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Case Report

Aplasia cutis congenita with rare association and unusual presentation

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Abstract

Aplasia cutis congenita (ACC) is a congenital absence of skin most commonly affecting scalp. In very rare instances it can involve extremities and trunk. It is usually diagnosed clinically at birth. Most cases are sporadic with a few reports of familial occurrence. Here, we present two cases of which one had scalp involvement correlating to antenatal methimazole intake and the other one had unusual presentation with involvement of deep tissues up to the periosteum of right lower limb. Both cases were managed conservatively and successfully discharged home. But unfortunately were lost to follow-up.

ACC is a rare condition with varying degree of severity depending upon the extent and depth of the involvement of the skin and underlying tissues. Mild cases do not need specific intervention while severe cases need to be managed by a multidisciplinary team of pediatrician, dermatologist and plastic surgeon. Methimazole is a known teratogen associated with this condition hence should be avoided in pregnancy.

Key words
Cutis aplasia, absent skin, methimazole.

Introduction

Aplasia cutis congenita (ACC) is developmental absence of skin in a localized or widespread area at birth. Lesion is solitary in 70% of cases but ACC can involve multiple sites.[1] It is estimated to affect approximately 3 in 10,000 live births.[2]

It was first reported by Cordon in 1767.[2] It can involve epidermis, dermis, subcutaneous tissue and rarely to periosteum. The depth and size of ulcer predicts its outcome. When the lesion occurs in early pregnancy it may also heal before delivery leaving a congenital atrophic alopecic scar.[3] The most commonly involved site is scalp but can occur rarely in extremities or trunk. Defects involving the trunk and extremities are often larger than those on the scalp. It can occur as an isolated skin defect or associated with other malformation syndromes.[4] Factors implicated in the etiology are genetics, compromised vasculature of the skin, infection, teratogens, fetus papyraceus and trauma. The main complication of larger defects includes infection, bleeding and thrombosis that may be fatal.[4]

We present a rare and unusual presentation of this medical condition encountered by our team in NICU of tertiary care center.

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Case 1

A newborn baby boy delivered through elective cesarean section at 38 weeks of gestation to a multigravida mother with normal APGAR, was noted to have a circular area of absent skin on parieto-occipital region of scalp measuring 2.0

cm x 2.0 cm. Rest of the examination was unremarkable. He was product of nonconsanguineous marriage. Mother was hyperthyroid and was receiving methimazole for treatment before and during pregnancy.

The baby was successfully managed conservatively

Case 2

A baby girl born at full term (38 weeks of gestation) after an elective cesarean section to a 25-year-old woman (gravida 2, para 0, aborta 1) in a peripheral hospital and was referred to us with skin lesion. APGAR scores at birth were normal. There was no history of consanguinity or any familial genetic disorders. Mother's antenatal history was uneventful.

On examination, the neonate was found to have absent skin over the anteromedial aspect of right lower limb. It started from the mid-thigh and extended upto the plantar aspect of the foot. The lesion had well-defined margins involved roughly 10-12cms of skin area. There was minimal serosanginous oozing at the floor of the lesion. No sloughing of skin or blister formation was noted after applying pressure. The nails appeared normal. It was associated with shortening of right leg. The rest of the skin had normal appearance and the infant had no other associated anomaly. A clinical diagnosis of aplasia cutis congenita was made. Multidisciplinary team including neonatology, Plastic surgery, orthopedics and dermatology was taken on board.

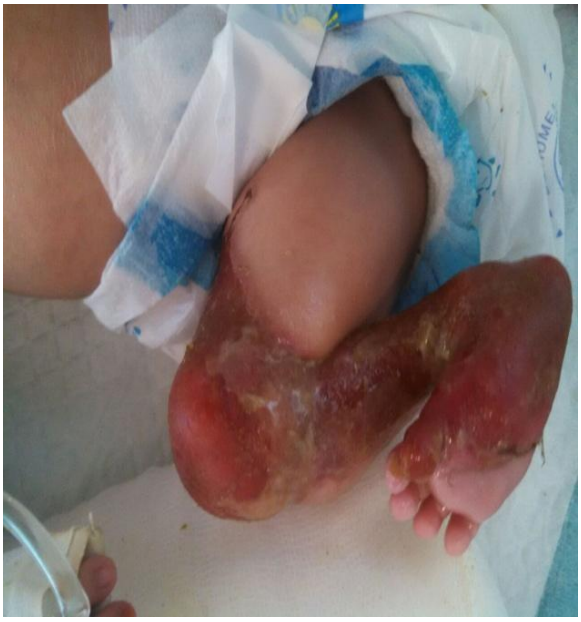

Figure 1 Well-defined eroded area involving right lower limb without blister.

Discussion

Aplasia cutis congenital (ACC) is a rare, heterogeneous congenital disorder. Frieden5 has classified it into 9 groups according to location of lesion and associated malformations. It can also be clinically categorized upon the depth of the lesion. Upper dermis and epidermis involvement cause minimal scarring or hair loss (mild disease) but those involving deep dermis, subcutaneous tissue (moderate disease) and periosteum causing major scarring (severe disease).[6]

The lesion described in our case 1 comes under group 8 of Frieden classification which occurs after teratogen exposure. Methimazole is a known drug with a teratogenic effect for cutis aplasia. and therefore should be avoided in pregnancy for treatment of hyperthyroidism.[8]

The case 2 can be characterized in group 7 which includes Aplasia cutis of extremities without epidermolysis bullosa. The extremity involvement is rare.[9] The patient had right lower extremity affected with limb shortening. The

depth of the lesion was upto the periosteum which is a severe disease and could compromise blood supply resulted in affected short limb

Management is conservative. Small Lesion usually heals in the first year of life spontaneously1 but large lesions may necessitate surgical interference with skin grafts or local skin flaps.[10] Use of cultured keratinocytes is promising but restricted to centers having tissue culture facilities.[11] We consulted plastic surgery for possible need of skin grafting in our case 2. But Parents and wanted to wait for spontaneous recovery with conservative management.

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