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BART SYNDROME: A SPORADIC CASE REPORT OF APLASIA CUTIS CONGENITA ASSOCIATED WITH EPIDERMOLYSIS BULLOSA AND NAIL HYPOPLASIA

Dermatology

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ABSTRACT

Bart syndrome is a rare congenital disorder characterized by a triad of aplasia cutis congenita (ACC), epidermolysis bullosa (EB), and nail anomalies. First described by Bart et al., it is considered a subtype of ACC with EB and may occur sporadically or be inherited in an autosomal dominant manner. We report a sporadic neonatal case presenting with extensive absence of skin over the right lower limb, scalp erosions, and nail hypoplasia. Systemic evaluation was unremarkable. Early diagnosis, meticulous wound care, and multidisciplinary support are crucial in preventing complications and improving outcomes in such cases.

KEYWORDS

Bart Syndrome, Aplasia Cutis Congenita, Epidermolysis Bullosa, Neonatal Genodermatosis, Nail Dysplasia

INTRODUCTION

Bart syndrome, first reported by Bart et al. in 1966, represents a unique genodermatosis defined by the coexistence of congenital skin absence (ACC), blistering lesions resembling EB, and nail hypoplasia or absence [1]. It is classified under type VI of aplasia cutis congenita. The inheritance is usually autosomal dominant, although several sporadic cases have been documented [2].

The clinical presentation is typically apparent at birth and includes sharply demarcated areas of skin absence, most commonly over the lower extremities. Blistering lesions may involve trauma-prone areas such as the scalp and limbs. Nail involvement can range from mild dystrophy to complete absence. The etiology is believed to involve structural protein defects in the dermo epidermal junction, similar to those seen in EB [3]. Early recognition is vital to initiate proper wound care and genetic counselling.

Case Description

A term female neonate was delivered via normal vaginal delivery to a non-consanguineous couple. The birth weight was 2.8 kg with uneventful perinatal history and normal Apgar scores. There was no family history of similar skin lesions, neonatal deaths, or genodermatoses.

Cutaneous examination revealed a large, well-demarcated area of skin absence over the anteromedial aspect of the right lower limb, extending from the thigh to the dorsum and plantar aspect of the foot. The lesion was ulcerated, raw, and glistening, without signs of infection at presentation.

Multiple bullous and erosive lesions were present on the scalp and retroauricular areas. The second toe showed nail hypoplasia. The remaining nails were intact. There were no mucosal erosions, and systemic examination revealed no abnormalities.

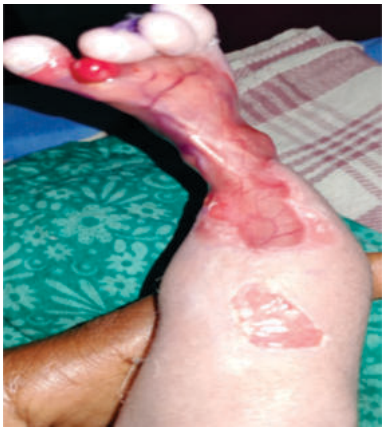

Figure 1: A Single Extensive Area of Absent Skin Noted in Right Leg

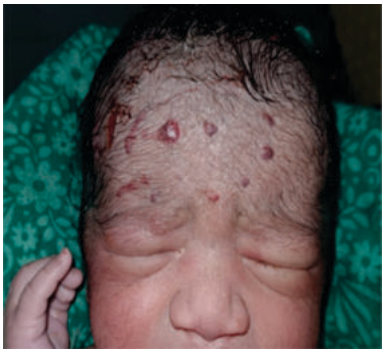

Figure 2:Multiple Bullous Lesions Seen Over Scalp

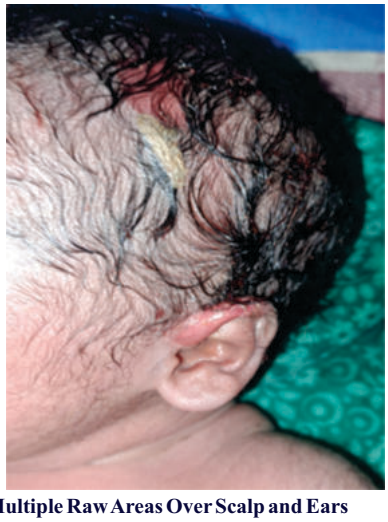

Figure 3 :Multiple Raw Areas Over Scalp and Ears

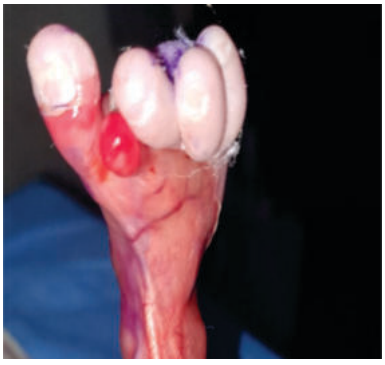

Figure 4: Hypoplasia of Second Toe Nail

Table – 1 Differential Diagnosis

Condition	Distinguishing Features
EB simplex	No congenital skin absence or nail dysplasia
Adams-Oliver syndrome	ACC with limb reduction defects, vascular anomalies
Neonatal HSV	Vesicles, mucosal involvement, systemic signs
Goltz syndrome	Fat herniation, atrophic patches, skeletal anomalies

Investigations and Differential Diagnosis

Routine hematological and biochemical investigations were within normal limits.

Tzanck smear from the blister was negative for acantholytic or multinucleated giant cells.

TORCH screen was negative.

Ultrasonography of the abdomen and 2D echocardiogram revealed no internal organ malformations.

Based on the constellation of congenital skin absence, bullous lesions, and nail hypoplasia, a diagnosis of Bart syndrome was made.

Differential Diagnoses Considered Included:
Epidermolysis bullosa simplex
Adams-Oliver syndrome
Congenital herpes simplex
Congenital varicella syndrome
Focal dermal hypoplasia (Goltz syndrome)

However, the absence of limb reduction defects, skeletal anomalies, or systemic signs helped rule out Adams-Oliver and Goltz syndromes. Negative viral studies excluded herpes and varicella. The diagnosis was thus consistent with sporadic Bart syndrome.

Management

The raw area over the leg was gently cleaned and dressed daily using non-adherent, petrolatum-based gauze. Topical mupirocin was applied to prevent secondary bacterial infection. The neonate was managed in a sterile environment to minimize trauma.

A multidisciplinary team involving dermatology, pediatrics, neonatology, and genetics was engaged. The parents were educated on the nature of the condition, the need for gentle handling, and the importance of long-term follow-up.

By the 10th day, the denuded area began to re-epithelialize, and no new bullae were observed. The neonate was discharged on day 14 with scheduled outpatient monitoring.

DISCUSSION

Bart syndrome is classified under Type VI ACC, wherein congenital absence of skin coexists with epidermolysis bullosa and nail dysplasia [2]. The pathophysiology involves structural protein defects in the basement membrane zone—especially in genes such as COL7A1, LAMB3, or ITGA6, depending on the EB subtype [4].

Clinical Features

ACC typically involves the lower extremities, but the scalp, trunk, or upper limbs may also be involved.

Blistering lesions may occur spontaneously or secondary to minimal trauma.

Nail changes include hypoplasia, dystrophy, or complete absence.

Histology usually shows subepidermal clefting. Genetic testing confirms EB-associated mutations but may not be immediately necessary in resource-limited settings.

Prognosis is generally favorable in isolated Bart syndrome. Major complications include secondary infection, scarring, and delayed healing. Rare associations with renal anomalies, cleft lip/palate, or musculoskeletal deformities have been documented [5].

Management Remains Supportive, Focusing on:

- Infection prevention
- Atraumatic wound care
- Genetic counseling
- Monitoring for late sequelae (alopecia, EB blistering, delayed dentition)

CONCLUSION

This case highlights the classical presentation of sporadic Bart syndrome, emphasizing the importance of clinical suspicion in neonates with congenital skin absence and blistering. Absence of systemic involvement, family history, or consanguinity supports a de novo mutation in this case. A multidisciplinary approach with supportive care ensures optimal recovery. Genetic evaluation and counseling are recommended to assess recurrence risk and long-term outcomes.

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