

Case Report

Bart's syndrome: a case report

Doulat Rai Bajaj, Asim Qureshi*

Department of Dermatology, Liaquat University of Medical & Health Sciences, Jamshoro

* Department of Histopathology, Agha Khan University Hospital, Karachi

Abstract Bart's syndrome is defined as congenital localized absence of skin (CLAS) associated with epidermolysis bullosa (EB). It may be associated with any type of EB but is mostly reported with dominant dystrophic epidermolysis bullosa (DEB dominant). Clinically it is characterized by raw beefy areas of denuded skin on trauma-prone areas of body e.g. hands and feet. Diagnosis is obvious clinically but requires ultrastructural microscopy for proper classification of the disease. Treatment suffices to palliative measures.

We describe here a case of newborn baby who presented with rich-red areas of denuded skin on the hands and feet. Clinical appearance was sufficiently distinct to suggest the diagnosis of Bart's syndrome. We report this case because of its rarity.

Key words

Bart's syndrome, epidermolysis bullosa, dominant DEB, congenital localized absence of skin, genetic mechano-bullous disorders.

Introduction

Bart's syndrome is a genetic mechano-bullous 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 (DEB) but ultrastructural and genetic linkage studies established firm association with dominant dystrophic EB.²

An interesting case of newborn baby who had large areas of denuded skin on hands, feet and face is presented here. There was no mucosal and nail involvement. This prompted us to make the diagnosis of Bart's syndrome. Light microscopy confirmed epidermal blistering. Further investigations to classify it could not be done. The patient was treated with palliative measures as no specific treatment exists there.

Case report

A female baby of 3 months was brought by her mother with complaints of absent skin on hands, legs and feet since birth. On

Address for correspondence

Dr. Doulat Rai Bajaj,
House No: 2970/4-3, Journalist Colony,
Hyderabad, Sind, Pakistan
Ph: +923003076504, + 92223008708
E-mail: doulat01@yahoo.com

examination large areas of denuded skin were seen on feet, lower legs and hands. Similar small areas were also found on perioral region. The affected parts appeared raw and rich red in colour and completely devoid of skin (**Figures 1**). An abrupt transition to normal skin where the lesions ended was clearly notable. No erosions were found in oral or nasal cavity. Similarly no significant lesions were seen on nails. There were no symptoms pertaining to upper or lower aero-digestive tracts. The baby's cry was sufficiently loud and she sucked well during feeding. This excluded involvement of pharynx and larynx. She was the first baby of couple.

A small piece of skin was taken and submitted for histopathological examination. It 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 (EBS) was made. We could not carry out ultrastructural, immunohistologic and genetic linkage studies because of non-availability. The patient was treated with antibiotic-cum-steroid (fusidic acid + hydrocortisone) cream and emollients. Her mother was told to prevent baby from trauma to avoid blistering. She was also counseled about the prognosis and outcome of disease. There was modest improvement in the lesions.

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



Figure 1 Large, bright red denuded areas on hands, feet of the patient.

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 ultrastructural 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 Joensenin⁵ 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 (CLAS).

Thus it is clear from literature review that CLAS occurs in association with all the three major types of inherited epidermolysis 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-to-arginine substitution in the triple helical domain of type VII collagen merits its retention as a unique clinical entity.⁸

Our case was not quite different from those reported earlier in the literature. Clinically it closely mimicked those described by Kanzler *et al.*⁷ The clinical picture was sufficiently obvious to label it 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. We searched the local literature to our best but could not find such case reported earlier.

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