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CASE REPORT

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Aplasia cutis congenita in a Nigerian child: A case report

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Abstract*:* Aplasia cutis congenita (ACC) is a rare skin disorder, the cause is not known but intrauterine infections, drugs, chromosomal and genetic disorders, vascular compromise and trauma have been implicated. Clinically the diagnosis is made based on physical findings indicative of intrauterine disruption of skin development. We present an eighteen hours old neonate with Aplasia cutis congenita, this is aimed at creating awareness in view of the rarity of this condition. Conservative treatment of the ulcers has yielded excellent result but not without complication of acquired syndactyly, the child is being followed up for possible surgery to release the digits.

Key words: Aplasia cutis congenita, Epidermolysis bullosa, Neonate, Honey dressing, Frieden classification, Nigeria

Introduction

Aplasia cutis congenita is a rare congenital developmental skin defects, characterized by well demarcated, oval or circular ulcers or scars.[1] It could be in a localized area or widespread at birth. The condition is most commonly seen on the scalp in 90% of cases,[2] but ACC can affect any part of the body.[2, 3] Aplasia cutis congenita is primarily a clinical diagnosis with no specific histological alterations. At birth most cases of ACC have ulcerated lesions which may show total absence of skin, sometimes extending to the bone or dura.[3] Aplasia cutis congenita can be associated with other physical abnormalities or malformation syndromes, chromosomal or other disorders such as ectodermal dysplasia and epidermolysis bullosa.[4]

Frieden created a classification system consisting of nine groups based on the number and location of the lesions and the presence of associated malformations.[4] In Frieden's classification group 1: This was a scalp ACC without multiple anomalies, a collar of hair was often seen around the defect. This can be autosomal or sporadic in occurrence. Group 2; was a scalp involvement with limb anomalies usually lower limbs with asymmetric lesions. Group 3; was a scalp ACC with epidermal and sebaceous nevi. Some patients reported in this group had ophthalmic and neurologic findings typical of epidermal nevus syndrome. Group 4; ACC had hair collar overlying deeper embryologic malformation. Examples were meningomyelocele, leptomeningeal angiomatosis, porencephaly, gastroschesis etc. In group 5: This was ACC associated with fetus papyraceous, while in group 6 ACC was associated with siplex, junctional or dystrophic types of epidermolysis bullosa. Group 7: This was ACC localized to the extremities without epidermolysis bullosa. Group 8; this was ACC specifically due to teratogens. Lastly in group 9: This was ACC associated with malformation syndromes such as Down syndrome, Patau syndrome etc.

The exact pathophysiological mechanism of this disorder is not clear but propositions included; intrauterine trauma, vascular compromise, maternal infections and medications. Aplasia cutis congenita is typically sporadic but autosomal dominant and less commonly autosomal recessive cases have been reported. Globally the estimated incidence of ACC is 3 in 10,000 live births.[2,4] If the defects is small recovery is usually uneventful, with gradual epithelization and formation of hairless, atrophic scar over several weeks. Sometimes surgical intervention may be necessary. The sex distribution is equal except if it is associated with an X-linked malformation syndrome.

The treatments of ACC depend primarily on size, dept and location of the defect and therapy of associated disorders.[5] Variety of dressings both adhesive and non adhesive materials have been used.[6, 7] Surgical repairs of large defects can be done.

This paper present Aplasia cutis congenita; with sole aim of creating awareness of this rare but treatable condition with good outcome depending on the size of the lesion.

Case Report

MBM, 18 hours old product of term female neonate was admitted into the Neonatology Unit of the Bingham University Teaching Hospital Jos on 13th Jan 2015. The mother was a 23 year old lady whose husband was a 30 year local security guard. The pregnancy was uneventful, supervised and delivered in a Plateau State Specialist Hospital Jos. The baby was referred to us when the lesions were noticed at birth. There was no family history of congenital malformation.

Examination revealed symmetrically distributed ulcers involving both upper and lower limbs. There were no

bullae, discharges, or bleeding. The baby was afebrile temperature of 36.7°C. The baby weighed 2.4kg; the length and the occipito-frontal circumference were within normal limits. The ulcers covering 5% of the total body surface area on each hand, while it covered 10% and 9% on both the right and left leg respectively (see figure bellow). There was no evidence of inflammation. Other systems examination was essentially normal. A diagnosis of aplasia cutis congenita was made with a differencial diagnosis of epidermolysis bullosa.

The patient's packed cell volume was 46%, WBC was 10×10^9/L with 68% neutrophils and 32% lymphocytes, the VDRL was non reactive and the HIV serology was negative. The blood culture yielded no bacterial growth and the wound swab culture was also negative. The serum electrolytes were within normal limits. Patient was commenced on 1/5 normal saline in 8.4% dextrose, intravenous cefuroxime 120mg 12 hourly, intramuscular gentamycin 6mg 12 hourly and a daily wound dressing with honey, she responded well to these treatments and within six weeks the wound had healed except for a complication of acquired syndactyly. Surgical intervention to release the digits is being planned.

Fig 1: Ulcers at presentation

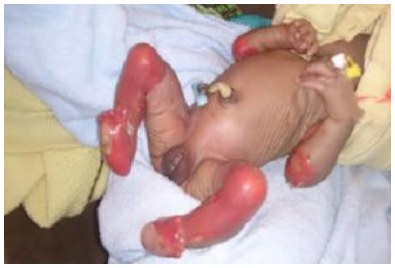
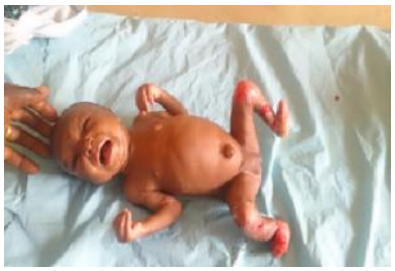


Fig 2: After 6 weeks of treatment



Discussion

Aplasia cutis congenital first reported by Campell in 1826,[8] is a rare disorder. Following this report there have been few cases reported in literature most of which were outside Africa. This condition is most often seen on the scalp in 90% cases.[2, 3] The index child's lesions affected the extremities making it one of the rare group of ACC. At birth most cases of ACC have ulcerated lesions which may show total absence of skin, sometimes extending to the bone or dura.[3] The later did not apply to the index infant, because lesion did not affect the bones; neither was there any lesion on the scalp.

The index child has no other associated malformation defects; this therefore put her in group 7 of the Frieden's classification.[4] In this group, neonates have ACC confined to the extremities without obvious malformation or epidermolysis bullosa. This particular type is said to be very rare, however there was a report of two families with multiple members having ACC on the pretibial lower extremities and the dorsal aspects of the hand and the feet,[9]these reports described features similar to the index child. (See figure).

The index infant has no history suggestive of fetus papyraceus or placental infarcts as the placenta was reported to be normal. In group 8 of Frieden classification of ACC teratogens are known causes. These had also been linked in few cases to intrauterine infections with herpes virus, varicella zoster virus or exposure to methimazole.[10] The mother of this child had no history suggestive of viral infections in pregnancy neither was she exposed to medications apart from the routine antenatal drugs. Ercan et al[1] reported 3 cases of ACC, the first one was linked to intrauterine infection (rubella), the second case was linked to trisomy 13 syndromes and the third case was linked to fetal valproate syndrome. None of these factors appeared to be relevant in our infant.

Pathologically the lesions in ACC are non inflammatory and well demarcated as was the case in the index infant. Laboratory investigations in the index infant were all within normal. This is not surprising; as it has been well documented in the literature that there were no specific laboratory abnormalities that were consistently found in this condition.[2, 3, 4] Elevated alpha feto protein in maternal serum and amniotic fluid as well as elevated acetylcholinestrases in amniotic fluid had been tried in the past, as biochemical markers but they have been abandoned because they lack sensitivity and specificity.[11]

The index child responded very well to medical treatment, within six weeks the wound were virtually completely healed. The decision to use medical, surgical or both modes of therapy depends primarily on size, dept, location of the defects and therapy of associated abnormalities.[5] This child had occlusive honey dressing, but in large defects ACC surgical repair including excision with primary closure, use of tissue expanders and rotation of flap to fill defects, skin and bone grafting may be required.

The prognosis in this child was excellent. This was in conformity with the usual excellent prognosis occurring in groups 7 of the Frieden classification to which this child belongs. But we are mindful of anticipated complications, such as large scars, contracture and acquired syndactyly. The index child had developed acquired syndactyly (See fig 1B). In other types of ACC the prognosis is dependent on the severity of the lesion and other congenital abnormalities.

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