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Case Series

Diagnosis, Management, and Outcome of Bart's Syndrome Observed in a Sub-Saharan African Country (Senegal, Dakar): 2 Case Reports

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Abstract
Introduction: Bart's syndrome is an uncommon inherited congenital disorder associating congenital cutaneous aplasia of the extremities and inherited epidermolysis bullosa. Bilateral and symmetrical involvement of the limbs is exceptionally described on black skin. In most cases, the diagnosis is clinical; however, the management remains very difficult and the extended forms are a real therapeutic challenge. We report 2 cases of Bart's syndrome observed in a sub-Saharan African country (Senegal, Dakar). ***Case Presentation:*** It was about 2 premature female and male newborns. On physical examination, the girl presented with a total absence of skin on the limbs, associated with cutaneous detachment of the trunk representing a detached and detachable skin surface of 46%; the boy underwent a total absence of skin of more than 50% of the skin surface. The diagnosis of Bart's syndrome was set based on the typical clinical aspect. The blood count and CRP were normal for the girl whereas it revealed some disorders for the

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boy. The 2 newborns were urgently admitted to an incubator, and the intensive care was started with hyperhydration, anti-staphylococcal prophylaxis, and daily dermatological care with antiseptic baths and fatty dressings. ***Conclusion:*** Bart's syndrome is an uncommon genodermatosis characterized by a clinical triad associating congenital cutaneous aplasia of the extremities, inherited epidermolysis bullosa suspected in the presence of bubbles, and areas of cutaneous fragility and nail deformity. All types of which can be associated with this syndrome. The easy clinical diagnosis but the difficult management encumber the vital prognosis of our cases.

Introduction

Bart's syndrome is a congenital cutaneous aplasia (CCA) of the extremities associated with inherited epidermolysis bullosa. It constitutes type 6 of CCA according to the classification of Frieden in 1986 [1]. It may be due to a genetic mutation of autosomal dominant or autosomal recessive transmission. CCA is defined as a congenital absence of skin tissue, localized or generalized reaching one or more areas [2]. The clinical diagnosis of Bart's syndrome is generally easy, characterized by the presence of well-limited ulcerated translucent membranes, with visualization of the underlying structures at the extremities associated with epidermal skin detachment (positive Nikolsky sign) [1, 3]. However, it poses a challenging management in our context because it is rapidly life threatening when the attack exceeds 30% of the skin surface by the risk of dehydration, hypothermia, infection, and hypoprotidemia. It is a disease whose description in the literature remains exceptional [4, 5]. In sub-Saharan Africa, 2 cases of Bart's syndrome were reported in Côte d'Ivoire [6] and a case in Nigeria [7]. In Senegal, particularly in Dakar, to our knowledge, this work constitutes the first case report on Bart's syndrome.

Case Presentation

The CARE Checklist has been completed by the authors for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000535038).

Case 1

The female newborn was referred from the health center on day 0 of life for an absence of skin on the 4 limbs observed from birth. Second-degree parental consanguinity was found. The physical examination on admission found a newborn with a weight of 2,800 g, a height of 49 cm (−3DS), a head circumference of 36 cm (+2DS), a temperature of 35.5°C, a respiration rate of 39 cycles/min, spo2 99% in ambient air, a heartbeat rate at 128 beats/min. The dermatological examination outlined a total absence of skin leaving bare a vascular network on almost all of the upper and lower limbs, associated with flabby bubbles with hemorrhagic skin detachment and positive Nikolsky's sign on the trunk, buttocks, wings of the nose, and the pavilion of the ears. There were also ulcerations of the oral mucosa and anonychia of the fingers and toes with vernix caseosa covering the areas without cutaneous aplasia (shown in Fig. 1). The involved skin surface was estimated at 46%. The rest of the examination was

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Fig. 1. Congenital skin aplasia of the limbs, skin detachment with positive Nikolsky sign on the trunk covered with vernix caseosa, anonychia. Body surface affected = 46%.

unremarkable and in particular without malformation. The diagnosis of Bart's syndrome was thus retained. Cutaneous histopathology of a bulla revealed a subepidermal bulla suggesting dystrophic epidermolysis. Direct immunofluorescence of skin bulla biopsy has not been realized due to its unavailability in our country. The newborn was urgently installed under a heated table due to a lack of space in an incubator. The child received intravenous prophylactic antibiotic therapy with cefotaxime 210 mg × 2/d and amikacin 45 mg/day. Daily local care was performed with a bath of chlorhexidine solution × 2/day, dressings with greasy tulle. The complete blood count noted a hemoglobin level at 14 g/L, leukocytes at 3.87 × 103 elements/mm^3, and platelets at 288,000 elements/mm^3. The CRP was negative. The death occurred on day 6 of hospitalization with an underlying hypothermia condition.

Case 2

It was a male newborn, received at 5 h of life who was referred from a health district for congenital skin absence. Second-degree parental consanguinity was found. The mother was 24 years old, second gravida, primiparous, an abortion. It was a premature newborn at 33 weeks of amenorrhea and 2 days. There was a 1-h premature rupture of the membranes. The child reportedly cried at birth with an Apgar 7/10 at the first minute and 8/10 at the fifth minute. The examination found the following parameters and measurements: hypothermia at 32.6°C, heartbeat rate at 119 beats/min, respiratory rate at 46 cycles/min, oxygen saturation at 100%, capillary hypoglycemia at 0.37 g/dL, a cranial circumference at 33 cm (DS), an arm circumference at 9 cm, a thoracic circumference at 28 cm, a height at 46 cm, a weight at 1,900 g (−3DS). Dermatological examination revealed cutaneous aplasia of the 4 limbs with an extensive cutaneous detachment of the anterior trunk, face, and skull (shown in Fig. 2). The involved skin surface was estimated at 50%. There was anonychia and erosions of the oral mucosa. The diagnosis of Bart's syndrome was retained based on the characteristic clinical course. Intensive care was performed with an incubator admission, a bolus of SG10% at a rate of 3 cc/kg, then an infusion of SG10% on vein guard. Blood count, urea, and serum creatinine were normal. The blood electrolyte measurement found hyponatremia at 119 mmol/L corrected by a bolus of NaCl 3% at a rate of 3 cc/kg in 30 min, antibiotic coverage based on vancomycin 40 mg/kg in four doses. He was also administered paracetamol 7.5 mL every 6 h. Local care was carried out with

Fig. 2. Congenital aplasia cutis with visualization of underlying structures, skin detachment with positive Nikolsky sign on the trunk and face covered with vernix caseosa, anonychia. Body surface affected = 50%.

potassium permanganate in a bath in a basin of water at 37°C, greasy tulle over the entire body surface devoid of skin, then sprayed with chlorhexidine solution. The evolution was marked by the persistence of hypothermia and the appearance of new areas of detachment. The death occurred on day 2 of life due to hypothermia.

Discussion

We reported 2 cases of Bart's syndrome observed in a pediatric dermatology department in Dakar. These observations are particularly interesting to report for several reasons. First, because of the scarcity. Indeed, Bart's syndrome is an exceptionally reported genodermatosis worldwide [5], especially in sub-Saharan Africa where 2 cases have been reported in Côte d'Ivoire [6] and 1 case in Nigeria [7]. Diagnosis is generally easy because it is a syndrome characterized by the clinical triad: CCA of the extremities, inherited epidermolysis bullosa regardless of the type, and nail abnormality [3]. The pathophysiology is still poorly understood; a recent study has shown that the *BMS1* gene plays a role in the occurrence of this anomaly [8]. However, the inherited epidermolysis bullosa with which it is associated also determines the type of mutation involved. Therefore, as it can be associated with all types of inherited epidermolysis bullosa, several genetic mutations can be sought. However, our patients presented with a dystrophic form of epidermolysis bullosa, which is linked to a mutation of collagen VII (*COL7A1*) [9]. In addition, our patients have a recessive form of dystrophic epidermolysis bullosa due to the absence of a family history of inherited epidermolysis bullosa in the patients. This pattern is rarely reported because in Bart's syndrome, it is a dystrophic epidermolysis bullosa in its autosomal dominant form, which is reported more than the autosomal recessive form [10]. Bart's syndrome is considered to be an extremely uncommon genetic disease. Multiple skin involvement is extremely rare [5]. Our case of Bart's syndrome presented with a bilateral and symmetrical CCA topography of the extremities, reaching, respectively, 46% and 50% of the body surface associated with a nail anomaly such as anonychia, and flaccid bubbles in the first case. In our African context, the difficulties reside because of the confusion with several neonatal bullous dermatoses such as staphylococcal epidermolysis bullosa, cutaneous candidiasis, neonatal syphilis, and other genodermatoses but above all

the lack of diagnostic means, which limit certain explorations such as immunostaining, electron microscopy, genetic tests, which make it possible to determine the type of genetic mutation in question. In some cases, the diagnosis of Bart's syndrome has been made based on the clinical presentation as described by Alfayez et al. [5] in 2017 about a case. In our two observations, the diagnosis was made based on the clinical presentation; however, sporadic cases were also reported. This syndrome can also be associated with other extra-cutaneous abnormalities with digestive, genito-urinary, and ocular mucosal involvement [2]. None of these abnormalities were found in our cases. The management of this condition is not dermatologically standardized and various local care is offered with the application of petroleum jelly, silver sulfadiazine, infusions of saline solution, and dressing changes twice a day. In our case, twice-daily local care with antiseptic and fatty dressings was used associated with intensive care measures with placement in an incubator in patient 2 and under a heated table in patient 1 for lack of an incubator with death observed despite all these intensive care measures. The prognosis of Bart's syndrome depends on the severity and extension of the cutaneous aplasia, the subtype of associated inherited epidermolysis bullosa, the existence of an associated malformation, and the success of the treatment [5]. So, there are different evolving modalities in the literature. Indeed, kulali et al. [11] reported a case with a good evolution while Ahogo et al. [7] noted a fatal evolution despite good management. In our patients, a fatal evolution was noted in both cases. This could be related to the type of associated dystrophic epidermolysis bullosa but also to the extent of the loss of skin barrier function with a body surface area of 46 and 50%.

Conclusion

Bart's syndrome is a genodermatosis rarely reported worldwide and in sub-Saharan Africa. The particularity of the cases that we report lies in the extent of the skin lesions leading to a loss of the crucial barrier function for the newborn in the first days of life. This case report allows us to draw the attention of practitioners for a better knowledge of this syndrome to take care of it quickly and adequately to limit deaths in our practice context. In the future, we plan to search for the genetic mutations involved in our regions if the case arises again in our practice.

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Statement of Ethics

Ethical approval is not required for this study by local guidelines. Written informed consent was obtained from the parent of the patients for publication of the details of their medical case and any accompanying images.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Mame Tènè Ndiaye Diop: contribution to medical care, article writing, and submission. Maïmouna Bassoum: contribution to medical care and article writing. Khadim Diop, Yaye Diood Dieng, Fatou Diasse Fall, and Charles Tchibinda Delicat: contribution to medical care. Birame Seck, Alassane Ndiaye, Assane Diop, Maodo Ndiaye, Pape Moctar Faye, Moussa Diallo, Ousmane Ndiaye, and Fatimata Ly: contribution to article writing. Suzanne Oumou Niang: contribution to article writing and supervised the medical care.

Data Availability Statement

All data generated or analyzed during this study are included in this article and its online supplementary material. Further inquiries can be directed to the corresponding author.

References

1 Frieden IJ. Aplasia cutis congenita: a clinical review and proposal for classification. J Am Acad Dermatol. 1986;14(4):646–60.
2 Belkhou A, François C, Bennis Y, Duquennoy-Martinot V, Guerreschi P. Aplasia cutis congenita: mise au point et prise en charge [Aplasia cutis congenita: update and management]. Ann Chir Plast Esthet. 2016;61(5):450–61.
3 E Rogvi R, Sommerlund M, Vestergaard ET. Aplasia cutis congenita is a rare and possibly overlooked congenital anomaly. Ugeskr Laeger. 2014;176(48):V05140276. Danish.
4 Itto AB, Lehlimi M, Habzi A, Benomar S. Bart's syndrome: a new neonatal observation. J Pediatr Child. 2013;26:232–5.
5 Alfayez Y, Alsharif S, Santli A. A case of aplasia cutis congenita type VI: Bart syndrome. Case Rep Dermatol. 2017;9(2):112–8.
6 Ogundipe KO, Kadiri IA, Oluwayemi IO, Ogundare EO, Wash YS. Wuraola IO Aplasia cutis congenita: dilemma of management in a resource-limited scenario. J Surg. 2020;5:1274.
7 Oriji PC, Afolabi SA, Omietimi JE, Allagoa D, Doni Doni P, Ozori SE, et al. Aplasia cutis congenita: uncommon finding of 2 cases occurring in one patient in two successive deliveries. Yen Med J. 2019;1(1):61–5.
8 Marneros AG. BMS1 is mutated in aplasia cutis congenita. PLoS Genet. 2013;9(6):e1003573.
9 Bardhan A, Bruckner-Tuderman L, Chapple ILC, Fine JD, Harper N, Has C, et al. Epidermolysis bullosa. Nat Rev Dis Primers. 2020;6(1):78.
10 Al-Gburi S, Namuq Z. Twin neonates with Bart's syndrome. Cureus. 2022;14(1):e21363.
11 Kulalı F, Bas AY, Kale Y, Celik IH, Demirel N, Apaydın S. Type VI aplasia cutis congenita: Bart's syndrome. Case Rep Dermatol Med. 2015;2015:549825.