Aplasia cutis congenita: A case report

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ABSTRACT

Aplasia cutis congenita (ACC) is a congenital absence of skin most commonly involves the scalp. There is no definite etiology 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. Here, we report the case of a late preterm male baby weighing 1.9 kg born at 36 weeks of gestation to a third gravida mother with a previous history of one stillbirth and one baby's death on day 3 of life. Multiple well-demarcated raw areas showing an absence of skin over the neck, extremities and back of varying diameter were seen. X-ray showed features suggestive of pyloric stenosis and ultrasound of the abdomen showed bilateral hydronephrosis. A skin biopsy showed a full-thickness absence of skin and dermal appendages in the involved areas, with dermis and epidermis ending abruptly. A final diagnosis of ACC was made based on the investigations. ACC of the trunk is less common than scalp, and different clinical presentations may be seen in infants with aplasia cutis.

Key words: *Aplasia cutis congenita, Neonatal skin defect, Skin defect*

Aplasia cutis congenita (ACC), a rare anomaly, is the congenital absence of skin. It commonly presents in the form of a solitary defect of the scalp but may also involve the trunk and extremities. The lesions are non-inflammatory, well-demarcated and have a variable range of extension from 0. 5 to 10 cm or more [1,2]. The implicated factors contributing to this anomaly are genetic factors, compromised vasculature to the skin, infection, teratogens, fetus papyraceous, and trauma. Truncal ACC has been reported in association with biliary atresia, distal duodenal atresia, intestine infarction, and multiple hepatic hematomas [3]. Although they are usually benign, may also be associated with other physical abnormalities and syndromes. Frieden classified them into nine groups based on the number and presence or absence of other anomalies. Almost, 86% belong to the first group with a solitary lesion [4].

CASE REPORT

A late preterm male baby weighing 1.9 kg was born at 36 weeks of gestation to a third gravida mother with the previous history of one stillbirth and one baby's death on day 3 of life. The father was mother's first-second cousin and the mother's prenatal course was unremarkable. There was no history of any infections, fever, exposures to medications, or teratogens

He was delivered by vaginal route and had Apgar scores of 7 and 9 at 1 and 5 min, respectively. On examination, this preterm baby had respiratory distress. There were multiple well demarcated raw areas showing an absence of skin over the neck, extremities, and back. The lesions were of varying diameter and had well-defined margins (Figs. 1 and 2). There was minimal bleeding at the lesion sites. The rest of the skin has a normal appearance. He had bilateral small dysplastic pinnae, flat nasal bridge. The baby had oral mucosal ulcers and positive Nikolsky's sign. He developed severe respiratory distress and was put on a ventilator, as he was not maintaining SpO_2 on continuous positive airway pressure, vasopressors were given for cardiovascular instability.

As the blood count and C-reactive protein showed sepsis, the patient was started with intravenous antibiotics along with the local dressing of the affected areas with mupirocin. There was a history of similar lesions in the second born male child, who died after 3 days of birth. X-ray showed features suggestive of pyloric stenosis (Fig. 3). An ultrasound of the abdomen was done to diagnose pyloric stenosis; it showed bilateral hydronephrosis as well. Echocardiography showed an atrial septal defect (3.1 mm) and ventricular septal defect (4 mm). Skin biopsy showed a full-thickness absence of skin and dermal appendages in the involved areas, with dermis and epidermis ending abruptly.

A final diagnosis of ACC was made on the basis of an absence of the skin over the extremities, face and associated pyloric stenosis with syndromic features. In spite of aggressive supportive management, baby succumbed to infection and died at 80 h of birth, due to septicemia.

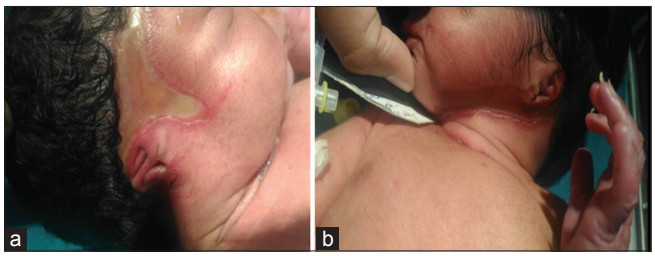

Figure 1: Raw skin seen over the face and around ear with right dysplastic pinna (a); fold of the neck and the left dysplastic pinna (b)

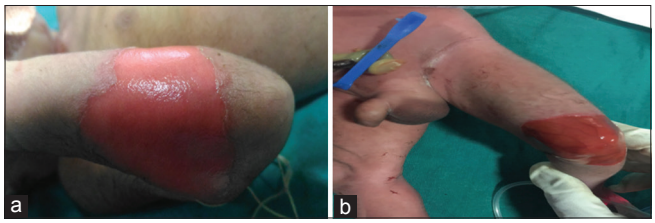

Figure 2: An absence of the skin with irregular margin on the right forearm (a) and the left knee with visible veins (b)

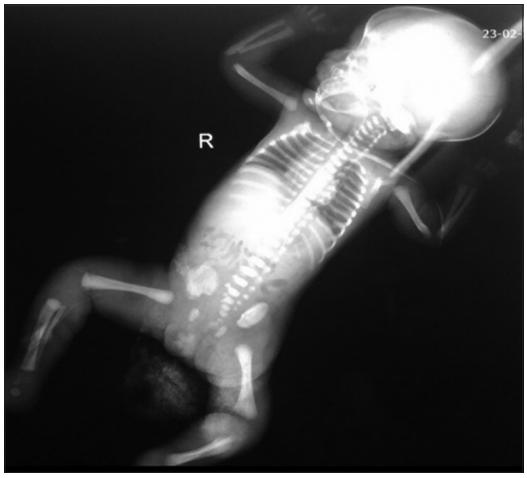

Figure 3: Roentogram showing distended stomach with distal gas shadow

DISCUSSION

ACC can appear anywhere in the body; however, in 84% of cases, the defect is found in the scalp and often solitary. It is also located in the midline vertex most of the times. Non-scalp lesions may include involvement of the trunk and/or extremities and are usually bilaterally symmetric, but asymmetric distribution has also been reported as well [5]. At birth, the appearance of the lesion may range from superficial erosion to a deep ulcer. The affected area might be covered with a thin, transparent membrane. The association of ACC with epidermolysis bullosa (EB) can be taken as a visible sign of other congenital anomalies, for example, pyloric or duodenal atresia, ureteral stenosis, renal abnormalities, craniofacial abnormalities, and nail dystrophy.

Bullous or membranous ACC is a clinical subtype of this condition, with extremely few cases reported in literature either due to underreporting or to it being so rare [6]. The present case did not have any bullae at birth, but features such as positive Nikolsky's sign suggests the diagnosis of EB causing intrauterine blistering and a presentation like an aplasia cutis. This in association with congenital malformations such as dysplastic pinnae, bilateral hydronephrosis places our case into a lethal autosomal recessive form of aplasia cutis type six [6,7].

The diagnosis is primarily clinical, and a biopsy is not needed nor suggested routinely, but characteristically there is an absence of epidermis and dermis and decreased subcutaneous tissue as deep as muscle and fascia. Healed lesions often demonstrate flattened epidermis, a proliferation of fibroblasts, newly formed capillaries, and complete absence of adnexal structures. Histological details are available in very few reports; depending on the depth of aplasia and duration, histological features vary. Ulcers are present at birth. After healing, the epidermis appears flattened with the proliferation of fibroblasts within a connective tissue stroma. A complete absence of the epidermal appendages is a characteristic feature [8,9]. The diagnosis can be suspected *in utero* with elevated amniotic fluid alpha-fetal protein levels, positive acetylcholinesterase, and normal ultrasound findings. In all patients with ACC, a complete obstetric and family history should be obtained, as well as a medical examination guided by the pattern of the lesions. Genetic counseling of the family regarding the risk of recurrence is recommended [10].

ACC (also known as "cutis aplasia," "congenital absence of skin," and "congenital scars") is the most common congenital cicatricial alopecia and is a congenital absence of epidermis in focal areas with or without the involvement of other layers of the skin. It can be associated with trisomy 13, Adams-Oliver syndrome, Johanson-Blizzard syndrome, and Wolf-Hirschhorn syndrome. It can also be seen if there is *in utero* exposure to methimazole and carbimazole. This dermatological manifestation has been linked to a deletion in chromosome 19 and peptidase D haploinsufficiency [11,12].

CONCLUSION

ACC is a rare congenital disorder characterized by an absence of skin, most frequently overlying the scalp. ACC of the trunk is less common than of scalp, can be associated with many anomalies. The baby had a sibling with a similar condition, as per history. However, while revising that was removed as lack of evidence. We can remove the whole line. Hence, a proper case history should be taken for the correct diagnosis of such anomaly.

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