

Isolated Aplasia Cutis Congenita of the Lower Limb in a Newborn: A Case Report

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ABSTRACT

Aplasia Cutis Congenita (ACC) is a rare congenital disorder characterised by a lack of skin at birth, most commonly affecting the scalp, though other body areas may also be involved. The lesions typically range from 0.5 cm to 10 cm in diameter and are usually well-defined, non-inflammatory, and devoid of epidermis and, in a few cases, dermis or deeper tissues. The exact aetiology of ACC remains uncertain, though various causative factors have been proposed, including genetic mutations, intrauterine vascular compromise, maternal infections, teratogenic drug exposure during pregnancy, fetus papyraceus, and traumatic events during delivery. This case report presents an unusual case of isolated ACC, corresponding to Group VII of Frieden's classification, involving the unilateral lower limb of a one-day-old full-term neonate, without any associated anomalies or systemic involvement. The lesion was present at birth, sharply demarcated, and devoid of signs of infection or ulceration. Conservative local wound care and gentle debridement were performed. Remarkably, the affected area showed spontaneous epithelialisation over the following weeks, healing completely without residual scarring or the need for surgical grafting. This case underscores the importance of recognising rare presentations of ACC beyond the scalp, as well as the potential for favourable outcomes with minimal intervention in selected cases. Early diagnosis, exclusion of underlying syndromic associations, and careful monitoring are essential for optimal management and prognosis.

Keywords: Congenital condition, Debridement, Genetic mutation, Non-inflammatory skin lesion

CASE REPORT

A full-term {38-week Period of Gestation (POG)} male baby weighing 3.8 kg presented to the department on Day one of life with the complaint of congenital absence of skin on the left foot. Baby was delivered via normal vaginal delivery, cried immediately after birth with stable vitals and was born to a 30-year-old primigravida mother. His antenatal history was uneventful. Routine prenatal ultrasounds done at 12 weeks POG and 22 weeks POG had shown a single viable intrauterine foetus without any gross malformations and adequate liquor for gestational age.

On examination, the child was afebrile with a heart rate of 128 beats/min and a respiratory rate of 38/min. Systemic examination has been normal except on local examination lesion was well demarcated, covered with a whitish membrane that involved toes and fingers along with two-thirds plantar and dorsal surface area of the left foot, as shown in [Table/Fig-1]. No other skin lesions were observed anywhere on the body surface.



[Table/Fig-1]: View of lesion involving two-thirds plantar and dorsal surface area of the left foot.

were noted. The mother's TORCH profile, Antinuclear Antibody (ANA), and Antiphospholipid Antibody (APLA) tests were all negative. Baby's detailed coagulation profile was also normal. There was no history of febrile illness during pregnancy, exposure to known teratogenic medications, radiological exposure, substance abuse, any chronic illness, or traumatic delivery, because lesions are sometimes mistakenly confused with scalp electrodes and obstetric trauma.

So based on clinical findings, a diagnosis of isolated ACC of the lower extremity was established.

The baby was managed conservatively, with local debridement performed and topical antibiotics (fusidic acid) applied. The wound healed on its own without any complications, as shown in [Table/Fig-2].



[Table/Fig-2]: Affected newborn after 3 days.

DISCUSSION

The ACC was a rare entity whereby pathogenesis involves either failure of the skin to fully develop on the body or degeneration of the skin lining. ACC was previously documented in literature as a rare case with isolated involvement of a unilateral limb, with even lower incidence than the overall incidence reported to be of 1-3 children out of 10,000 live births [1]. It was initially reported in the

literature by Cordon in 1767 [2]. It is defined by the local lack of skin tissue after birth, most often affecting the scalp, although it may also encompass the trunk and extremities [3]. Exact aetiology is not known, but it can be due to chromosomal abnormalities, intrauterine trauma, and amniotic fluid abnormality, foetus papyraceus [4]. It has been linked to many genetic disorders, including trisomy 13, Wolf-Hirschhorn syndrome, Johanson-Blizzard syndrome, and Adams-Oliver syndrome. Mutations in some genes, like ribosomal GTPase BMS1, have been associated with autosomal dominant ACC [5]. A classification system for ACC was proposed by Frieden IJ and it has nine distinct subgroups, categorised by the quantity or location of lesions, as well as the presence or absence of other abnormalities [6].

Large defects of aplasia cutis can lead to complications such as infection, bleeding, and thrombosis. Management is usually conservative [7], as was done in the index case, and surgical interventions are done in large defects. The management of ACC is mostly guided by size and the anatomical site of the defect. In most cases, a conservative approach is favoured, which supports natural skin regeneration and typically results in the formation of a thin atrophic scar and hairless skin over the course of a few weeks. For small lesions that are <4 cm with no other abnormality, daily cleansing of the lesion and daily application of topical antibacterial ointment (Fusidic acid, as used in the index case) are recommended until healing is complete. Lesions will take a few weeks to a few months for healing, and after healing, a hairless atrophic scar forms at the lesion site. Larger lesions (>4 cm) are more prone to complications. Early surgical intervention is recommended to avoid complications and to achieve adequate wound closure. A fully synthetic dermal substitute can be used to manage large defects [8].

A comparable case published in the Indian Journal of Paediatric Dermatology described a five-day-old male neonate with extensive bilateral ACC affecting the anteromedial aspects of both lower limbs. The infant was otherwise healthy, with normal imaging of the abdomen and central nervous system. Management was conservative, utilising topical antibiotics and cotton padding [9].

CONCLUSION(S)

The ACC has rarely been reported in the literature; however, diagnosis is usually made on clinical findings. It is important to assess for underlying causes and other congenital anomalies. The overall short-term and long-term prognosis of ACC is good with no permanent defects if no underlying malformation is present. In smaller defects, there is a laying down of epithelial tissue over a period involving epidermis and dermis, leading to the formation of an atrophic scar. In conclusion, we believe that newborn babies have an excellent capacity for rapid regeneration of skin with minimal complications. So, the message of reporting the Index case is to bring out in the public domain that most cases of ACC could be managed conservatively.

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