

Single Case

A Case of Aplasia Cutis Congenita Type VI: Bart Syndrome

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Keywords

Aplasia cutis congenita type VI · Bart syndrome · Epidermolysis bullosa · Conservative treatment

Abstract

Aplasia cutis congenita type VI, also known as Bart syndrome, is a rare genetic mechano-bullous disorder characterized by congenital localized absence of skin, mucocutaneous blistering lesions, and nail abnormalities. We present the case of a 4-h-old male newborn who presented with complete absence of skin over the anteromedial aspect of both lower legs associated with nail dystrophy since birth. After a few days, he developed blisters that were consistent with epidermolysis bullosa in histopathological examination. There was no systemic involvement such as pyloric atresia, ureteral stenosis, renal abnormalities, or arthrogryposis. All laboratory work and imaging studies were normal. A diagnosis of Bart syndrome was made based on previous presentation. We managed the patient with conservative methods. Complete epithelialization occurred after several weeks.

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Introduction

Bart syndrome is an exceedingly rare disorder [1]. It was first described in a large family almost half a century ago [2]. Bart syndrome is a genetic mechanobullous disorder charac-

terized by congenital localized absence of skin, mucocutaneous blistering lesions, and nail abnormalities, such as congenital absence or nail dystrophy [3]. The lesions of Bart syndrome are mainly unilateral and involve the medial and/or dorsal surface of the limbs. They usually appear on the limbs as sharply demarcated, glistening red ulcerations that extend upward from the dorsal and medial surface of the foot to the shin [1].

We report in this paper a rare case of Bart syndrome in a patient presenting with congenital localized absence of skin over the medial aspect of both lower legs, blistering of the skin, and nail abnormalities.

Case Report

A 4-h-old male newborn presented with complete absence of skin over the anteromedial aspect of both lower legs since birth. The baby was born to a 23-year-old primigravida mother via normal vaginal delivery. Both pregnancy and delivery were uncomplicated. The mother denied any history of exposure to medications or radiation during her pregnancy. The parents were not relatives. There was no family history of similar conditions or any skin diseases.

The baby's Apgar scores were normal, but he was admitted to the neonatal intensive care unit due to this skin lesion. Otherwise, the baby seemed to be in a good health. He had normal weight, length, head circumference, and vital signs. On physical examination, there were symmetrical bilateral absences of skin over the anteromedial aspect of the legs, starting from the knees and extending to the dorsal and medial plantar aspect of the feet. The lesions had sharply demarcated borders covered by a red ultrathin translucent membrane, and vascular structures were easily visualized (Fig. 1). Some nails of the fingers and toes were dystrophic. The scalp and the mucous membranes were not affected. Systemic examination was normal. After 5 days, the baby developed blisters over the preexisting lesions. Blistering also appeared over normal skin in response to minor trauma or friction (Fig. 2).

Several laboratory investigations were performed; the baby's complete blood count, electrolytes, as well as liver and renal function tests were within normal limits, and serologic tests for infection were negative. Ophthalmological examination, abdominal and cranial ultrasound screening, and echocardiography revealed normal findings.

A skin biopsy was taken from a fresh blister over the thigh. Histopathological study of the skin biopsy showed subepidermal blister formation, which was consistent with epidermolysis bullosa (Fig. 3). Immunofluorescence staining and electron microscopy were not available in our hospital. The combination of congenital localized absence of skin over the lower extremities, epidermolysis bullosa, and nail abnormalities led to the diagnosis of aplasia cutis congenita (ACC) type VI (Bart syndrome).

We performed conservative wound care with topical antibacterial cream (fusidic acid cream 2%) applied twice per day and nonadhesive dressing. After 1 week, the infant was discharged, and the mother was given detailed instructions about handling the baby and continuing local wound care.

The patient returned for follow-up in the outpatient dermatology clinic every 2 weeks for 2 months. At the 2-month follow-up visit, the patient was in good health and was well fed on formula without any complications. There was no difficulty in urination or defecation. The previous lesion healed with scarring and milia formation (Fig. 4, Fig. 5).

Discussion

ACC is a group of heterogeneous diseases representing failure of the skin to fully develop. Frieden [4] created a classification system for ACC consisting of nine groups based on the number and location of the lesions and the presence or absence of associated malformations. Group six of ACC is a genodermatosis characterized by a triad of clinical manifestations: ACC that presents usually over the lower extremities, any type of epidermolysis bullosa, and nail abnormalities. This clinical triad is also called Bart syndrome, first described by Bruce J. Bart in 1966 [2], who reported a family consisting of 26 members who presented with congenital absence of skin on the lower legs, widespread blistering of the skin, and nail dystrophy. Bart syndrome is considered an exceedingly rare genetic disorder. Our case of Bart syndrome presented with the classic triad of congenital localized absence of skin over both lower legs, blistering of the skin, and nail dystrophy.

In more severe cases of Bart syndrome, particularly those associated with junctional epidermolysis bullosa, the patient can present with other congenital anomalies, such as pyloric atresia, ureteral stenosis, renal abnormalities, rudimentary ear development, flattened nose, broad nasal root, and wide-set eyes [5]. However, in our case, there were no associated anomalies. The inheritance pattern of Bart syndrome appears to be autosomal dominant. Nevertheless, several sporadic cases have been reported [6]. Bart syndrome has no known underlying cause. Although various hypotheses have been proposed, both the etiology and pathophysiology of Bart syndrome are still much debated [7]. Our case was sporadic; there was neither consanguinity between the patient's parents nor any family history of a similar lesion.

Bart syndrome is usually diagnosed based on the clinical presentation. In some cases, analysis may require skin biopsy to determine the type of epidermolysis bullosa and genetic study to look for the exact gene mutation that may help to confirm the final diagnosis. In our case, the diagnosis of Bart syndrome was made based on the characteristic clinical presentation, including congenital localized absence of skin over the medial aspect of both lower legs, blistering of the skin, and nail dystrophy. Skin biopsy from a fresh blister was performed to confirm the diagnosis. Histopathological examination of the skin biopsy specimen was consistent with epidermolysis bullosa.

The management of Bart syndrome is generally conservative and includes wound care, allowing the affected area to declare itself in order to optimize future reconstruction, control of infection, and prevention and treatment of complications. Kuvat and Bozkurt [8] discussed in detail conservative treatment for patients with Bart syndrome. They used daily hydrodebridement with 1/200 diluted povidone-iodine (100 mL povidone iodine/20 L of boiled water) and fusidic acid cream; the wound was closed with dexpanthenol plus chlorhexidine-impregnated sterile gauze bandages. These conservative methods led to rapid epithelialization. A possible explanation for this rapid recovery might be that the translucent membrane, whose histopathology is nonspecific, acts like an ultrathin skin graft. These findings suggest that neither surgical nor nonsurgical methods, such as repeat Alloderm grafting or application of cultured keratinocytes, are required in many cases of Bart syndrome. In this report, we managed our patient conservatively with topical fusidic acid cream twice per day and nonadhesive dressings changed every 5 days. The recovery time in our case was similar to that in the previous literature that reported using these methods.

The prognosis of Bart syndrome depends on many factors, such as the severity and extension of ACC, epidermolysis bullosa subtype, associated anomalies, and efficacy of treatment. In general, the prognosis of patients with Bart syndrome is good [6]. They have a nor-

mal life expectancy and in most cases are able to live a normal life. However, close follow-up for serious complications such as hemorrhage, infection, hypothermia, and hypoglycemia is important. In our case, no serious complications were observed as of the writing of this paper.

Conclusion

Bart syndrome is a rare congenital skin disorder characterized by a unique clinical presentation. Looking for other associated anomalies is important. Generally, the syndrome has a good prognosis, but it should be managed as early as possible to reach the best outcome [6]. Management can be conservative with relatively simple methods for rapid and optimal healing without the need for complex interventions. Close follow-up of the patient is recommended.

Statement of Ethics

The authors have no ethical conflicts to disclose.

Disclosure Statement

The authors have no conflicts of interest directly relevant to the content of this case report. No sources of funding were used to assist in the preparation of the manuscript.

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Fig. 1. a, b Symmetrical bilateral absence of skin over the anteromedial aspect of the legs, starting from the knees and extending to the dorsal and medial plantar aspects of the feet. The lesions have sharply demarcated borders covered by a red ultrathin translucent membrane, and vascular structures were easily visualized.



Fig. 2. Over the inner aspect of the left arm, there were blisters and erosion.

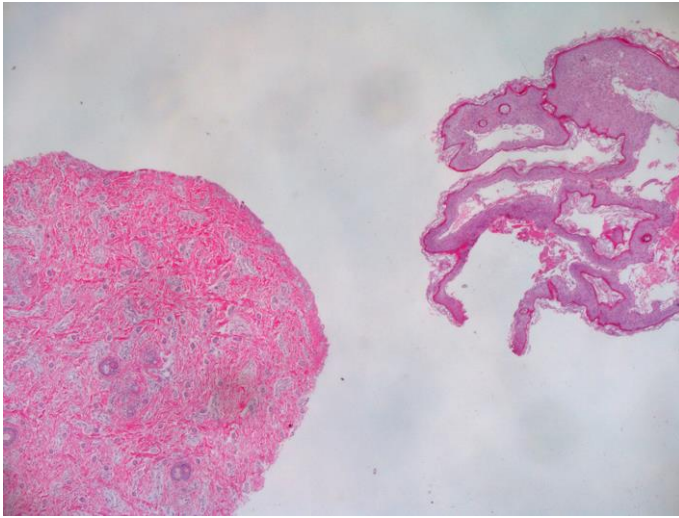


Fig. 3. Histopathological study of a skin biopsy showed subepidermal blister formation consistent with epidermolysis bullosa. H&E, $\times 4$.



Fig. 4. a, b After 2 months, the lesions were completely healed with hypopigmented scarring and milia formation. There were also some bullae, as shown by the arrows.



Fig. 5. Milia formation.