution along the flanks and the lateral aspect of the limbs[15,16]. Therefore, prenatal ultrasound has been a great help in allowing a better understanding of this disorder.

The cause of the symmetrical type of aplasia cutis is a vascular disruption inducing abnormal dermoepidermal development or cutaneous defect through ischemic and thrombotic events. The intrauterine death of one of the fetuses should cause the release of the thrombosis-promoting material from the dead fetus. These substances can cause placental infarction, disseminated intravascular coagulation and cutaneous lesions [16,17].

Other abnormalities such as hepatic hematoma, dudonal atresia, biliary atresia may be observed. This findings prove hypothesis of the vascular origin of the disorder [3,6,16]. Our patient did not have any other organ abnormalities.

It is suggested that death of the twin in earlier gestation, could lead to more extension and increase of truncal form of this lesion in the survivor twin. However, in other cases no relationship was found between the extension and localization of the disorder in the viable twins [3,17].

Our patient in 18 months of age had minimal growth and developmental. Wu reported a 1-year old boy with scar of scalp, aplasia cutis associated with the clinical manifestation of intractable seizures and developmental delay,[18] but mental deficit in others is not reported. Relationship between aplasia cutis congenita and developmental delay needs more studies.

Conclusion

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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