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# Conservative Treatment for Single Wound due to Aplasia Cutis Congenita Type IV ( Bart's Syndrome) in the Lower Limb: Case Report

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## Abstract

The Aplasia Cutis Congenita (ACC) is a rare malformation, and it does characterize by the absence of skin layers and subcutaneous cellular tissue. It occurs most frequently in the scalp region, but can affect any anatomical region; the incidence is in between 0.5 to 1 of 10,000 live births being more common in female gender.

The etiology may be associated with Epidermolysis Bullosa (EB), intrauterine infections, ectodermal dysplasia; it may be associated with chromosomal anomalies such as trisomy 13, and also with the autosomal recessive and dominant inheritance in 35% to 50% of cases.

To report the case of a patient with ACC in the lower limbs treated exclusively with biological dressing.

**Keywords: Aplasia; Cutis anomaly; Epidermolysis bullosa**

## Introduction

The Aplasia Cutis Congenita (ACC) is a rare malformation, and it does characterize by the absence of skin layers and subcutaneous cellular tissue [1]. It occurs most frequently in the scalp region, but can affect any anatomical region; the incidence is in between 0.5 to 1 of 10,000 live births being more common in female gender [2].

The etiology may be associated with Epidermolysis Bullosa (EB), intrauterine infections, ectodermal dysplasia [3], it may be associated with chromosomal anomalies such as trisomy 13, and also with the autosomal recessive and dominant inheritance in 35% to 50% of cases [1].

The diagnosis is clinical and its characterize by large tissues defects in the skin of the newborn, the treatment is usually conservative, however there are records of surgical treatments, such as autologous partial skin grafting, this conduct is recommended specially when this malformation consists of total absence of skin and subcutaneous cellular tissue [4,5].

## Objective

To report the case of a patient with ACC in the lower limbs treated exclusively with biological dressing.

## Case Presentation

Male newborn at 39 weeks gestation. Mother 28 years old, Gestation II and Para II, vaginal deliveries. She had done prenatal care, had not taken any vitamin or mineral supplements. She reported episode of urinary tract infection during the gestation period that have being treated with first generation cephalosporin. She had made the use of paracetamol as an analgesic. She denies the use of any other medication, exposure to teratogens, and family history of skin diseases or chromosomal syndromes.

There have been no intercurrences during the childbirth, and the placenta had no visible changes. At the birth the child received an Apgar score of 9/10, and on physical examination have presented areas of absent skin on the left lower limb, beginning at the calf and extending to the pododactyles (Figure 1), a small area on the scalp without total loss of dermis and on the right hand a small area of necrosis on the third chirodactyl.

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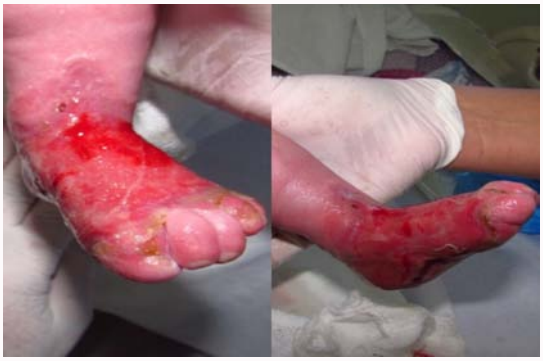

**Figure 1:** Initial wound.

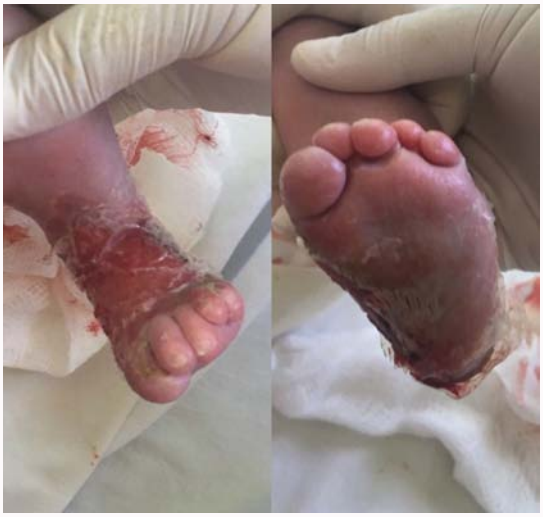

**Figure 2:** Fifteen days post treatment.

During the first 10 days, have proceeded with daily dressing with Dersani and gaze rayon. Diet only with maternal milk. The patient had no other clinical abnormalities, complete blood count and urine type I within normal range.

According to Frieden's classification (Table 1), the case was diagnosed as ACC associated with Epidermolysis Bullosa (EB) (subgroup 6) (Figure 2). To accelerate the healing process and prevent wound infection, have chosen the use of the laminar biological dressing Omiderm® on the 11th day of birth, and patient was discharged. The patient has returned to the outpatient clinic for 30 days, twice a week, when have proceeded the changes of dressing with Omiderm (Figure 3).

**Table 1:** ACC Frieden's classification.

| TYPE | Feature |
|---|---|
| TYPE 1 | ACC on scalp without multiple abnormalities. |
| TYPE 2 | Scalp associated with limb abnormality (Adams–Oliver syndrome). |
| TYPE 3 | ACC associated with epidermis or organoid nevus. |
| TYPE 4 | ACC superimposed on embryological malformation such as meningomyelocele, gastrochisis or emphalocele. |
| TYPE 5 | ACC associated with papyraceous fetus or placental infarction. |
| TYPE 6 | ACC associated with bullous epidermis. |
| TYPE 7 | ACC located at the edges without bubbles. |
| TYPE 8 | ACC caused by specific teratogens. |
| TYPE 9 | ACC associated with malformation syndromes. |

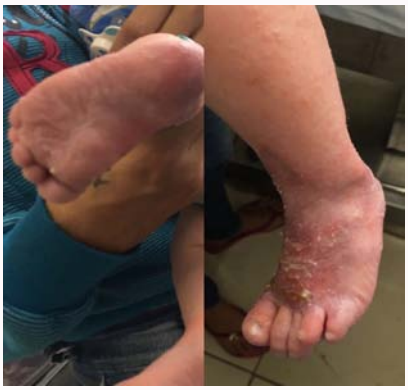

**Figure 3:** 40 days Post treatment.

## Discussion

Current evidences suggest associations of ACC with some etiological factors such as: exposure to maternal smoking [6], drugs such as methimazole, diclofenac sodium, valproic acid, and drugs such as cannabis sativa and cocaine [7]. The clinical aspect of ACC lesions is heterogeneous. In some cases it may present in the early stages of gestation and then heal as a fibrous and atrophic scar at birth [8]. Lesions that remain until birth present as erythematous, moist, granulation tissue ulcers covered by thin membranes. The depth of the lesions may affect all layers of the skin with decreased subcutaneous cellular tissue, and may affect muscle and bone [9].

The diagnosis is clinical; generally the lesions are well delimited, non-inflammatory, 0.5 cm to 10 cm in extension, and may be larger [10,11]. The prognosis is good in the medium and long term [12,13], mortality (20% to 50%) is proportional to the extension of these lesions [14].

According to statistical data, the involvement of the lower limbs occurs in 25% of all cases of aplasia cutis congenita; 12% affect the trunk and flanks, being that the highest incidence occur in the scalp, with about 60% of the cases [15].

The non-cranial ACC disease has an incidence of 17% to 30% of cases [8]. The association with Epidermolysis Bullosa (EB) is rare [16]. Nevertheless, in this study ACC was associated with EB characterizing Bart's Syndrome (BS) [17].

The Bart's syndrome is a congenital cutaneous aplasia and epidermolysis bullosa, and may be associated with nail abnormalities such as congenital absence, ungual dystrophy or additional loss. Its etiology as well as its pathophysiology are still controversial, mainly because it is considered an extremely rare genetic disorder [17,18].

The Bart's syndrome was first described in a family with congenital absence of skin on the lower limb, vesicles spread to other regions of the skin, and atrophy of the nails [17,18].

In this context, lesions are almost always unilateral and involve the dorsal and medial surface of the limbs. They appear on the extremities with intense red ulceration extending upward from the dorsal and medial surface of the foot to the calcaneus [18].

It is worth noting that in severe cases of BS, especially those associated with junctional epidermolysis bullosa, the patient may have other congenital anomalies such as atresia, urethral stenosis, rudimentary ear development, flattened nose, kidney abnormality, staring eyes, and wide nasal root [18,19]. In this case, there was no

other malformation associated.

Most lesions heal spontaneously with conservative treatment, but very large and deep wounds that affect the basal cells of the skin may need cultured epidermal grafts when the wound has basal cells [20]. The treatment aims to accelerate epithelialization when there are basal cells in the skin and decrease the risk of atrophic scarring, hemorrhage, infection, the response to this treatment will depend very much on the patient [21]. Some authors recommend early surgical treatment [22]. A conservative treatment plan provides protection against infection keeps the wound moist and provides good conditions for healing by secondary intention [23].

The conservative treatment as well as surgical treatment has similar risks, but conservative treatment is recommended as the first option, making use of dressings, thus trying to reduce possible surgery complications [24].

Dressing options for conservative treatment should include petrolatum-derived oils, silver sulfadiazine, or Bacitracin 3 times a day. The petrolatum is considered the best option because of its low risk of allergy and systemic absorption [2].

The wounds epithelialized 100% in this case and were treated conservatively and on an outpatient basis without requiring further hospital admission. Outpatient treatment decreased the risk of infection by resistant microorganisms and maintained exclusive breast milk feeding.

The patient's prognosis is very well until complete healing and the evaluation of possible retractions and consequent difficulty in moving the affected limb will be analyzed as soon as the patient starts walking.

## Conclusion

ACC associated with epidermolysis bullosa is one of the less common forms of presentation of this disease. Conservative treatment with biologic dressing 3 times a day, recommended preventing infection, speed healing, and decreases the risk of bleeding and scar retraction.

Outpatient follow-up should be continued until a more adult age. At the moment, there are few studies related to the treatment of this pathology, leaving it up to define the therapeutic strategy.

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