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Fatal Neonatal Sepsis in Suspected Bart Syndrome: A Case Report

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Abstract

Bart syndrome is defined by the association of a congenital absence of skin on the limbs with inherited epidermolysis bullosa. We report the case of a male newborn born at term to first-cousin parents, whose older brother had died on day 7 of life with similar skin lesions. At birth, the infant presented with well-demarcated areas of absent skin on all four limbs and a crusted nasal lesion. Initial laboratory tests were normal, and he was managed with non-adhesive dressings, without antibiotic prophylaxis. Skin fragility with a positive Nikolsky sign appeared on day 1, without blisters, and non-bilious vomiting began on day 3. On day 5, he developed septic shock with leukopenia and thrombocytopenia, and the blood culture grew methicillin-susceptible *Staphylococcus (S.) aureus*. Despite broad-spectrum antibiotics started at the onset of shock, he died on day 6. Skin biopsy and genetic testing were not available, and the diagnosis of Bart syndrome therefore remained presumptive, based on clinical findings and family history. This case highlights the severity of suspected familial Bart syndrome, in which infection may be rapidly fatal. In newborns with extensive skin defects, close monitoring for infection is essential; any empirical antibiotic therapy for suspected infection may need to cover *S. aureus*, and early storage of a DNA sample for later diagnosis and genetic counseling should be considered.

Categories: Pediatrics, Infectious Disease, Dermatology
Keywords: aplasia cutis congenita, bart syndrome, congenital absence of skin, consanguinity, epidermolysis bullosa, neonatal sepsis

Introduction

Aplasia cutis congenita (ACC) is the congenital absence of skin. In a population-based European study, its live-birth prevalence was 4.81 per 100,000 births [1]. It involves the scalp in most cases [2]. Frieden classified ACC into nine groups [2]. Group VI, or Bart syndrome, combines ACC of the limbs with epidermolysis bullosa (EB) [2,3]. Bart described the syndrome in 1966 in a family with autosomal dominant inheritance. The disease in this family was later linked to a *COL7A1* mutation [3,4]. EB is a group of inherited disorders in which the skin is abnormally fragile and blisters after minor trauma [5,6]. Current EB classifications no longer consider Bart syndrome a separate entity and use the term EB with congenital absence of skin [5,6]. Congenital absence of skin can occur in EB simplex, junctional EB, and dystrophic EB [6]. These types differ by the level at which the skin separates, within the epidermis, within the basement membrane, or below it, and this partly explains their different severity [5,6].

The main neonatal risks are infection, fluid loss, and bleeding [7-9]. Skin biopsy and genetic testing are often unavailable in low-resource settings. In this situation, the diagnosis relies on clinical findings alone, and clinicians must manage a high-risk neonate and counsel the family without knowing the exact EB type or its prognosis. We report a fatal familial case diagnosed on clinical grounds.

Case Presentation

A male infant was born at 37 weeks by vaginal delivery. Birth weight was 3,460 g, and Apgar scores were 10 at 1 and 5 minutes. The parents were first cousins with no skin disease. The pregnancy and three antenatal ultrasound scans were normal. There was no maternal illness or drug exposure. The couple had two older sons: one died on day 7 of life with similar skin lesions, and the other, now three years old, is healthy.

At birth, the baby was in good condition. He had several well-demarcated areas of absent skin on all four limbs, covered by a thin membrane (Figures *1A*-*1C*). The largest lesion, on the lower limb, extended from the knee to the dorsum of the foot (Figure *1A*). A small crusted lesion was present on the nose (Figure *2*). Nails and mucosa were normal. There was no scalp defect or other malformation.



FIGURE 1: Congenital absence of skin on the limbs

(A) Lower limb: a well-demarcated area of absent skin, covered by a thin, shiny membrane through which vessels are visible, extending from the knee to the dorsum of the foot. (B) Lower limb held up: a well-demarcated area of absent skin with visible vessels. (C) Upper limb: a well-demarcated area of absent skin on the hand, extending to the thumb.



FIGURE 2: Nasal lesion at birth

Small crusted lesion on the nose.

Echocardiography, transfontanellar ultrasound, and abdominal ultrasound were normal. On admission, the white blood cell (WBC) count, neutrophil and lymphocyte counts, hemoglobin level, and platelet count were normal, as were the C-reactive protein (CRP) and lactate levels (Table *1*). A blood culture taken on admission, before any antibiotic administration, was sterile, and serology for toxoplasmosis, other agents, rubella, cytomegalovirus, and herpes simplex (TORCH) was negative. Herpes simplex polymerase chain reaction, skin biopsy, and genetic testing were not available.

Parameter	On admission (day 1 of life)	Reference range (day 1)	Day 5 of life	Reference range (day 5)
WBC count (/mm³)	14,000	9,000–30,000	2,200	5,000–21,000
Neutrophils (/mm³)	10,000	7,500–28,500	1,400	2,700–13,000
Lymphocytes (/mm³)	4,000	2,000–11,000	806	2,000–11,500
Hemoglobin (g/dL)	15	14.5–22.5	14.4	13.5–21.5
Platelets (/mm³)	188,000	≥150,000	50,000	≥150,000
CRP (mg/L)	4	<10	34	<10
Lactate (mmol/L)	1	0.3–2.2	5	0.3–2.2

TABLE 1: Laboratory investigations on day 1 and day 5 of life

Laboratory results of the patient on admission (day 1 of life) and on day 5 of life, at the onset of septic shock. Reference ranges are age-specific values for the first week of life.

WBC: white blood cell; CRP: C-reactive protein

He was nursed in an incubator with minimal handling. The lesions were covered with non-adhesive petrolatum gauze, without adhesive tape. No antibiotics were given on admission, as there were no clinical or biological signs of infection.

From day 1, the nasal lesion extended into an erosion. Erosions of the lips and oral mucosa then appeared (Figure *3*). Gentle friction detached the epidermis, showing a positive Nikolsky sign. No blister appeared at any time. On day 3, he developed non-bilious vomiting after feeds. An abdominal X-ray was normal, with gas distributed throughout the bowel. Abdominal girth remained normal, and no significant gastric residuals were observed. His general condition then worsened.



FIGURE 3: Nasal, labial, and oral mucosal erosions on day 4 of life

Crusted, eroded lesion of the nose with yellowish crusts, and red, eroded areas on the lips and oral mucosa.

On day 5, he developed septic shock with tachycardia, a mean arterial pressure of 30 mmHg, prolonged capillary refill time, and hypothermia. Laboratory tests showed leukopenia with low neutrophil and lymphocyte counts, thrombocytopenia, an increased CRP level, and a high lactate level, while the hemoglobin level remained normal (Table *1*). He received fluid resuscitation and supportive care for septic shock. Blood cultures were taken before the first antibiotic dose, and intravenous imipenem and amikacin were then started, a broad-spectrum regimen chosen because of the septic shock and the bacterial ecology of our unit. The blood culture grew methicillin-susceptible *Staphylococcus aureus* (MSSA), susceptible to all antibiotics tested. The patient died on day 6, before the antibiotic regimen could be de-escalated. Skin swabs taken before day 5 and on day 5 were sterile. Genetic counseling was offered to the parents.

Discussion

Three findings suggested Bart syndrome in our patient: ACC of the limbs, skin fragility from day 1, and the death of a brother with the same lesions. The diagnosis remains presumptive. No blister was seen, and no biopsy was done. Immunofluorescence mapping and genetic testing are needed to confirm the EB type [10]. Similar clinical diagnoses without biopsy have been reported in Africa [9,11].

Outcomes reported in clinically similar cases vary widely. Yerram et al. described a boy of consanguineous parents with ACC of the four limbs, whose two siblings had died in the first week of life; this infant had also been exposed to maternal propylthiouracil, and his outcome was not reported [12]. Yao et al. reported a newborn of parents who were cousins, who died of sepsis and hemorrhage on day 19 [9], and Alwafi et al. reported a preterm infant of consanguineous parents who died on day 19 [8]. Other reported cases, with lesions limited to the legs, had a favorable course with conservative care [11,13]. These cases differ in clinical severity and associated anomalies, and no conclusion on the prognostic role of consanguinity can be drawn from them.

The family described by Bart had autosomal dominant inheritance [3,4]. In our case, the parents were healthy first cousins, and a sibling was affected. This suggests autosomal recessive inheritance, as in recessive dystrophic or junctional EB [5], although a dominant mutation with parental germline mosaicism cannot be excluded without genetic testing. Bart syndrome is therefore a clinical pattern rather than a single disease.

The vomiting raised the possibility of junctional EB with pyloric atresia. This recessive form is linked to *ITGB4* or *ITGA6* mutations and may include ACC [10,14]. Its mortality is high, mainly from sepsis and organ failure [14]. Gastrointestinal atresia has also been reported in Bart syndrome [7]. In our patient, the vomiting was non-bilious, and the abdominal X-ray was normal, with gas throughout the bowel and no isolated distended gastric bubble, which argued against complete pyloric atresia. However, partial obstruction could not be excluded, as no contrast study was performed.

The positive Nikolsky sign and the MSSA bacteremia, whose source was not identified, raised the possibility of staphylococcal scalded skin syndrome. This diagnosis was not retained because the skin defects were present at birth, skin fragility appeared on day 1 before any sign of infection, and an older brother had died with similar lesions. In addition, no diffuse erythema was observed, and exfoliative toxin testing was not performed. Neonatal herpes can cause erosions at birth [7]. TORCH serology was negative, but herpes PCR was not done. Adams-Oliver syndrome and amniotic band sequence were unlikely without scalp defect, limb reduction, or constriction rings.

Infection is a major cause of death [9,14]. Open and fragile skin is a possible route of bacterial entry. In our patient, the source of the MSSA bacteremia was not established, as the skin swabs were sterile. Admission tests were normal, yet MSSA septic shock developed on day 5. Leukopenia and thrombocytopenia indicated severe sepsis. *S. aureus* is a common pathogen in open wounds. Our patient did not receive antibiotic prophylaxis. When infection is suspected in a newborn with extensive skin defects, empirical therapy may need to cover *S. aureus*, according to local guidelines and bacterial ecology. Once MSSA is identified, an anti-staphylococcal penicillin such as oxacillin or cloxacillin is preferred; in our patient, the broad-spectrum regimen was maintained because of the septic shock, and he died before de-escalation could be considered. Regular skin swabs may help guide therapy. Care otherwise remains supportive, with minimal handling, non-adhesive dressings, no adhesive tape, and careful fluid management [7,8].

A precise diagnosis is the most useful outcome for the family. When testing is unavailable, a blood sample collected in ethylenediaminetetraacetic acid can be stored for later analysis, even after death. Genetic counseling can be provided based on the clinical presentation and family history, but targeted prenatal diagnosis requires identification of the causative variant. No sample was kept in our case. The main limitations of this report are the lack of histological and genetic confirmation and the absence of a contrast study.

Conclusions

Bart syndrome should be suspected in a newborn with congenital absence of skin on the limbs and skin fragility, particularly when the parents are consanguineous or a sibling has had similar lesions. This

presentation corresponds to epidermolysis bullosa with congenital absence of skin, and in such families, recessive inheritance is likely, although a dominant mutation with parental germline mosaicism cannot be excluded without genetic testing. Our case shows that the course can be rapidly fatal even when the initial examination and laboratory tests are reassuring. In our patient, infection developed despite minimal handling and non-adhesive dressings, which supports close monitoring for infection in these neonates. When infection is suspected, empirical therapy may need to cover *Staphylococcus aureus*, according to local guidelines and bacterial ecology. As a single case report, these observations cannot support general recommendations. Vomiting may have several causes in this setting, including sepsis, mucosal erosions, or reflux, but pyloric atresia should be considered, and a contrast study may be performed when the plain X-ray is not conclusive. Genetic counseling can be offered on the basis of the clinical findings and family history. When skin biopsy and genetic testing are unavailable, storing a DNA sample before death may allow later identification of the causative variant, which is required for targeted prenatal diagnosis in future pregnancies.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

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Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. Not applicable issued approval Not applicable. Ethics committee approval was not required for this single retrospective case report at our institution. . **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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