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CASE REPORT

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Aplasia cutis congenita in a Nigerian newborn: Case report from South-western Nigeria

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Abstract: We report an uncommon presentation and conservative management of Aplasia Cutis Congenita (ACC), a rare congenital defect of the skin in a newborn. The skin defects was located on the abdominal region, irregularly shaped, symmetrical, each measuring about 5 x 3 cm. There was no discharge, bleeding, bullae or anomalies on clinical examination. The child was managed conservatively with antibiotics and wound dressing. The wound granulated well, there were no local or systemic complications. By the 8th day of admission, defect had almost completely healed leaving a hypo-pigmented scar. At 2 weeks on follow-up the scar had become hyper-pigmented.

Résumé: Nous rapportons une présentation inhabituelle ainsi qu'une prise en charge conservatrice de l'aplasie cutanée congénitale (ACC), une anomalie congénitale rare de la peau chez le nouveau-né. Les lésions cutanées étaient localisées au niveau de la région abdominale, de forme irrégulière, symétriques, mesurant chacune environ 5 × 3 cm. Il n'y avait ni écoulement, ni saignement, ni bulles au niveau des lésions. Aucune autre anomalie congénitale associée n'a été mis en évidence à l'examen clinique.
L'enfant a bénéficié d'une prise en charge conservatrice associant une antibiothérapie et des soins locaux. L'évolution a été favorable avec une bonne cicatrisation des lésions, sans complication locale ni systémique. Au huitième jour d'hospitalisation, les lésions étaient presque complètement cicatrisées, laissant une cicatrice hypopigmentée. À deux semaines de suivi, la cicatrice est devenue hyperpigmentée.

Introduction

Aplasia cutis congenita (ACC), first described by Cordon in 1767,[1] is a rare congenital skin defect characterized by the localized or widespread absence of epidermis, dermis and occasionally subcutaneous tissue.[2]It's a disease entity of unidentified specific aetiology. However, several causative factors have been identified such as teratogenic substances (such as methimazole, carbimazole, misoprostol, benzodiazepines, valproic acid, methotrexate, cocaine, marijuana), intrauterine infections (such as varicella, herpes simplex, congenital rubella syndrome), genetic mutation in BMS1 gene, fetal/placental ischemia, vascular accidents, amniotic irregularities (such as early rupture of amniotic membrane), the most frequently accepted theory focuses on the tension that disrupts the skin from converging during intrauterine development.[2-6] ACC has also been shown to be associated with some syndromes such as Patau syndrome (trisomy 13), Adams-Oliver syndrome, Bart syndrome and Setleis syndrome.[2,4]
It has an incidence of about 1 - 3 in 10,000 births[7] with no racial or sex predilection. As of 2015, approximately 500 cases of ACC had been reported in the literature; all outside Africa.[3,6,8]

ACC is a clinical diagnosis,[9] with 70-80% of cases occurring on the scalp, however, lesions could also involve the face, trunk or limbs, sometimes in symmetrical patterns[3,5,10]. There are few case reports of ACC from Nigeria with most involving either the limbs or the scalp and none on truncal involvement. This report, being the first from Nigeria involving truncal ACC, aims to create awareness of a rare presentation of this rare disease condition and highlight a mode of conservative management.

Case report

Baby O, a female infant from the Yoruba tribe delivered at term via spontaneous vaginal delivery at a traditional birth attendant home to a 21-year-old primigravida mother. The child was said to have cried well at birth. She was brought in at the 1st hour of life because of skin lesions noticed at birth. Pregnancy was uneventful and the mother was regular with antenatal care medications and visits. Mother denied the use of any orthodox medication or herbal preparations. The antenatal ultrasound scan during the second trimester was reported as normal. She is the first child in a monogamous family setting. There is no history of congenital anomalies among family members. Mother is a 21-year-old fashion designer and father is a bricklayer.

Physical examination revealed a conscious and irritable female neonate, pink in room air, with a normal temperature of 37.1℃, anicteric, not pale, not dehydrated, and no oedema. The heart rate was 150 beats per minute with first and second heart sounds, respiratory rate was 60 cycles per minute with broncho-vesicular breath sounds. She had symmetrically distributed and well-demarcated skin defects over the abdominal region each measuring about 5 x 3 cm that were irregularly shaped. Hyperpigmented scars surrounded these ulcers. There was no discharge, bleeding or bullae from the ulcers. The child weighed 2.790kg; occipito-frontal circumference was 33cm and chest circumference was 32cm. Other systemic examinations were essentially normal. A diagnosis of aplasia cutis congenita was made.

Blood sample was taken for complete blood count which revealed a packed cell volume of 51%, white blood cell count of 15.6 x 109/L (neutrophils: 56%, lymphocytes: 34% and monocytes: 10%)s, Platelets: 160 x 109/L. The child was commenced on intravenous 10% dextrose water (which was discontinued the next day) and put to breast, intravenous ceftriaxone 140mg daily, intravenous gentamicin 7mg 12 hourly, syrup vitamin C 2.5mls twice daily, wound cleaning and dressing with normal saline and mupirocin cream plus honey respectively three times daily and the parents were reassured. She has responded well to treatment with no signs of infection and the wound granulated well. By the 8th day of admission, defect had almost completely healed leaving a hypo-pigmented scar. At two weeks on follow-up the scar had become hyper-pigmented and stable.

Fig 1a: The neonate at presentation showing skin defects over the right side of the trunk

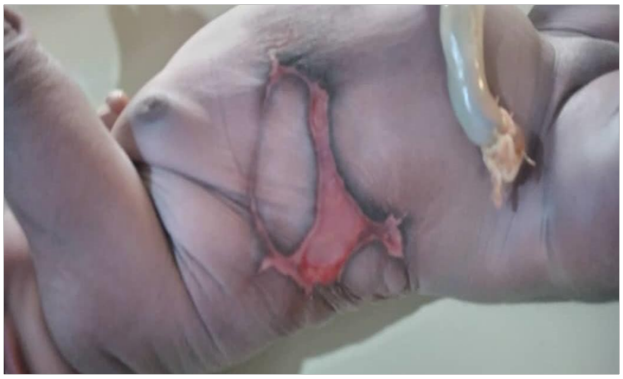


Fig 1b: The neonate at presentation showing skin defects over the left side of the trunk

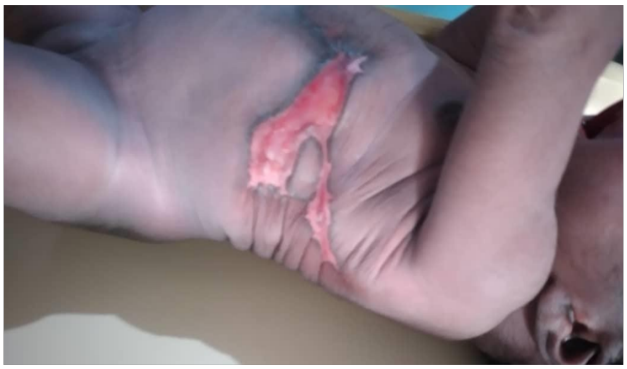


Fig 2a: The skin defects over the right side of the trunk on the 5th day of admission

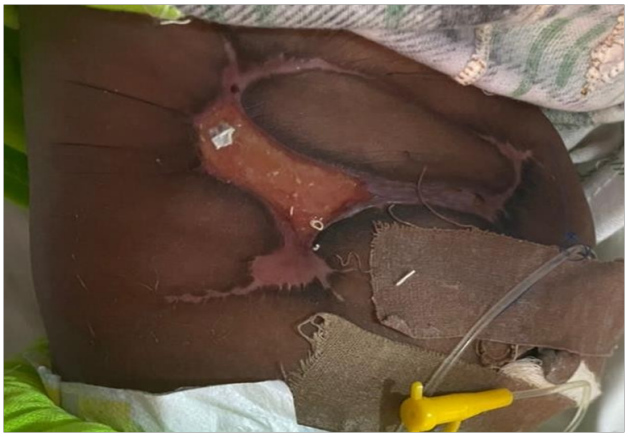
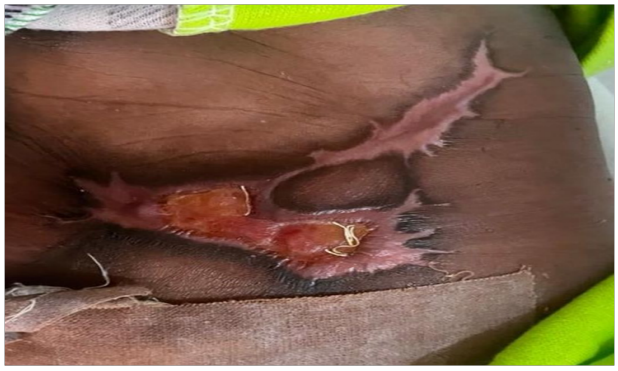


Fig 2b: The skin defects over the left side of the trunk on the 5th day of admission



Discussion

Aplasia Cutis Congenita: Overview and Case Analysis

Aplasia Cutis Congenita (ACC) is a rare congenital condition characterized by the absence of a portion of skin in localized or widespread areas at birth. The etiology of ACC remains largely unclear, but it is believed to involve genetic, environmental, and possibly teratogenic factors. The case of Baby O provides a poignant example of ACC, highlighting several important aspects related to its presentation, diagnosis, and management.

Epidemiology and Etiology

ACC can present as an isolated skin defect or in association with other anomalies. The condition occurs in approximately 1 in 10,000 live births. The etiology of ACC is multifactorial. It can result from genetic mutations, intrauterine trauma, vascular compromise, teratogenic exposures, or amniotic band syndrome. In Baby O's case, the mother's pregnancy was uneventful, with no exposure to harmful substances or significant familial history of congenital anomalies, suggesting a potentially sporadic occurrence of ACC.

Clinical Presentation

When non-scalp locations are involved, they are often bilaterally symmetrical.[11] Baby O presented with well-demarcated skin defects over the abdominal region that were bilaterally symmetrical, a classic presentation of ACC. The lesions were surrounded by hyper-pigmented scars, indicative of prior healing attempts. There were no associated anomalies on clinical examination. The absence of systemic symptoms and normal systemic examination findings, except for the skin defect, aligns with typical isolated ACC. The size and distribution of the lesions can vary, but they are often symmetrically distributed, as observed in Baby O.

Diagnosis

Diagnosis of ACC is primarily clinical, based on the appearance of the skin lesions at birth. In this case, the symmetrical and well-demarcated nature of the ulcers, along with the surrounding hyperpigmentation, supported the diagnosis of ACC. Additional laboratory investigations, such as the complete blood count performed for Baby O, are essential to rule out underlying infections or hematological abnormalities, which can complicate the condition. Baby O's laboratory results were within normal ranges, further confirming the isolated nature of her ACC.

Management

The management of ACC can be conservative or surgical depending on the size and location of defect and the presentation. The conservative management of ACC focuses on preventing infection, promoting wound healing, and providing supportive care. In Baby O's case, intravenous antibiotics (ceftriaxone and gentamicin) were initiated to prevent secondary infections, a common and serious complication of ACC. Topical wound care with normal saline, mupirocin cream, and honey was employed to promote healing. The use of honey, known for its antibacterial and wound healing properties, is particularly noteworthy and reflects a blend of modern and traditional therapeutic approaches. Additionally, Vitamin C supplementation was given to support overall health and wound healing.

The importance of regular wound cleaning and dressing cannot be overstated, as it reduces the risk of infection and facilitates granulation tissue formation. Baby O responded well to the treatment, with no signs of infection and good wound granulation, demonstrating the effectiveness of the management plan.

Prognosis and Long-term Care

The prognosis for infants with ACC is generally good if the condition is isolated and managed appropriately. Potential complications include secondary infections, scarring, and, in rare cases, more extensive involvement of underlying structures such as bone or dura. Regular follow-up is essential to monitor for complications and ensure proper wound healing. In Baby O's case, continued monitoring and parental education on wound care will be crucial to her long-term health and well-being.

Conclusion

This index case highlights the successful management of ACC through prompt diagnosis, appropriate antimicrobial therapy, and meticulous wound care and close monitoring. It underscores the importance of a multidisciplinary approach in managing congenital skin conditions and the potential for blending traditional and modern treatments to optimize patient outcomes. Further research into the etiology and best practices for managing ACC will continue to improve care for affected infants.

Contributions to Author
Dr Agelebe Efeturi: Concept, design, definition of intellectual content, literature search, data acquisition, manuscript preparation, manuscript editing and manuscript review
Dr Arinde Adebola V: Concept, design, definition of intellectual content, literature search, manuscript preparation, manuscript editing and manuscript review
Dr Odeyemi Abimbola O (guarantor): Concept, definition of intellectual content, literature search, manuscript preparation, manuscript editing and manuscript review.
Dr Odafen Oseiga P: Concept, definition of intellectual content, data acquisition, manuscript preparation and manuscript review.
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