Case Study

A rare case of Aplasia Cutis Congenita type VII in a Nigerian newborn: Diagnostic and psychological challenges in a low-resource setting

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ABSTRACT

Background: Aplasia cutis congenita (ACC) is a rare congenital anomaly characterized by the localized absence of skin. Type VII ACC, which predominantly affects the lower limbs, is extremely rare and usually not associated with blistering, making diagnosis challenging.

Case presentation: This reports a term male neonate delivered by caesarean section with normal APGAR scores and anthropometric parameters, who presented with bilaterally symmetrical areas of absent skin over both lower limbs and several smaller lesions elsewhere. The denuded areas were covered by translucent membranes with visible vasculature, and the calves appeared swollen and hypoplastic. No blisters or mucosal lesions were observed. The infant was markedly irritable on touch. Supportive wound care with warm saline-soaked gauze and breastfeeding were initiated, but the father declined referral and further investigations due to financial and cultural constraints. Partial epithelialization occurred within 24 hours, and minor lesions healed rapidly. The family discharged themselves after three days; the baby died at home 11 days later.

Conclusion: This case highlights the diagnostic dilemma of extensive, symmetrical, non-blistering ACC Type VII and the challenges of managing rare congenital conditions in low-resource settings. While differential diagnoses include severe forms of epidermolysis bullosa and vascular-disruption variants, definitive classification requires histopathologic or molecular studies. Early counselling and attention to sociocultural barriers are vital to improve outcomes.

Keywords: Aplasia cutis congenita, Low-resource setting, Neonate, Type VII ACC, Vascular disruption.

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INTRODUCTION

Aplasia Cutis Congenita (ACC) represents a heterogeneous group of congenital disorders characterized by localized or widespread absence of skin at birth.[1] The first report of ACC was by Cordon in 1767.[2] The condition has an estimated incidence of between 1 and 3 in 10,000 live births and is most commonly observed on the scalp, accounting for over 80% of cases.[1,3] Frieden's classification of ACC describes nine subtypes, of which types VI and VII involve the extremities, particularly the lower limbs.[4] Type VI is associated with epidermolysis bullosa (EB), while Type VII ACC is exceedingly rare and may present with symmetrical or asymmetrical skin defects, often associated with underlying malformations or constrictive bands that can impair limb development.[5]

The aetiology of ACC remains unclear, though proposed mechanisms include genetic mutations, vascular disruptions, exposure to teratogens, and amniotic band syndrome.[1,4] Management is typically conservative, focusing on wound care and infection prevention, with surgical intervention reserved for extensive or complicated lesions.[5] When blistering is absent, distinguishing non-genetic, vascular, or ischaemic variants of ACC becomes difficult. Accurate classification is essential for counselling, recurrence-risk estimation, and family support. We report a term male neonate with symmetrical limb ACC lesions and extreme irritability but no visible blistering, delivered at Obio Cottage Hospital, a primary health care facility in Port Harcourt, Nigeria.

CASE PRESENTATION

Clinical History: A term male neonate was delivered by elective caesarean section (indication: two previous caesarean sections) at 40 weeks of gestation to non-consanguineous parents. The pregnancy, labor, and delivery periods were uneventful. APGAR scores were 9 and 10 at 1 and 5 minutes respectively, and birth weight was 3.4kg. Other anthropometric parameters were appropriate for gestational age. The mother's antenatal history was unremarkable, with negative screening for HIV, hepatitis, and syphilis. She denied any medication use beyond routine antenatal supplements. She had no history of teratogenic exposure or systemic illness. The neonate was the fifth child of his parents, with all older siblings being healthy females without any similar lesions or obvious congenital abnormalities.

Clinical Examination Findings: Postnatal examination revealed a male neonate with normal vital signs and anthropometry. There were no dysmorphic facial features or signs of systemic illness. However, striking bilateral symmetrical areas of absent skin were noted on the lower limbs, extending from the distal third of the thighs to the toes, including the soles. The lateral aspects of the calves and soles were spared. The affected areas were covered by thin, translucent membranes, through which the underlying vasculature was visible. The calves appeared swollen, and the distal legs were markedly hypoplastic and deformed, although limb mobility was preserved. Additionally, smaller skin defects were seen at the base of the umbilical cord, tip of the penis, dorsomedial aspects of both wrists and thumbs, the left cheek, and beneath the lower lip. The lesions were non-blistering, sharply demarcated, and did not involve the scalp or mucous membranes. Examination of other systems did not reveal any remarkable findings. Views of the lesions seen at birth are shown in Figure 1 below.

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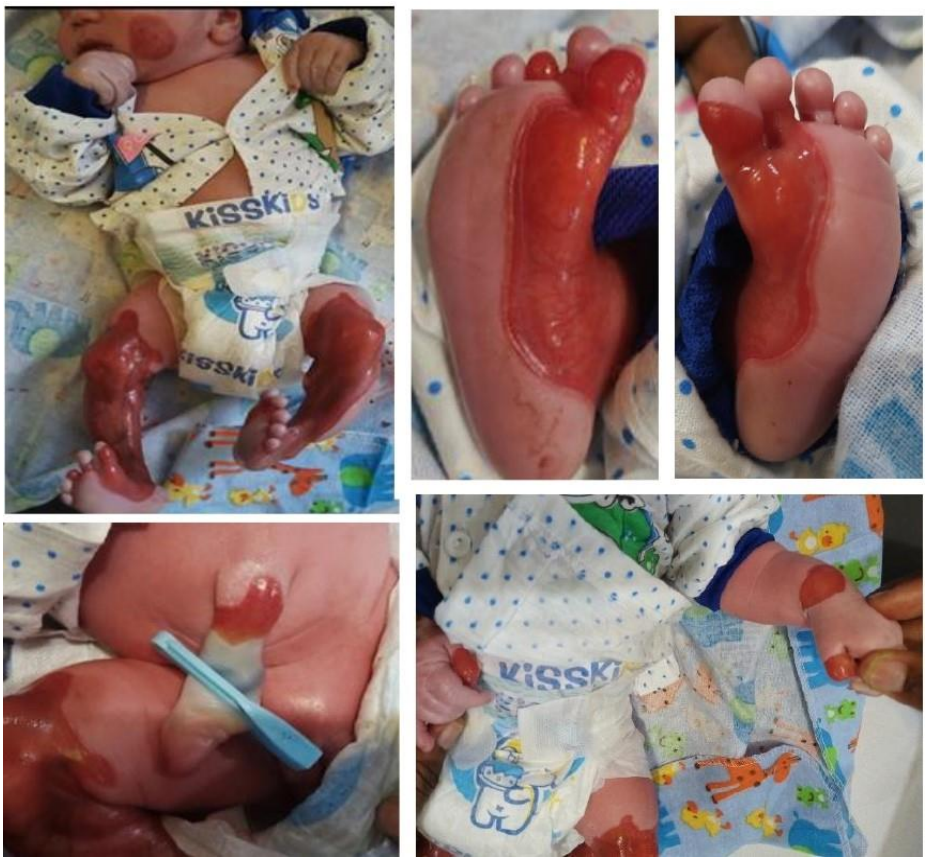

Figure 1: Images of the lesions as seen at birth

The neonate was highly irritable and cried excessively when touched. The attending paediatrician diagnosed it as ACC, possibly Type VII.

Clinical and Laboratory Investigations: Not done.
Treatment Given: Within an hour of delivery, warm saline-soaked gauze dressings were applied to the denuded areas, with a plan to start non-occlusive honey-based dressings. The mother initially became acutely distressed upon seeing the baby with the malformed lower limbs with the lesions and required mild sedation. She however started breastfeeding within hours of delivery.

Social Consequences, and Outcome: The father refused to handle the infant and declined further care and referral due to financial constraints and traditional beliefs. Remarkably, epithelialization of the lesions began within 24 hours, and the smaller defects healed rapidly. Pictures of the lesions after 24 hours are shown in Figure 2 below.

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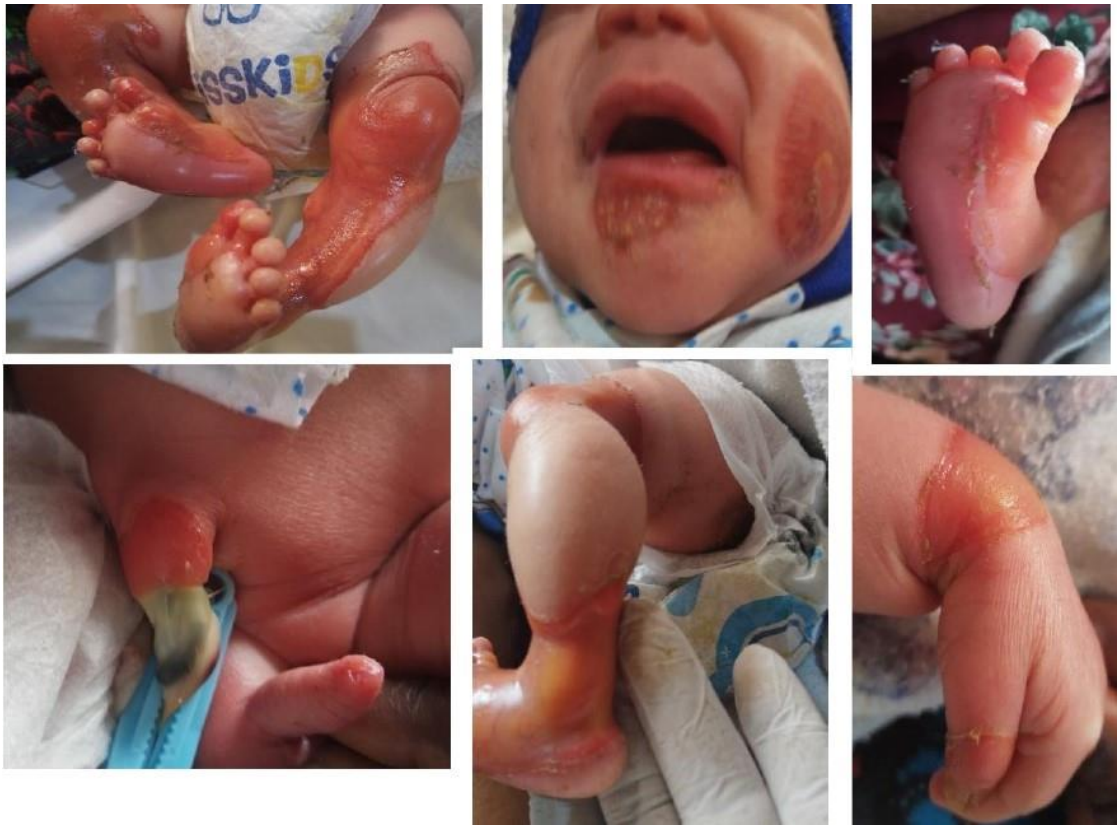

Figure 2: Images of the lesions as seen at 24 hours of life

Despite counseling, the parents discharged themselves after three days, refusing further investigations or treatment.

Follow Up: A follow-up phone call to the family revealed that the infant had remained highly irritable and had died at home, reportedly due to increasing restlessness and poor feeding. No post-mortem examination was performed. Consent for publication was granted by the father.

DISCUSSION

This case highlights a rare presentation of ACC Type VII with bilateral symmetrical involvement of the lower limbs and multiple other sites. The absence of scalp involvement and the presence of translucent membranous lesions are consistent with previously reported cases from different parts of the world,[1,4,6] and Nigeria.[5,7] The absence of visible blisters but a high degree of irritability in this infant broadens the differential diagnosis. The bilateral symmetrical distribution and constrictive appearance, lower-limb predilection, and absence of mucosal or scalp lesions suggest a vascular disruption or amniotic band aetiology, possibly due to compromised intrauterine blood flow to the lower limbs. However, no definitive cause could be established due to a lack of investigations. In the case of this neonate, this is likely a sporadic event, as many published cases have no positive family history, and recurrence risk for future pregnancies is very low.[8] However, some forms of EB, such as junctional EB or recessive dystrophic EB, can manifest with subtle or rapidly ruptured blisters, leaving erosions that mimic ACC, with possible modes of inheritance being autosomal recessive, or autosomal dominant with reduced penetrance or parental mosaicism, or X-linked.[9,10]

The rapid epithelialization observed within 24 hours supports a non-EB mechanism, as well as the notion that many ACC lesions, even extensive ones, may heal

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spontaneously with conservative management.[5,6] This aligns with literature advocating for non-invasive treatment approaches unless complications arise. Most reports from Nigeria in which different types of ACC were managed conservatively had generally good outcomes.[5,7,11-13]. These cases were managed in tertiary health care facilities. However, reported ACC cases in which there were extensive bleeding lesions[14] or associated cardiac involvement [13,15,16] required more intensive management with longer hospital stays and financial involvement, and thus had poor outcomes.

The psychosocial dimension of this case was particularly striking and played a decisive role in the newborn's outcome. The father's rejection of the newborn, motivated by deep-rooted cultural beliefs associating congenital anomalies with spiritual or moral transgressions, exemplifies the intersection of sociocultural perceptions and neonatal health outcomes in sub-Saharan Africa. In many African communities, congenital malformations are sometimes viewed as omens, curses, or evidence of parental wrongdoing.[17-20] Such beliefs can foster stigma, social isolation, and outright denial of the affected child, as was evident in this case.

This rejection was compounded by financial constraints that prevented the family from accessing advanced diagnostic and surgical interventions. The combination of cultural stigma and poverty created a barrier to timely referral, multidisciplinary consultation, and supportive neonatal care. These challenges are emblematic of the realities in many low-resource settings, where the management of rare congenital conditions such as ACC is hindered not only by limited technical capacity but also by societal attitudes and inadequate psychosocial support systems.[21]

This case underscores the need for an integrated approach to care that addresses both the biomedical and sociocultural determinants of health. Early parental counseling, empathetic communication, multidisciplinary support, and community education are critical for mitigating stigma and facilitating acceptance of affected infants.
Ultimately, the infant's death was likely due to sepsis or metabolic complications from extensive skin loss and poor home care. Histopathological and molecular investigations – such as skin biopsy for immunofluorescence antigen mapping or next-generation sequencing panels – would have been crucial for definitive diagnosis and genetic counseling but are not available in this country. This tragic case illustrates that without deliberate attention to psychosocial and cultural factors; even potentially manageable congenital conditions may lead to preventable mortality in resource-limited environments.

CONCLUSION

ACC Type VII involving bilateral lower limbs is a rare but clinically recognizable condition. Extensive types without blistering may pose a diagnostic dilemma. In this case however, early epithelialization and healing favoured a non-genetic vascular cause. Nevertheless, comprehensive dermatologic and genetic evaluation remains essential whenever feasible, to define aetiology and guide family counseling. Addressing socioeconomic and cultural barriers is vital to improving outcomes for such rare congenital conditions in low-resource settings. This case underscores the importance of early recognition, supportive care, and culturally sensitive counselling to optimize neonatal outcomes.

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