BART SYNDROME: CONGENITAL APLASIA CUTIS TYPE VI

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ABSTRACT

Bart's syndrome is described as absence of skin in a localized area by birth also called Aplasia cutis congenital type VI. It may be concomitant with any type of EB but the most common cases reported are allied with dystrophic epidermolysis bullosa (DEB dominant). The clinical presentation of this disease is that the areas of the body which are prone to injury area lacking skin e.g. hands and feet. Diagnosis is done by clinical examination and but for proper disease classification microscopic examination is required only symptomatic treatment is done.

Our case is of a neonate who presented with wide-denuded erythematous areas on the neck, upper and lower limbs. Clinical examination helped us to reach the final diagnosis that is Bart's syndrome.

Keywords: Bart's syndrome; Aplasia cutis; Epidermolysis bullosa; Congenital localized lack of skin.

INTRODUCTION

Aplasia cutis congenital type VI or Bart's syndrome is a described by the focal absence of skin due to genetic defect and it involves formation of bullae.[1] it is an autosomal dominant trait and it was first described in a family in 1996.[2] The baby suffering from Bart syndrome had areas of bared skin over the body, along with sharp demarcation between diseased and healthy skin. Any area of the skin can be affected but the skin of body parts which are more prone to trauma and friction is a favorite target in Bart baby such as hand, feet, arms and legs along with the skin around mouth.[3,4] First of all there is blister formation and later on erosions and trauma lead to extension of blisters over large area and ultimately denudation of skin over large area of body. Sometimes the affected person may have absence of bones along with skin. The disease follows Mendelian inheritance pattern with autosomal dominant manner.

We report an attention-grabbing case of a newborn baby who was born with absence of skin over large areas of body involving the face upper and lower limbs. But There mucosa over the surfaces including mouth was intact. The diagnosis was confirmed by Light microscopy which showed epidermal blistering. The patient was managed conservatively.

CASE REPORT

A female premature baby was brought to us with the absence of skin on the neck, upper and lower limbs. Examination revealed denuded areas on the neck, feet, lower limbs, and hands, where the skin was completely lacking, leaving raw, erythematous tissue with visible blood vessels as shown in Figure 1.

The patient did not have any signs of nasal or oral mucosal involvement or erosions, but did have dystrophic nails and a tracheoesophageal fistula. Since the baby's cry was normal, it was determined that the pharynx and larynx were not affected. This was the first occurrence of the disease in the family and advanced diagnostic tests, such as genetic analysis for gene identification, were not available. Skin biopsy was not performed due to lack of permission from the family.

To address the symptoms, the patient was treated with Fusidic H, which is an antibiotic and steroid ointment. Additionally, a thermo-controlled environment was provided to maintain skin moisture, and the attendants were instructed to prevent trauma to minimize blistering. The attendants were also educated on the nature of the disease, potential complications, transmission patterns, and potential outcomes.

DISCUSSION

A family of 26 members was reported by Bart in 1996 with all of the members having a inborn absence of skin on the lower limbs, blisters on the skin and mucous membranes along with dystrophy of nails.[5] Later Zelickson et al reported that the original kindred described by Bart were cases of dominant dystrophic EB along with the nonexistence of skin.[6] Later on, similar cases were reported by Joensenin in 1973 and

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

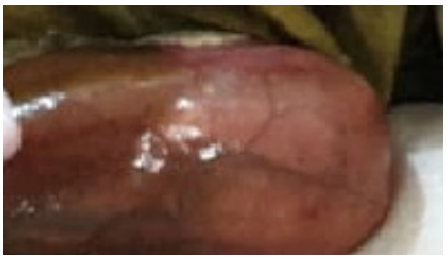

Figure 2: Right leg with fragile skin and visible vessels

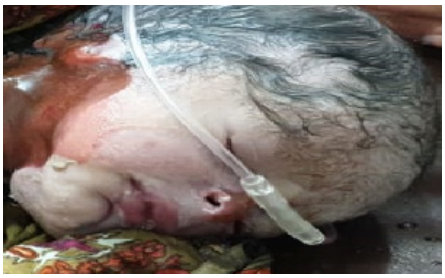

Figure 3: Large, bright red denuded areas on head of the patient

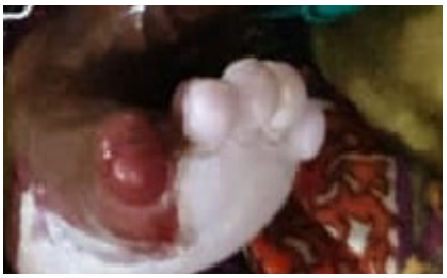

Figure 4: Large, bright red denuded areas on feet of the patient with sharp demarcation

Drzewiecki in 1979. Kanzler et al reported a family having 4 generations with epidermolysis bullosa simplex with the localized inborn absence of skin. There patient went detailed examination including both electron microscopy and immunofluorescense which showed a similarity between Aplasia cutis and epidermolysis bullosa.[7] Mc Kinster et al in 2001 reported six cases of Bart syndrome and proposes that inborn absence of skin in Bart's syndrome follows the lines of Blaschko.[5]

We report a similar case to those reported earlier in the literature. Our case presented with the classic triad of the Bart syndrome including all three features e.g. congenital lack of skin over localized areas including both lower legs, formation of skin blisters, and nail dystrophy. Clinically there are three presentations of Bart syndrome these include: epidermal, junctional or dermal epidermolysis bullosa. The classification of our case could not done because ultramicroscopic and other detailed examinations were not done because of lack of facilities. The variant named junctional epidermolysis bullosa is a severe variant in which, the patient may present with other inborn anomalies, such as pyloric atresia, ureteral stenosis, ear, nose and renal problems. There has also been association of Bart syndrome with ambiguous genitalia,[4] but our patient had normal genitalia. The death usually occurs due to metabolic abnormalities like decrease glucose level in blood and hypothermia and septicemia due to congenital absence of skin o barrier to pathogens like bacteria and viruses. However, in our case, there were no associated anomalies. It is autosomal dominant disease. The diagnosis of disease is usually clinical but can be confirmed by advanced investigations like electron and immunofluorescence microscopy and genetic analysis.[8] the treatment is palliative. The prognosis of Bart syndrome depends on many factors, such as the variant of the disease the areas involved and other associated abnormalities. The prognosis of patients with Bart syndrome is good.

CONCLUSION

Bart syndrome is dominantly inherited but rare and diagnosed immediately by clinical examination. More cases should be reported after genetic analysis and a lot more trials are awaited for identifying effective treatment options.

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Author's Contribution

SR, MN, NHB, and FA contributed in data collection, article writing, and final approval of the manuscript. Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflict of Interest

Authors declared no conflict of interest

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None

Data Sharing Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.