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## Isolated bilateral symmetrical aplasia cutis congenita of lower limbs in a newborn: A rare entity

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**Abstract**

Aplasia cutis congenita (ACC) is a heterogenous group of disorders characterized by the absence of a portion of skin in a localized or widespread area at birth. The scalp is the most commonly involved area with lesser involvement of trunk and extremities. We hereby report a rare presentation of this entity with isolated bilateral symmetrical involvement of lower extremities.

**Keywords:** Aplasia cutis, lower extremities, rare

**Introduction**

Aplasia cutis congenita (ACC) is a heterogeneous group of congenital disorder characterized by a localized absence of skin, dermal appendages and subcutaneous tissues since birth of the child<sup>[1]</sup>. More than 500 cases have been documented since it was first described, but because of significant underreporting, the precise frequency is unknown. An estimated incidence is approximately 3 in 10,000 births.

First reported by Cordon in 1767, aplasia cutis congenita manifests as a solitary defect on the scalp in 70% of cases, but it may sometimes occur as multiple lesions and can affect any part of the body, including the trunk and limbs. It may occur in isolation or may be associated with other anomalies, eg Adams Oliver syndrome, trisomy 13. Distorted hair growth, known as the "hair collar sign", is a marker for an underlying cranial defect such as encephalocoele, meningocoele. The ACC lesions are typically non-inflammatory and well-demarcated, ranging widely in size. A classification system was devised by Frieden in 1986 consisting of nine main ACC types based on the number, the location of the lesions, and the presence or absence of associated deformities<sup>[1]</sup>.

Most cases of ACC occur sporadically, though familial cases have been reported<sup>[2, 3]</sup> with the mode of transmission either autosomal dominant with reduced penetrance or autosomal recessive<sup>[2]</sup>. It does not show any sexual or racial predilection. There is no unifying theory for etiology and pathogenesis. Proposed mechanisms include the incomplete closure of the neural tube or embryonic fusion lines, intrauterine trauma, vascular compromise from placental insufficiency, ischemic or thrombotic event from fetus papyraceus (the demise of a twin fetus in the late first or early second trimester of pregnancy), amniotic membrane adhesions, intrauterine infections, genetic mutations and teratogens (Methimazole, valproic acid, angiotensin-converting enzyme inhibitors, benzodiazepines, misoprostol and cocaine).<sup>(4)</sup> Prognosis is usually excellent especially when no other anomaly is co-existent. Majority of the cases are treated successfully with conservative management comprising of wound cleaning, dressings and antibiotics leading to gradual epithelisation and formation of hairless atrophic scar over weeks. Surgical correction in the form of primary wound closure, skin grafts, rotational flaps is considered in cases with large or multiple defects.

We report a rare presentation of ACC with isolated bilateral symmetrical lower limb involvement.

**Case:** 30 yrs, G4P2L1A1, previous 1 cesarean delivery, with non-consanguineous marriage at 38 weeks presented to labour room with pain in abdomen. She was a booked case with uneventful antenatal course till last visit. Past drug, medical & surgical history was insignificant. Family history was also not significant.

Obstetric scans of only 26 weeks and 36 weeks were available and revealed no congenital anomaly.

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Patient underwent cesarean section i/v/o previous cesar with oligohydramnios and delivered a Female child of 2.3kg.

On physical examination, there was absence of skin over bilateral anteromedial aspect of lower limbs involving knees and dorsum of feet. The child showed no signs of neurological impairment and no other organ abnormalities. Routine investigations were within normal limits. Ultrasonography of the abdomen and central nervous system imaging revealed no abnormalities. Child was managed conservatively with wound cleaning, cotton padding and topical antibiotics.

### Discussion

Aplasia cutis congenita is one of the uncommon disorders of skin embryonal development presenting at birth and characterized by a localized absence of skin, dermal appendages and subcutaneous tissues. Because of its benign course in most the cases, this entity remains underreported and thus, exact incidence is difficult to cite. The ACC lesions may occur on any body surface although localised agenesis of the scalp is the most frequent pattern. In approximately 20% of cases underlying bone defects are also found.

Though most reported cases are sporadic, familial occurrence is known.

Exact pathogenesis of ACC is unknown. However, there are multiple factors that probably contribute to the development of ACC- Trauma, amniotic irregularities, teratogens (especially methimazole), vascular lesions, thrombosis. Mutations in ribosomal GTPase BMS1 have been identified as a potential cause of autosomal dominant ACC [5]. A recent study has also implicated the role of UBA2 gene and the SUMOylation pathway in the ACC pathogenesis [7].

Although ACC is usually benign, they can be associated with physical anomalies, syndromes and chromosomal disorders e.g; Adams-Oliver syndrome, trisomy 13, Johanson Blizzard syndrome.

Prognosis usually excellent, is influenced by site, size, number of defect and co-existing defects/anomalies. E.g; Full-thickness defects of the scalp, skull and dura are associated with a mortality rate of greater than 50%.

Decision regarding conservative or surgical management or both depends primarily on the size, location and depth of the defect. Small defects are better managed by conservative approach which help preserve moisture, prevent desiccation and allow spontaneous epithelialization. Truncal and limb defects, despite their large size, also respond well to conservative management. Surgical management is usually favoured in cases with large or multiple defects with excision and primary closure or with the use of tissue expanders and rotation of a flap. Complications are uncommon except larger scalp defects which are predisposed to complications such as hemorrhage and infection, placing patients at risk for death.

Our newborn is a case of ACC, type 7 of Frieden classification with b/l symmetrical distribution of ACC involving the antero-medial aspects of distal thighs and both lower limbs including dorsum of feet. She didn't show any others organ abnormalities & no obvious neurological deficit. There was no identifiable risk factor in her case. Imaging studies were within normal. This child was managed with conservative management at our tertiary centre and later referred to Nagpur as per parents' wish for further consultation & management.



Fig 3: The neonate under study on conservative management.

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Fig 1-2: ACC involving antero-medial part of distal thighs, lower limbs and dorsum of B/L lower limbs