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Barts Syndrome: A Case Report

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Abstract
Bart's syndrome is characterized by aplasia cutis congenita and epidermolysis bullosa. A male baby was presented on day 3 of birth with congenital absence of skin on the bilateral legs with associated bullous lesion over left thumb and rim of right nostril along with oral mucosal eroions. Barts syndrome was diagnosed clinically and patient was managed conservatively. There was no similar family history. His vitals, systemic examinations and blood investigations were all within normal limits.
Key Words: *Aplasia cutis congenita, Barts syndrome, epidermolysis bullosa*

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I. Introduction

Barts syndrome (BS) is characterized by striking clinical findings, including congenital localized absence of skin (CLAS) affecting the lower extremities, blistering of skin and/or mucous membranes, and absence and/or deformity of nails.[1]There is sharp demarcation between affected and normal skin. Any part of skin can be involved but most frequently trauma prone areas such as feet, hands, arms, and legs are involved.[2]Bart syndrome is a very rare disorderwhich was first described in a large family almost half a century ago.[3]Although the inheritance pattern appears to be autosomally dominant, isolated cases have been recognized.[4]

BS is diagnosed clinically based on the disorder's unique signs and symptoms but histologic evaluation of the skin can help to confirm the final diagnosis.[4] The management of BS is usually conservative, preventing infection of affected area and to accelerate healing and reduce the risk of scarring.[5]

II. Case Report

A newborn on day 3 was referred to us for congenital absence of skin. He was the second child of a non-consanguinous couple. He was delivered via normal vaginal delivery at term.

The pregnancy and delivery was normal. Both parents were apparently healthy and had no abnormalities of skin, skin appendages or mucous membrane. Weight, height, head circumference were normal and baby cried immediately after birth. APGAR score was 8 and 10 at 1 and 5 minutes, respectively. His vital signs were normal. His elder sister was born 2 years back and died after 10 days of birth due to pneumonia. However, there were no any skin lesions or bulla. There is no family history of similar lesions.

On physical examination, he had absence of skin over the anteromedial aspect of left leg starting from the knees and extending to dorsal and medial plantar aspect of the feet while on right foot there was absence of skin at lower third leg extending upto dorsal and medial plantar aspect of foot as shown in figure1. The affected parts appeared raw and rich red in colour and completely devoid of skinwith sharply demarcated borders in a broad linear pattern. The skin adjacent to these defects appeared normal.On the second day of birth, he developed tense bulla over left thumb and another at the rim of right nostril as shown in figure 2 and 3. Erosions were present over oral mucosa shown in figure 3. However, nail involvement was not present. Sucking reflex was normal and his vitals were all within normal limit. Spontaneous activity was present and the tone of the muscles were normal.

All the systemic examination were also normal. Blood investigations that includes complete blood count, renal function test, liver function test, blood sugar were all within normal limits. Biopsy couldnot be done as the parents did not give consent. The patient was treated with antibiotic-steroid (fusidic acid + hydrocortisone) cream and emollients along with wet gauze dressings 2 times a day. Parents were counselled regarding the nature of disease and its prognosis and were advised to avoid any trauma.

We reported this case as it is a very rare disease encountered only once in a while during practice.

Figure 1: large bright red denuded areas on bilateral legs

Figure 2: tense blister over right nostril and left thumb

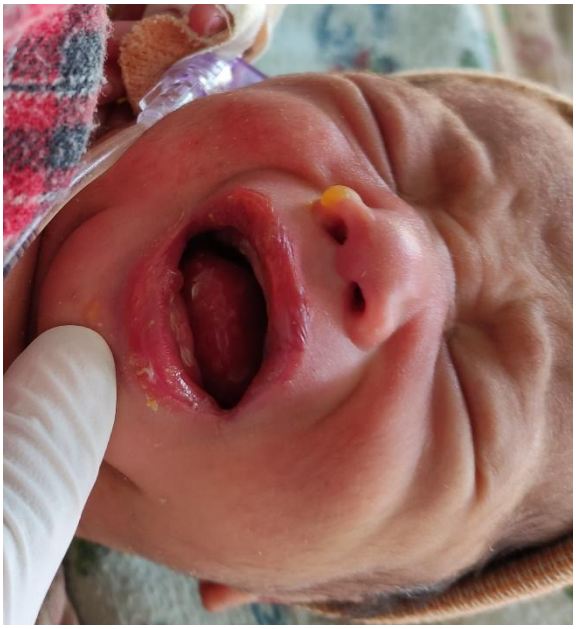

Figure 3: erosions over oral mucosa and blister over right nostril

III. Discussions

Bart syndrome was described by Bart et al. in 1966 as a new syndrome. It is an inherited disorder characterized by epidermolysis bullosa, aplasia cutis congenita, and nail deformities. The genetic abnormality has been linked to chromosome 3, with an autosomal dominant pattern of inheritance.[6]

BS is sometimes accompanied by nail abnormalities which are not absolutely required for making the diagnosis.[4] In our patient, there was congenital localized absence of skin over bilateral legs and blistering lesions but there was no involvement of nail.Though the predominant inheritance pattern is autosomal dominant, while some cases with unaffected parents are believed to occur due to sporadic mutation. Chiaverini et al. reported sporadic cases to be associated with mutations of the triple helix domain of collagen VII gene.[7] Our patient had no family history of congenital localized absence of skin and blistering lesions; therefore he might have a sporadic mutation.

Lesions in BS are usually unilateral and involved the medial and/or dorsal surface of the limbs. They appear on extremities as sharply demarcated, glistening red ulceration that extend upward from the dorsal and the medial surface of the foot to the shin.[8] Our patient also presented similarly though there was bilateral involvement of legs. Also, multiple oral mucosal ulcerations were present which was similar to the another case reported by Aygun etal.[5]BS can also be associated with other anomalies as pyloric atresia, rudimentary ear development, flattened nose, broad nasal root, and wide-set eyes.[6] The present case had no associated anomalies.

Bart syndrome is usually diagnosed clinically. In some cases, skin biopsy is done to determine the type of epidermolysis bullosa and genetic study to look for the exact gene mutation that may help to confirm the final diagnosis.[3] In our case, the diagnosis of Bart syndrome was made based on the characteristic clinical presentation, including congenital localized absence of skin over the anteromedial aspect of both lower legs, blistering of the skin, and erosions over oral mucosa. Biopsy was not done because of lack of parental consent.

The prognosis of Bart syndrome depends on many factors, such as the severity and extension of ACC, epidermolysis bullosa subtype, associated anomalies, and efficacy of treatment. In general, the prognosis of patients with Bart syndrome is good. They have a normal life expectancy and most cases are able to live a normal life. The management of BS is usually conservative that includes the proper wound care, control of infection and prevention of complications.[3]The goal of treatment is to accelerate healing and reduce the risk of scarring.[5] We also managed the patient with antibiotic-steroid cream, emollients and wet gauze dressings two times daily.

In conclusion, Bart syndrome is a rare syndrome, which should be considered as a differential diagnosis in a patient with ACC and bullous lesions. Management can bedone conservativelyfor rapid and optimal healing without the need for complex interventions. Close follow-up of the patient is recommended.

Consent was taken from the parents of the patient

Conflict of interest: None

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