Bart's Syndrome: A rare genetic disorder

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Abstract

Bart syndrome is a genetic disorder characterized by three cutaneous manifestations- congenital localized absence of skin (CLAS), mucocutaneous blistering, and nail abnormalities. The syndrome is a clinical variant of dominant dystrophic Epidermolysis Bullosa (EB) which imparts a spectrum of blistering diseases showing a disturbance in the top layer of the skin (epidermis) causing it to blister. Dystrophic EB is accompanied by scarring. The present study highlights a case of newborn baby affected with rich-red areas of denuded skin on the left leg and and foot. Clinical appearance was sufficiently distinct to suggest the diagnosis of Bart's syndrome. This typical case is reported because of its rarity.

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Introduction

Bart's syndrome detected in a large family was earlier described in 1966 which 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 now regarded as a manifestation of epidermolysis bullosa (EB)[1]. Although Bart is considered, but not a disorder but a clinical sign seen in many forms of EB. The diagnosis of Bart syndrome can be made clinically in a patient with a strong family history of the disease along with the classic cutaneous findings[2] .There is no cure or accurate therapy for Bart syndrome. Gene therapy is currently being investigated. Treatment of symptoms is the goal of therapy. Saline compresses and topical antibiotics can be applied to the affected areas to keep the area clean and prevent infection. In the case of inflammation, topical steroids may be used. Severely affected patients are treated in a burn unit. Gentle bathing and cleansing are followed by protective emollient and nonadherent dressing application. In dystrophic EB control of cutaneous infections is an important issue because scarring can lead to fusion of digits and limb contractures.

The present study reflects an investigation report a case of Bart's syndrome, which is exceedingly a rare disorder.

Case history

A new-born boy got admitted into the department of Pediatrics of Community Based Medical College, Bangladesh and later referred to the Department Dermatology and Venereology for congenital absence of skin. The baby was the third child of a non-consanguineous couple.

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The history revealed that the pregnancy and delivery were normal. Both parents were found apparently healthy and had no abnormalities of skin, skin appendages or mucous membrane. However there was a positive family history of one female child born earlier having blistering skin lesions, who died after a few days of birth.

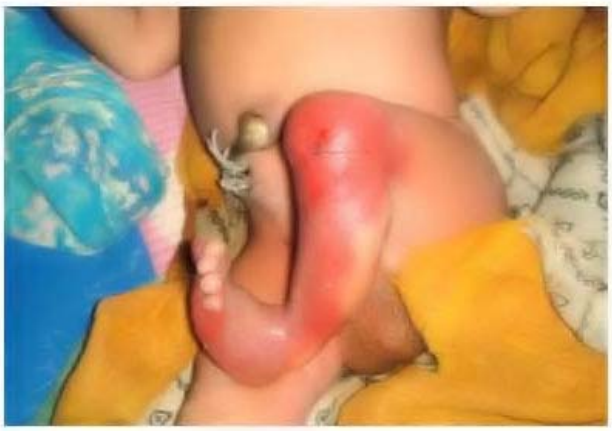

Fig. 1 : Showing localised absence of skin over left leg & foot of Bart's syndrome.

On examination of the child, there was absence of skin on the anterior and lateral surface of the left leg and dorsum of the left foot (Fig. 1). The defect extended proximally up to the knee and distally up to lateral half of sole. Erosions were present on distal phalanges of left hand, fourth finger nail of left hand and big right toe nail. There were no oral erosions. Systemic examination was normal.

Skin biopsy from ulcerated skin lesions revealed ulcerated epidermis. Upper dermis showed neutrophils, eosinophils and an occasional hair follicle. Skin biopsy from the blister evidenced a sub-epidermal blister; PAS stain revealed basement membrane along with dermis suggesting the diagnosis of EB simplex or junctional EB. Electron microscopic studies could not be performed due to crisis of laboratory facilities and other limitations.

The child was discharged on request after a week. From subsequent home visits we got information that the child died within 10 weeks of birth. The exact cause of death could not be ascertained in two affected babies under this investigation.

Discussion

Bart considered congenital absence of skin as an occasional manifestation of epide-rmolysis bullosa simplex and attributed it to in utero blistering[3] However, he could not properly classify the disease as ultrastructural and immunochemical studies were not available at that time. Later Zelickson et al[2] 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[4] in 1973 and Skoven and Drzewiecki[5] in 1979 reported analogous cases. Kanzler et al[6] described a family in which members in 4 generations demonstrated epidermolysis bullosa simplex with congenital localized absence of skin (CLAS). Thus it is evidenced from literatures that CLAS occurs in association with all the three major types of inherited epidermolysis bullosa. Keeping this in view Kanzler et al6 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[7].

The present study case is not quite different from those reported earlier in literatures. Clinically it closely mimicked those described by Kanzler et al. 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. Effort was made to search the native literatures but no report about this case was found any where in the country.

Conclusion

Genetic counseling for this rare familial disorder is extremely important for any affected families. In Bangladesh we should be aware of this disorder and as far as practicable establish laboratory facilities about DNA-based prenatal diagnosis using Chorionic villus sampling or Aminocentesis for junctional and dystrophic forms of EB, wherein the mutationscould be characterized. Future avenues are currently under investigation for early prenatal diagnosis, including preimplantation genetics.

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