

Bart's Syndrome: A Case Report

V.V.V. Satyanarayana

Department of Dermatology, Venereology(DVL) and Leprology, Rajiv Gandhi Institute of Medical Sciences (RIMS), Srikakulam, Andhra Pradesh, India.

Abstract

Bart syndrome is a genetic disorder characterized by the association of congenital localized absence of skin (CLAS), epidermolysis bullosa (EB), lesions of the mouth mucosa, and dystrophic nails. It may be associated with any type of EB but is mostly reported with dominant dystrophic EB (DEB dominant). Clinically it is characterized by raw beefy areas of denuded skin on trauma prone areas of body. E.g. Hands & Feet. Diagnosis is obvious clinically but it requires ultra structural microscopy for proper classification of the disease. Treatment is only palliative measures. Here we report a case; A mother gave birth to two female child with 3 years gap. Both sisters showed clinical features of Bart's syndrome. 1st child Complaining of scars on both legs and bulla and 2nd child complaining of red-rich area and absence of skin on Right leg and foot with hemorrhagic bulla. We report these cases because of its rare disorder.

Keywords: Bart's Syndrome, EB, dominant DEB, Congenital localized absence of skin, Genetic Mechanobullous disorder.

I. INTRODUCTIONS

Bart's syndrome was described in a large family in 1966 and consisted of any one or a combination of the following three characteristics: congenital absence of skin, blistering and associated nail abnormalities. Congenital absence of skin is regarded now as a manifestation of epidermolysis bullosa (EB) ⁽¹⁾ Epidermolysis bullosa or EB strikes about 1 in 20,000 children and there are about 30,000 cases nationwide, half a million globally. Bart's syndrome occurs just one in a million. Bart's Syndrome is a genetic mechanobullous disorder characterized by the focal absence of skin. The affected baby is born with areas of denuded skin over body. These appear as raw, rich red plaques on different parts of the body. Any part of the skin can be involved but the disease tends to occur more on those parts of body which are exposed to friction and trauma. Such as feet, hand, arms, legs and skin around and oral cavity. The Phenomenon starts with blisters and erosions which lead to loss of skin over large areas of body. The mode of inheritance is suggested to be autosomal dominant. ⁽²⁾ Though it has been reported with any subtype of epidermolysis bullosa (EB) i.e. simplex (EBS), junctional (JEB) or dystrophic (DEB) but ultrastructural and genetic

linkage studies established firm association with dominant dystrophic EB. ⁽²⁾

II. CASE HISTORY

This is a special case where a mother had given birth to two child with Bart's syndrome. Two sisters are of less than 3 years of age, elder 2½ years, younger of 6 months presenting with some of the features of Bart's Syndrome. Both parents were apparently healthy and had no abnormalities of skin, skin appendages or mucous membrane. The presenting features of two sisters are, 1) Scarring, 2) localized absence of skin 3) Bulla 4) raw rich-red areas of denuded skin of the right Foot. No mucosal & nail involvement. This prompted in to make a diagnosis of Bart's Syndrome. Light microscopy confirmed the epidermal blistering. Further investigations to classifying it could not be done. The two sisters were managed palliatively.

Case :1 Elder Sister of age one year brought by her mother with complaining of scars on the medial aspect of the both legs and bulla on the medial aspect of right great toe since birth.

Case: 2 Younger Sister of age of 8 months brought by same months after 3 years of gap of 1st child, complaining of red-rich area and absence of skin on the medial aspect of the Right leg and foot with hemorrhagic bulla.

On examination of both sisters Case 1 showed scarring extends from below knee joint medial aspect on to the both legs and feet. A bulla is present on medial aspect of the great toe. The affected areas are hyper and hypo pigmented with well defined margins (Figure 1, 2, 3). No lesions in oral or nasal cavity. No significant lesions on the nails. Cry was sufficiently loud and she sucked well during feeding.

Case 2: After 3 years of gap. The couple came with the complaint of the similar absence of skin and hemorrhagic bulla on the right leg and foot for second female child of 6 months age (Figure 3, 4, 5). The present features are present since birth. So the couple was advised not to have children like this and the mother was sterilized (tubectomy done).

A small piece of skin including bulla was taken and submitted for Histopathological examination. Case 1: It showed epidermal detachment with intact basal cell layer and sparse infiltrate of lymphocytes with few eosinophils in the dermis of the 1st Child.

Case 2: The second child biopsy showed the similar histopathological appearance.

Thus diagnosis of Bart's Syndrome with EB simplex (EBS) was made. We could not carryout ultra structural, immune histological and genetic linkage studies because of unavailability in our

hospital. The patients were treated with antibiotics and mild topical steroids (topical hydrocortisone) and Emollient. Her mother was educated about prognosis and disease. Advised her to avoid trauma.

FIG:1 1st child of age one year with scars on the medial aspect of the both legs and bulla on right great toe since birth.



FIG:2 A bulla is present on medial aspect of the great toe .



FIG: 3 scarring extends from below knee joint medial aspect on to the both legs and feet

FIG:4 2nd child with red-rich area and absence of skin on Right leg and foot with hemorrhagic bulla.



FIG:4 Localized congenital



FIG:5 Hemorrhagic bulla Absence of skin with Epidermolysis bullosa (Bart's syndrome)

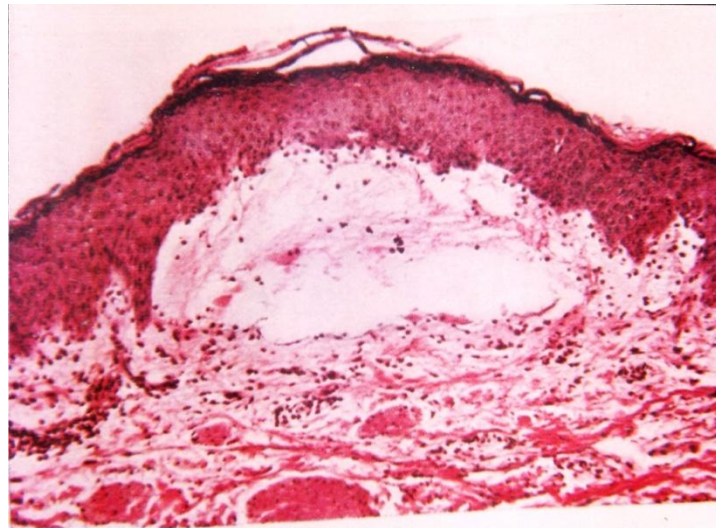


FIG:6 Micro photo graph is showing sub epidermal vesicle with leucocyte infiltration in the cavity and upper dermis

III. DISCUSSION

In 1966 Bart and his colleagues reported a family of 26 members; all of whom were having congenital absence of skin on the Figure 1 Large, bright red denuded areas on hands, feet of the patient. Lower extremities, blistering of skin and mucous membranes, and congenital absence or deformity of nails. This unique association came to be known after his name as Bart's syndrome. ⁽¹⁾

Kanzler et al. ⁽³⁾ (1992) described a family in which persons in 4 generations had epidermolysis bullosa simplex with congenital localized absence of skin (CLAS). At birth a large area of denuded skin was present on the right leg as well as a smaller area on the dorsal aspect of the left wrist and hand. The areas of CLAS healed completely by 3 months of age with minimal scarring. Nails were normal at birth. The proband's mother had an identical lesion on her leg at birth that resulted in a barely perceptible scar. She displayed no nail changes but did have skin fragility.

Later Zelickson et al. ⁽⁴⁾ carried out these studies on the original kindred described by Bart and proved that these were cases of dominant dystrophic EB associated with congenital absence of skin. Subsequently Joensen ⁽⁵⁾ in 1973 and Skoven and Drzewiecki ⁽⁶⁾ in 1979 reported similar cases.

In Our case it was not quite different from those reported earlier in the literature. Clinically it closely mimicked those described by Kanzler et al. ⁽³⁾ the clinical picture was sufficiently obvious to label it as Bart's syndrome. This suggested the benign nature of disease as mostly is seen in cases of EB simplex and was also the reason of our tendency to associate it with EB simplex. . However, in our patients there was no involvement of nails, oral and nasal cavity and both the sisters have the Bart's syndrome.

Bart's syndrome is a genetic disease caused by structural abnormalities in the anchoring fibrils of tissue. Anchoring fibrils make up part of the supportive framework that helps keep the skin and mucosa intact. When these fibrils are dysfunctional the skin loses its strength and sloughs off easily forming big blisters called bullae. Bart's original kindred demonstrated ultrastructural abnormalities in the anchoring fibrils and linkage of the inheritance of the disease to the region of chromosome 3 near the type VII collagen gene (COL7A1). These results disclosed a G-to-A transition within exon 73 of COL7A1, which results in a glycine-to-arginine substitution within the triple-helical domain of type VII collagen in affected individuals. ⁽⁷⁾ As far as my search to the literature to our best I could not find such case reported earlier.

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