

CASE REPORT

CONGENITAL LOCALISED ABSENCE OF SKIN (BART'S SYNDROME) RARE CASE REPORT

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ABSTRACT: It is defined as congenital localised absence of skin associated with epidermolysis bullosa. It may be associated with any type of epidermolysis bullosa. Three cutaneous manifestation are characteristics of Bart syndrome: congenital localised absence of skin (CLAS), mucocutaneous blistering and nail abnormalities. It present as raw beefy areas of denuded skin on trauma prone site; extremity like hands and feet, also perioral area. Diagnosis is obvious clinically but requires ultrastructural microscopy. We describe here a case of newborn baby in our OPD who presented with raw beefy red areas of denuded skin on the hands, feet, neck area with few flaccid bulla near thigh. The patient was treated with antibiotic and steroid (fusidic acid + hydrocortisone) cream and emollients. Thus, Clinical appearance was sufficiently distinct to suggest the diagnosis of Bart's syndrome.

KEYWORDS: Bart's Syndrome, Epidermolysis Bullosa, Inherited Mechano Bullous Disorder, Congenital Localised Absence of Skin.

INTRODUCTION: 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 body. There is sharp demarcation between affected and normal skin. Any part of 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, hands, arms, legs and skin around oral cavity. The phenomenon starts with blisters and erosions which lead to loss of skin over larger areas of body. The mode of inheritance is suggested to be autosomal dominant.¹ Though it has been reported with any subtype of epidermolysis bullosa (EB) i.e. simplex (EBS), junctional (JEB) or dystrophic (EBS) but ultrastructural and genetic linkage studies established firm association with dominant dystrophic EB.²

A female baby of day one was brought by her father with complaints of absent skin on hands, legs, feet and neck area since birth. On examination, the affected parts appeared raw and rich red in color and completely devoid of skin.

An abrupt transition to normal skin where the lesions ended was clearly notable. The child also had blisters on the right and left thigh.

No erosions were found in oral, genital or nasal area. No significant changes were seen on nails and hair. There were no symptoms pertaining to upper or lower aero-digestive tracts (no choanal atresia and esophageal atresia). The baby's cry was sufficiently loud and she sucked well during feeding. This excluded involvement of pharynx and larynx.

Routine blood and urine test was normal. Histology of skin showed epidermal detachment with intact basal cell layer and sparse infiltrate of lymphocytes with few eosinophils in the dermis. Thus the diagnosis of Bart's syndrome with epidermolysis bullosa simplex was made. Her family

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member was told to prevent baby from trauma to avoid blistering. She was also counseled about the prognosis and outcome of disease. The child was admitted in nicu.

A subsequent visit revealed that the child had died within three days of birth.

The exact cause of death could not be determined. Ultra structural study, immunohistochemistry and gene study could not be carried out due to non-availability.

DISCUSSION: In 1966 Bart and his colleagues reported a family of 26 members; all of whom were having congenital absence of skin on the 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. Complete penetrance was noted in all the cases.³ Bart considered congenital absence of skin as an occasional manifestation of epidermolysis bullosa simplex and attributed it to in utero blistering.⁴ However, he could not properly classify the disease as ultra-structural and immunochemical studies were not available at that time. Later Zelickson et al. 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 Joensen⁵ in 1973 and Skoven and Drzewiecki⁶ in 1979 reported similar cases. Kanzler et al.⁷ described a family in which members in 4 generations had epidermolysis bullosa simplex with congenital localized absence of skin. Thus it is clear from literature review that CLAS occurs in association with all the three major types of inherited epidermolysis. The clinical picture was sufficiently obvious to label it as Bart's syndrome. However, in our patient there was no involvement of mucosa and nails. 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, electron microscopy and immunochemical studies are essential for the more accurate classification of disease bullosa. Keeping this view Kanzler et al.⁷ suggested abandoning Bart's syndrome as separate disease entity. However its familial occurrence and association with specific mutation in COL7A1 with glycine-toarginine substitution in the triple helical domain of type VII collagen merits its retention as a unique clinical entity.⁸

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Figure 1: Showing raw,beefy red denuded skin over both legs



Figure 2: Showing denuded skin over hand and neck region



Figure 3: Showing flaccid bullae over rt thigh region

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