Avens Publishing Group
J Clin Investigat Dermatol
June 2026 Volume 14, Issue 1

A Neonatal Case of Bart Syndrome: First Reported Case from Yemen

Abstract

A 10-day-old female infant presented with skin erosion and atrophy of the left leg, accompanied by an absence of most fingernails, evident since birth. In addition, she developed flaccid bullae and erosions at the sites of trauma. On the basis of these clinical findings, the patient was diagnosed with Bart syndrome, a rare type of genodermatosis characterized by the clinical triad of aplasia cutis congenita, epidermolysis bullosa, and nail abnormalities.

Introduction

Aplasia cutis congenita (ACC) is an inherited absence of skin [1]. Among its different manifestations, Bart syndrome (BS), also referred to as ACC type VI, is an extremely rare autosomal dominant or recessive genodermatosis characterized by the classic triad of ACC of the lower limbs, epidermolysis bullosa (EB), and nail dystrophy [2]. BS was first described in 1966 by Bart, based on findings in members of a family displaying one of three features, namely ACC, EB, or nail dystrophy [2]. To date, fewer than 200 BS cases have been reported worldwide, and herein we describe the first case from Yemen.

Case report

A 10-day-old female infant presented to our clinic with congenital skin erosion and atrophy of the left leg, along with an absence of most fingernails (Figure 1-3). She had no feeding difficulties, and her growth was normal. Dermatological examination revealed a longitudinal denuded erythematous band 3 x 20 cm in diameter, extending from the knee to the base of the big toe of the left leg and including the medial portion of the sole. In addition, she developed bullae and erosions on the left thumb, due to minor trauma, but signs of systemic infection were absent. The nails of the left thumb and left big toe were dystrophic. Mucous membranes and hair were not affected. There was no family history of similar lesions or consanguinity, and the pregnancy and delivery were uneventful. The differential diagnosis included isolated ACC, inherited forms of EB, and Adams–Oliver syndrome; however, the coexistence of congenital localized skin absence, trauma-induced blistering, and nail abnormalities strongly supported the diagnosis of BS. Histopathological examination, immunofluorescence antigen mapping, and genetic analysis were not performed because of limited local diagnostic resources. Based on these clinical findings, the patient was diagnosed with BS, for which she was prescribed topical mupirocin ointment and fusidic acid fatty gauze for wound care. She responded well, with substantial healing observed within 2 weeks.

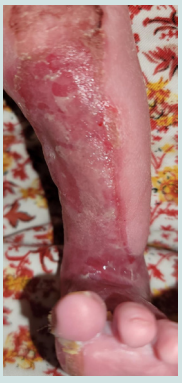

Figure 1: Aplasia cutis congenita involving the medio-anterior aspect of the left leg.

Open Access Case Report

Journal of Clinical & Investigative Dermatology

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Submission: 13 April, 2026
Accepted: 01 June, 2026
Published: 05 June, 2026

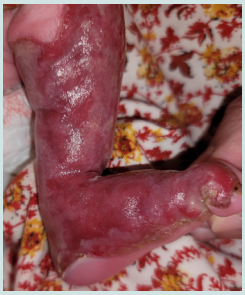

Figure 2: Medial view of the left leg, Glistening red aplasia cutis congenita extending to the sole.

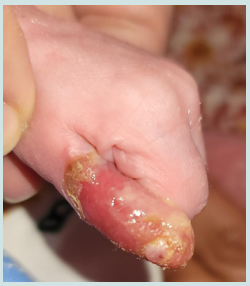

Figure 3: Epidermolysis bullousa, along with onychodystrophy of the left thumb

ISSN: 2373-1044

Discussion

BS, an exceedingly rare genodermatosis, comprises ACC of the lower extremities, EB, and nail abnormalities [3]. A glycine substitution mutation in the type VII collagen gene underlies BS [4]. ACC is a congenital anomaly characterized by a localized absence of skin. Based on its distribution and associated anomalies, this disorder has been categorized into nine groups, among which BS is classified as ACC type VI [1,2]. EB, a further type of genodermatosis, is characterized by increases in the development of skin and mucous membrane fragility-related blisters; it can be broadly classified into four major types based on the ultrastructural level of skin cleavage [5,6] ACC is associated with all types of congenital EB, particularly dystrophic dominant and recessive EB [7]. In BS, the distribution of ACC is generally uniform, involving the anterior aspects of the lower extremities and the dorsum of the feet. The lesions are typically characterized by an S-shaped, sharply demarcated involvement of the toe webs along the Blaschko lines, with distinct borders [8]. The differential diagnosis included isolated ACC, inherited forms of EB, and Adams–Oliver syndrome. The coexistence of congenital localized skin absence, trauma-induced blistering, and nail abnormalities strongly supported the diagnosis of BS. Regarding management, both conservative and surgical approaches have been advocated, each of which has distinct advantages and disadvantages [9,10]. In the present case, we adopted a conservative approach, which yielded excellent results within 2 weeks, consistent with previous findings.

Written informed consent for publication of clinical details and images was obtained from the patient's parents.

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