

Bart syndrome: A case report

Renata Teles Albernaz¹, Adriana Prazeres da Silva²

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Abstract

Bart's syndrome is a genetic disorder characterized by the absence of localized skin (present at birth), epidermolysis bullosa (EB) and ungual changes. It is an autosomal dominant inheritance, caused by mutations in the type VII collagen gene on chromosome 3p. It is described the case of a female patient who expressed these characteristics at birth. In the delivery room, it was observed that there was no skin in the lower limbs, and the next day, intact blisters and routes oral mucosa, and the absence of some nails. The diagnosis is clinical, so it has been confirmed through the typical characteristics; laboratory confirmation was not possible due to lack of integrity bubbles for biopsy and direct immunofluorescence, while monitoring the patient. The treatment, which in most cases involves conservative measures, was performed with daily bandages in the lower limbs, fluid removal from some bubbles and skin and mucous care, avoiding trauma. It was observed a significant improvement of all injuries and the patient currently has been in an outpatient follow-up with a pediatric dermatologist with cicatricial lesions on the lower limbs and without bullous lesions.

¹ Physician - Pediatric Resident. Hospital Regional de Mato Grosso do Sul (HRMS), Campo Grande, MS, Brazil.

² Pediatric Dermatologist (HC-UFPR) - Preceptor of the medical residency in pediatrics program of the HRMS, Campo Grande, MS, Brazil.

Correspondence to:

Adriana Prazeres da Silva.

Hospital Regional de Mato Grosso do Sul (HRMS). Avenida Nelly Martins, nº 1838, Apartamento 1204, Bairro Carandá Bosque, Campo Grande, MS, Brasil. CEP: 79032-295.

INTRODUCTION

In 1966, Bart et al. reported the case of 26 members of a family who had congenital absence of skin in the lower limbs, cutaneous mucous blisters, and absence or deformity of the nails. This unique association became known as Bart's syndrome¹.

Currently considered to be a genetic disease,² Bart's syndrome is an autosomal dominant inheritance caused by mutations in the type VII collagen gene on chromosome 3p³.

Aplasia cutis may be unilateral or bilateral and is identified at birth. The disease tends to occur on the parts of the body that are subjected to rubbing and trauma (feet, hands, arms, legs, and oral cavity),¹ and may be linked to any subtype of epidermolysis bullosa (EB): simplex, junctional, or dystrophic; it is more often associated with dominant dystrophic EB⁴.

Despite being a rare hereditary bullous skin disorder, one of the lesser known manifestations of EB,³ Bart's syndrome appears to have a distinct and clear clinical Figure and has a favorable prognosis; its spontaneous progressive improvement emphasizes the importance of early diagnosis and conservative treatment⁵.

CASE REPORT

EVL, a female born by cesarean section at 39 weeks of gestation, weighing 2,740 grams, Apgar 8/9, without the need for neonatal resuscitation.

The mother 24 years old; she has attended all prenatal appointments, with no reported baseline conditions; only the first phase of serology was performed, evidencing susceptibility to toxoplasmosis, rubella, and cytomegalovirus.

Oligohydramnios was reported at 25 weeks of gestational age, but a subsequent ultrasound detected a normal amount of amniotic fluid and no malformations in the fetus.

She is the second child of non-consanguineous parents, born in Campo Grande, MS, Brazil. The first child was born with omphalocele and other unspecified malformations, and died 2 h after birth.

From birth, the patient presented an extensive area with no skin on the lower limbs, extending from the knee to the toes, mainly following medial parts; aplasia cutis was diagnosed by a pediatric dermatologist (Figure 1).

On the second day of life, nail abnormalities were observed: absence of some toenails and presence of intact bullous lesions on the lips and oral mucosa, which burst at minor friction (Figure 1).

The provisional diagnosis was Bart's syndrome because of the triad-aplasia cutis, nail abnormalities, and epidermolysis bullosa. To complement the investigation, abdominal and transfontanelar ultrasound, echocardiogram, and ocular fundus were performed, which showed no abnormalities. During follow-up, no intact blisters emerged; therefore, it was not possible to perform biopsy and direct immunofluorescence.



Figure 1. Aplasia cutis, absence of nails, and burst blister in oral mucosa.

The proposed treatment was conservative, using gauze soaked in oil with essential fatty acids; the dressings were changed daily by a nurse with experience in dressing wounds. The mother was taught how to do the procedure and currently does so at home, but less frequently due to the almost complete healing (Figure 2).



Figure 2. Evolution of injuries.

The intensivist at the neonatal unit opted for antibiotic use; initially, oral antibiotics were administered in order to avoid using a venous access, to prevent unnecessary trauma. However, during hospitalization, the patient developed electrolyte disturbance due to excess insensitve loss and low intake due to sucking difficulty caused by the bullous lesions in the oral mucosa, requiring intravenous hydroelectrolytic replacement. At that moment, as a peripheral venous access had been arranged, intravenous antibiotic was administered; furthermore, the laboratory tests indicated worsening of the infection when compared with admission.

The child is currently 3 months old; she has no blisters, and the lower limb injuries are fully healed. Care is being taken by the parents to avoid trauma and friction, and the child attends monthly follow-up consultations with a pediatric dermatologist.

DISCUSSION

Bart's syndrome, although rare, should be diagnosed early, so that the most appropriate treatment can be established in order to prevent complications such as local infection, sepsis, bleeding, excessive fluid loss due to the injuries, hypothermia, electrolyte disturbances and, later, hypertrophic and atrophic scars⁶.

In the present case, the diagnosis was early, but the patient developed electrolyte disturbances due to losses, especially of bloody fluid, during the dressing, which even led to anemia and required transfusion of red blood cells.

It is known that this is an autosomal dominant inheritance caused by mutations in the type VII collagen gene on chromosome 3p³. However, its pathomechanism has not been fully elucidated. Some authors propose that it is caused by intrauterine physical trauma, while others argue that the affected area follows Blaschko's lines, which are metamereric segments such as dermatomes that direct the growth of skin cell clones⁷. In the present case, the aplasia manifested bilaterally in the lower limbs, which would be compatible with both theories.

The diagnosis is clinical, based on sign and symptoms that are exclusive to the disease, i.e., localized skin absence (present at birth), EB, and nail alterations¹. Nonetheless, confirmation is obtained by a histological evaluation of the skin with immunofluorescence and/or electron microscopy to classify the subtype of EB⁸. The present case was not laboratory confirmed, as there were no injuries with intact blisters for biopsy.

Immunofluorescence may show specific antibodies to antigens and type VII collagen³. Electron microscopy characterizes the ultrastructure of the skin at the level of the dermal-epidermal junction⁸. In the case reported by Kim et al., electron microscopy showed evidence of separation of the dermal-epidermal junction and disruption of the basal lamina, similar to what is observed in junctional EB. Genetic testing is

the most accurate method for analyzing EB and may provide a definitive diagnosis of its type and subtype; however, it is expensive and seldom performed⁸.

The management of this syndrome involves conservative measures for treatment of wounds and a multidisciplinary approach (pediatric dermatologist, pediatrician, nurse with experience in treating wounds, and speech therapist, among other professionals). The administration of systemic antibiotics is not routine, but can be used if an infection is suspected⁸. In the present case, the authors opted for treatment with systemic antibiotics; but in other reported cases, such as that of Kim et al., topical antibiotics were associated with dressings due to the lack of signs of systemic infection⁸.

In cases of blistering, fluid can be removed with a sterile needle; if necessary, mechanical debridement can be performed⁸. In the course of hospitalization, it was necessary to burst some blisters on the upper limbs, but most were located in the oral mucosa; the very action of suction caused these blisters to burst. This was one of the greatest difficulties faced in the present case.

The dressings can be initially applied with petrolatum-impregnated gauze, aiming to protect the site of trauma and decrease the risk of chafing⁸. While most patients can be treated conservatively, deep wounds may need surgical intervention with a local skin graft or flap⁸.

The reported case did not require surgery; only dressings with gauze soaked in oil with essential fatty acids were used. As an example, in the case described in Kim et al., mechanical debridement and hydrodebridement with normal saline solution were conducted; petrolatum and sterile gauze were applied to induce wound contraction and maintain hydration⁸.

CONCLUSION

This study presented a case of Bart's syndrome manifested at birth. Although rare, the syndrome must be recognized by pediatricians so that, when faced with a newborn with absence of skin, mucocutaneous blisters, and nail changes, they can start general conservative treatment measures. Nowadays, several medical treatment options are available for this condition; it is very important to minimize trauma and friction during manipulation of the patient. Non-adherent dressings that need not be changed daily should be preferred, to promote the healing process.

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