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CASE REPORT

Bart syndrome: case report and review

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Abstract

MAB, newborn male, e.t , gestational age of 37 weeks and 5 days. During his initial care, no changes were noted, except for skin lesions: cutaneous aplasia on limbs, back, inguinal region and mucous membranes, as well as bullous lesions and nail alterations. The e.t o nt the anatomopathological exam was epidermolysis bullosa. The patient had a bad e.t o nt and developed neonatal sepsis, and several antibiotic therapy and dressings were performed. After presenting n e.t o ntent clinical worsening and refractoriness to the proposed treatment, he died at 57 days of life. Barts syndrome is a genetic condition, and its diagnosis is clinical, made through the triad: cutaneous aplasia, epidermolysis bullosa and nail dystrophies. This syndrome is linked to autosomal dominant inheritance and occur e.t oto a mutation in the type VII collagen gene. Despite having a good prognosis, its diagnosis must be early, so the treatment, which is based on topical dressings and antibiotics, can be started as soon as possible to prevent possible complications such as: sepsis, hemorrhages, hydroelectrolytic disorder and death.

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INTRODUCTION

Bart Syndrome was first described in 1966 by Bart et al[1], on a study that included 26 members of a family.[2] It is defined as the coexistence of epidermolysis bullosa and aplasia cutis congenita.[3] This genetic syndrome is diagnosed when a triad of findings is present: epidermolysis bullosa, aplasia cutis congenita and nail abnormalities.[4]

Bart Syndrome differs from other causes of aplasia cutis congenita by the presence of blisters and skin fragility. Additionally, it predominantly affects the acral region.[3]

This autosomal dominant disease4 is caused by a mutation in the type VII collagen gene, located on chromosome 3.[5]

Diagnostic confirmation is achieved based on histopathology and genetic tests. Prognosis is good in the absence of other abnormalities. Early diagnosis and conservative treatment are followed by gradual spontaneous improvement.[4,6]

CASE REPORT

MAB, a male Caucasian infant born from a vaginal delivery at 37 weeks and five days weighing 2,350 g, had no alterations on physical examination other than skin lesions. A dermatologist was called to evaluate the patient for skin aplasia (Figure 1) in the arms and legs, back, oral mucosa, nose and inguinal region, in addition to bullous lesions (Figures 2 and 3) and nail abnormalities (Figure 4). A skin biopsy was performed, and a diagnosis of epidermolysis bullosa was considered. Due to the high risk of contamination, systemic antibiotic therapy was initiated with daily dressing changes. After 20 days, despite the escalation of antibiotic therapy and testing for CHAD, the patient developed sepsis. His condition deteriorated considerably, with oliguria and hyponatremia, several extubations, respiratory distress, increases in oropharyngeal lesions, and atelectatic pneumonia. He had melena, diarrhea, hematuria and vomiting. The patient was sedated and analgesia was optimized due to scabs on the limbs and ruptured blisters on the perineum. He died 57 days after birth.



Figure 1. Aplasia cutis congenita on both legs, on the day of birth.



Figure 2. Bullous lesions on the oral mucosa and face.



Figure 3. Disseminated bullous lesions and aplasia cutis congenita on the right leg.

DISCUSSION

Bart Syndrome is diagnosed based on clinical findings consisting of a triad of symptoms: aplasia cutis congenita, epidermolysis bullosa (blisters, pustules, vesicles) and nail abnormalities[7]. The syndrome is considered a variant of aplasia cutis congenita and epidermolysis bullosa. Diagnostic confirmation



Figure 4. Nail dystrophy in the fingers of the left hand.

is achieved with skin histology tests with immunofluorescence and/or electron microscopy to describe the epidermolysis bullosa subtype[1]. Histology tests were carried out in our patient and in the case described by Reddy et al[8].

This autosomal dominant condition occurs due to a mutation in the type VII collagen gene on chromosome 3[4]. However, the mechanism by which the disease develops has not been fully described. Authors still differ on whether the lesions stem from intrauterine trauma or if the affected areas follow dermatomes and, therefore, are symmetrical and bilateral[7], affecting mainly the medial portions of the legs, from the knees to the heels[3].

Early diagnosis is extremely important, so that effective treatment is promptly initiated and complications prevented[4]. Care measures include sterile gauze dressings and topical antibiotics to prevent secondary infection. In the case reported by Reddy et al[8], in which there was no need for systemic therapy, the patient was prescribed oral steroids to control the formation of new blisters and warm water and potassium permanganate to clean the lesions. Systemic antibiotic therapy may be considered if the patient is suspected with infection, combined with lesion debridement[7].

Prognosis is relatively good[7] for patients diagnosed early on and when treatment is promptly initiated. However, complications can be serious and lead to death, mainly due to sepsis, bleeding, local infection, excessive loss of fluids due to lesions, and fluid and electrolyte disorders, as in the case of our patient[4].

REFERENCES

1. Kim D, Lim H, Lim S. Bart Syndrome. Arch Plast Surg. 2015;42(2):243-5. DOI: https://doi.org/10.5999/aps.2015.42.2.243
2. Amichai B, Metzker A. Bart's syndrome. Int J Dermatol. 1994;33(3):161-3. https://doi.org/10.1111/j.1365-4362.1994.tb04939.x
3. Bolognia JL, Jorizzo, JL, Shaffer JV. Dermatologia. 3ª ed. Rio de Janeiro: Elsevier; 2015. p. 1574-80.
4. Albernaz R, Silva A. Bart syndrome: a case report. Res Pediatr. 2017 Ago;6(2):94-97. https://doi.org/10.25060/residpediatr-2016.v6n2-11
5. Christiano AM, Bart BJ, Epstein EHJ, Uitto J. Genetic basis of Bart's syndrome: a glycine substitution mutation in type VII collagen gene. J Invest Dermatol. 1996 Apr;106(4):778-80. DOI: https://doi.org/10.1111/1523-1747.ep12346304
6. Neema S, Mukherjee S, Shaw SC. Bart Syndrome. Indian Pediatr. 2019 Dec;56(12):1083.
7. Almeida NAP, Serafini F, Marchiori J, Moro JG. Você conhece esta síndrome? An Bras Dermatol. 2010 Fev;85(1):119-21. DOI: https://doi.org/10.1590/S0365-05962010000100023.
8. Reddy CU, Reddy KS, Reddy JJ. Bart Syndrome: a rare entity. Arch Oral Res. 2011 Jan/Apr;7(1):69-73. DOI: http://dx.doi.org/10.7213/aor.v7i1.23146