

## CASE REPORT

### Failed conservative management of a case of aplasia cutis congenita in a low-income country

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## Introduction

Aplasia cutis congenita (ACC) is a rare condition characterized by the congenital absence of all skin layers [1]. The first description of ACC dates back to 1767 by Cordon [2]; since then, more than 500 cases have been reported in the literature. Most commonly, ACC presents as a localized solitary lesion involving the vertex of the scalp. However, it may affect larger areas of the face, trunk, buttocks, and extremities. The reported incidence ranges from 0.5 to 2.8/10,000 newborns, with a male:female ratio of 1:7 [3, 4]. Aplasia cutis congenita is usually an isolated clinical finding, but it can also occur in several syndromes, and different classifications based on etiology and/or presentation have been so far proposed [5, 6]. The

#### Key Clinical Message

Aplasia cutis congenita is a rare disease characterized by absence of skin layers. Usually the scalp is affected, but the whole body can be involved. We report extensive aplasia of a baby born of a HIV-positive mother taking antiretroviral drugs. Conservative treatment was not enough to ensure her survival.

#### Keywords

Aplasia cutis congenita, dermatology, pediatrics.

management of ACC in a newborn with a widespread skin defect presents a dilemma, since best treatment has not yet been determined. Surgical reconstruction of the skin with and without grafts, as well as conservative cures has been attempted, with different outcomes [7–10]. We report a challenging case of severe ACC conservatively treated in a low-income country.

## Case Report

A newborn female was presented to us, in Beira Central Hospital (Mozambique), after a few hours of birth at home (Apgar score unknown). Fifth-born child, born at term, from a 28-year-old HIV-positive mother, in therapy for 2 years and treated with the following antiretroviral

drugs: Tenofovir 300 mg, Lamivudina 300 mg, and Efavirenz 600 mg. The child's birth weight was 1480 g, her body length 47 cm, and her head circumference 33 cm. Despite a rosy complexion, the crying sound, a good tone, and responsiveness, her general conditions were compromised. She showed extensive discontinuity of the skin on the scalp, trunk, abdomen, limbs, and bilaterally on the extremities for more than 60% of the body surface. In these areas, a thin, shiny, and diaphanous membrane was present, and a network of small vessels was visible. Other abnormalities and dysmorphic features were not recognized. In the absence of diagnostic tests, ACC was clinically diagnosed (Fig. 1).

Systemic intravenous treatment was as follows: cefotaxime 70 mg for 12 h, gentamicin 6 mg for 1 day, and azidothymidine 0.5 mL every 12 h. The only pain therapy available was paracetamol, 15 mg every 8 h. Skin lesions were treated with silver sulfadiazine dressing. For the first 5 days, she was able to breastfeed, she had no fever or other complications, and we registered a weight loss of 180 g. Despite recording attempt to cicatrization, areas of necrosis with scattered leak fluid remained. From the 6th to the 10th day, her general conditions worsened: her body temperature reached 38°C, necrotic areas increased and began to tear. On the 15th day, she weighed 1010 g, showed extensive areas of tissue necrosis, desquamation of healed areas, distended abdomen, inability to feed, and irritability. Intravenous vancomycin was administered (with a first administration of 15 mg, and then of 10 mg every 12 h), as

well as cefepime (30 mg every 12 h), and fluconazole (6 mg every 72 h), still in association with silver sulfadiazine dressing. General conditions continued worsening, her weight decreased to 950 g, and fistulas appeared in abdominal necrotic areas. On the 24th day, she died.

## Discussion

Aplasia cutis congenita is a multifactorial skin alteration usually presenting as an isolated small lesion at the scalp. In the case reported here, however, we faced an extensive congenital malformation. The diagnosis is mainly based on the clinical presentation and on histological demonstration of complete lack of skin. Some authors suggested the following tests as complement: maternal and fetal alpha-fetoprotein and acetylcholinesterase dosage as prenatal screening, as well as ultrasound, X-ray, and magnetic resonance imaging (MRI) as postnatal tests [11]. In the absence of other inherited abnormalities, prenatal tests, information about the placenta, or about a possible *fetus papyraceus*, we hypothesize a teratogenic role of maternal HIV infection and/or long-standing antiretroviral treatment in the pathogenesis of the disease. Indeed, drug consumption (e.g., methimazole, valproic acid, misoprostol, marijuana, and cocaine) during pregnancy is a well-known factor associated with ACC. Virus transmission (e.g., herpes simplex virus or varicella zoster virus) from the mother to the fetus is another recognized teratogen. These may cause placental alterations affecting the



**Figure 1.** (A) At birth – Characteristic scalp lesion. (B) At birth – Extensive skin discontinuity on the trunk, abdomen, limbs, and extremities. (C and D) At 18 days. Extensive areas of tissue necrosis and desquamation of healed areas.

normal skin development in the fetus. Unfortunately, it is not possible neither to confirm nor to refute our hypothesis and, in the future, only a proper follow-up of the mother during pregnancy – of particular importance in low-income countries with high HIV prevalence – will allow us to clarify the issues hypothesized above, as well as all other related issues.

The correct management and treatment of ACC is still debated. Both surgical and conservative approaches have been attempted in order to achieve a complete closure of the skin defect, while avoiding complications such as meningitis, hemorrhages, trauma, and thrombosis [12, 13]. The management of ACC presenting extensive lesions is even more difficult in low-income countries due to the absence of specialized clinicians, diagnostic tools, and, frequently, medicaments.

In our case, appropriate diagnostic tests were not available. However, we can state with of ACC certainty that, due to its characteristic presentation and because only few other conditions, we can rule out the following, presenting with ulcerations or scars in the newborn period: epidermolysis bullosa, focal dermal hypoplasia syndrome, and nevus sebaceous. Lacking even a plastic surgeon, conservative medical therapy was the only empirical treatment provided. Although contraindicated in newborns, we used silver sulfadiazine dressing as reported in several papers [12, 14, 15]. Moreover, we administered systemic antibiotic therapy – another medication that is not appropriate in the management of large skin defects, mainly because it may cause the emergence of resistant bacteria – to prevent infection and sepsis. The only available analgesic treatment, albeit possibly inappropriate and certainly inadequate, was paracetamol.

Considering the bad prognostic factors, this case would be difficult to treat even in high-income countries, particularly more so when taking into account the possible complications that may occur during treatment, such as infection and sepsis, fluid leakage, weight loss, electrolyte imbalance, bleeding, and pain. Therefore, and very sadly, failure of successfully treating this case of ACC, which presented extensive lesions in a setting with limited therapeutic options, was almost a foregone conclusion. This case, once again, points out that it is mandatory to improve maternal and child health in countries with limited resources in order to guarantee the possibility of a more appropriate therapeutic approach while at the same time allowing research progress.

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## Conflict of Interest

No author has any conflict of interest.

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