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Association of Congenital Cutaneous Aplasia and Congenital Bullous Epidermolysis Bart Syndrome: A Case Report

I. Agouzoul[1*], A. Ayad[1], M. Sellouti[1], M. Bahous[1], A. Agadr[1], R. Abilkassem[1]

[1]Pediatrics and Neonatology Service, Mohamed V Military Hospital, Rabat, Marocco

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***Corresponding author:** I. Agouzoul
Pediatrics and Neonatology Service, Mohamed V Military Hospital, Rabat, Marocco

Abstract **Case Report**

Background: Bart syndrome is a rare neonatal condition that combines congenital skin aplasia and congenital bullous epidermolysis. The clinical diagnosis is straightforward, but its management is intricate, and the extensive forms pose a true therapeutic challenge. We report a case of a newborn diagnosed in the pediatric intensive care unit at the Military Hospital of Rabat. ***Case Description:*** A female newborn from a well-followed pregnancy, delivered via cesarean section due to a narrow pelvis. The Apgar score was 10/10, and the birth weight was 2600g. The mother had been diagnosed with hypothyroidism during pregnancy and was taking synthetic antithyroid medication. The infant was admitted to the neonatal unit at 2 hours of life due to multiple skin ulcers noted at birth. Clinical examination revealed bilateral and symmetrical cutaneous aplasia affecting almost the entirety of both lower limbs. The vascular network was clearly visible. Other parts of the skin showed areas of skin fragility and some flaccid blisters. A clinical diagnosis of Bart syndrome associated with bullous epidermolysis was made. Hydration and antibiotic coverage were initiated, leading to good clinical improvement. ***Discussion:*** Bart syndrome represents clinical features of aplasia cutis congenita associated with congenital epidermolysis bullosa suspected by areas of fragility and sometimes by blisters. All types of congenital epidermolysis bullosa can be associated with this syndrome. Clinical diagnosis is usually easy, but the therapeutic management is difficult, and the prognosis is poor.

Keywords: Aplasia cutis, Congenita, Newborn, Black, Skin.

BACKGROUND

Bart syndrome is a rare neonatal condition that combines congenital skin aplasia and congenital bullous epidermolysis. The clinical diagnosis is straightforward, but its management is intricate, and the extensive forms pose a true therapeutic challenge. It was first described in a family by Bart et al., in 1966 [1]. The pathophysiology is still poorly understood. The goal of management is to prevent secondary complications and prepare the ground for potential reconstructive surgery. The prognosis is generally good and directly depends on the proper management of skin lesions. We report a case of a newborn diagnosed in the pediatric intensive care unit at the Military Hospital of Rabat.

CASE DESCRIPTION

A newborn female infant born to non-consanguineous parents. The mother was a 24-year-old primigravida, had an uncomplicated pregnancy managed to full term. She underwent a cesarean delivery due to a narrow pelvis. The Apgar score was 10/10, and the birth weight was 2600g. The mother had been diagnosed with hypothyroidism during pregnancy and was on synthetic antithyroid medication. The infant was admitted to the neonatal unit at 2 hours of life due to multiple skin ulcers noted at birth.

The clinical examination found a full-term, eutrophic newborn, with a weight of 2600g, a length of 49cm (within average range), and a head circumference of 33cm (within average range). Hemodynamically, the baby was stable, with a skin recoloration time of less than three seconds. The heart rate was normal at 145 beats/min, and the respiratory rate was 40 cycles/min.

Skin examination (Fig 3 & 4) revealed bilateral and symmetrical cutaneous aplasia affecting almost the entirety of both lower limbs. The vascular network was clearly visible. Other parts of the skin showed areas of skin fragility and some flaccid blisters. Over the first few days of life, the condition progressed to the formation of blisters followed by post-blister erosions on the cheeks, forehead, chest, upper limbs,

lower limbs, buttocks, and oral mucosa (Fig 1). The remainder of the clinical examination was unremarkable. Infectious workup came back normal. A clinical diagnosis of Bart syndrome associated with bullous epidermolysis was made.

Treatment was initiated with hydration to counter fluid losses and daily care, including applying occlusive dressings. The patient showed excellent clinical improvement. The lesions dried out, and skin re-epithelialization began in areas affected by epidermolysis.

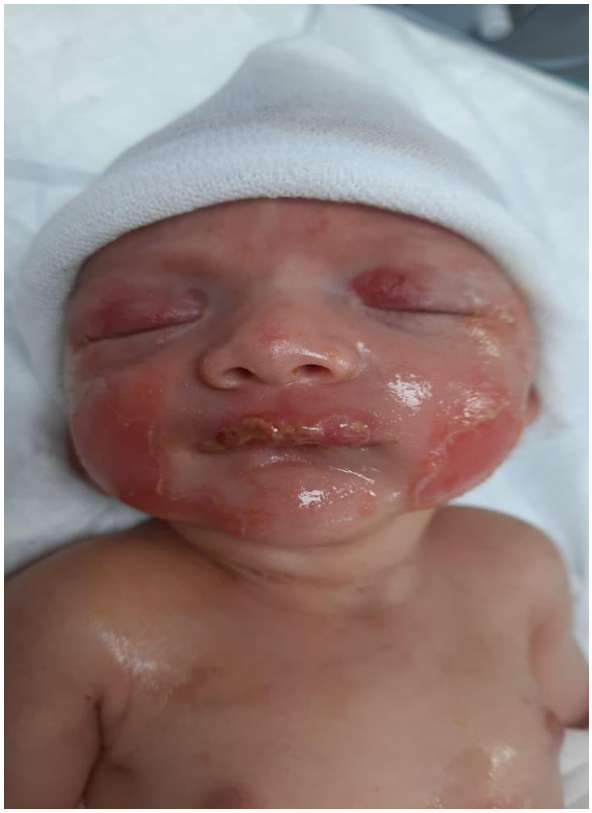

Figure 1: Picture of the newborn at birth

Figure 2: Picture of the newborn after clinical improvement

Figure 3: Congenital cutaneous aplasia on the feet

Figure 4: Drying out and initiation of skin lesion healing

Figure 5: Extended post-bullous erosions

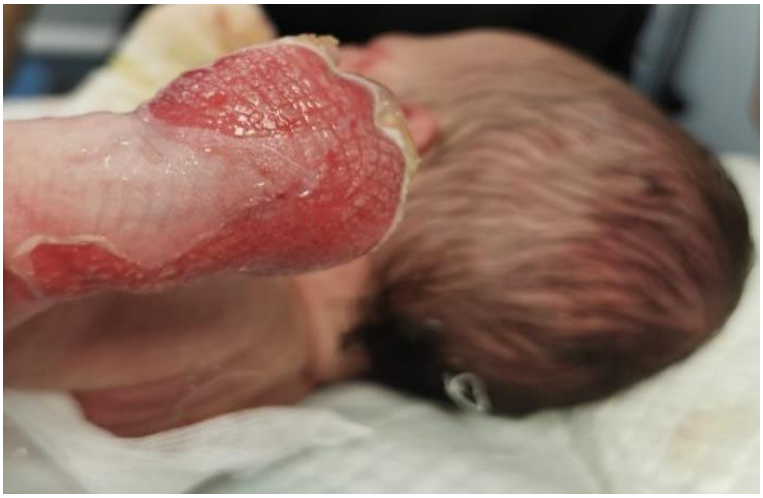

Figure 6: Extended post-bullous erosions

DISCUSSION

Bart syndrome corresponds to a clinical presentation of congenital cutaneous aplasia associated with suspected congenital bullous epidermolysis, indicated by areas of fragility, and sometimes blisters. Congenital cutaneous aplasia is a rare dermatosis with 9 clinical-genetic types. It is often sporadic but familial cases have been reported. This congenital defect is generally limited to the vertex in 85% of cases. Limb involvement is uncommon, generally bilateral, and symmetrical but less extensive compared to our case. Aplastic lesions are acquired in utero through mechanisms likely differing by type. Several pathophysiological hypotheses have been proposed: genetic, vascular, infectious, teratogenic, as well as amniotic band syndrome and fetus papyraceus syndrome. Extended and fatal forms are rare [2]. Genetic and environmental factors have been implicated in ACC development. Some ACC forms have been linked to specific genetic mutations [9], such as anomalies in the TP63 and SATB2 genes. These mutations can disrupt normal skin development, leading to congenital cutaneous aplasia that might be associated with minor malformations or syndromes. Our patient showed no other malformations, but the aplastic lesions were associated with bullous epidermolysis lesions. This lesion combination constitutes Bart syndrome [3].

Congenital bullous epidermolyses (CBE) are a group of rare genetic diseases affecting the skin and mucous membranes. They are characterized by extreme skin fragility, resulting in blister and vesicle formation upon mild friction or trauma.

The pathophysiology of CBE is primarily linked to abnormalities in structural skin proteins [4], notably intercellular junction proteins. CBE is mainly caused by mutations in genes encoding these proteins.

There are several types of congenital bullous epidermolyses, including simple epidermolysis bullosa (EB), junctional epidermolysis bullosa (JEB), and dystrophic epidermolysis bullosa (DEB). EBS features intra-epidermal cleavage within the basal layer, EBJ involves cleavage at the level of the basal membrane itself, and EBD features cleavage below the basal membrane, specifically at the anchoring fibrils of the superficial dermis. Each type of EB is associated with specific mutations [9]. In junctional epidermolysis bullosa, mutations affect proteins at the dermo-epidermal junction, such as laminin-332, collagen XVII, and integrin alpha-6/beta-4. In dystrophic epidermolysis bullosa, mutations affect collagen VII, which is essential for dermo-epidermal junction stability [9].

These mutations alter protein structure or function, leading to reduced skin mechanical resistance. Consequently, the skin of individuals with CBE is extremely fragile, prone to lesions and blisters [5].

Research in the field of congenital bullous epidermolysis pathophysiology aims to better understand underlying mechanisms, identify new therapeutic targets, and develop more effective treatments for this rare and debilitating condition. Spontaneous healing in the form of atrophic scarring, sometimes intrauterine, is possible. Extended and fatal forms are rare. In Bart syndrome, aplastic lesions are often extensive and generally located bilaterally and symmetrically on the lower limbs [7]. The association of these extended lesions with congenital bullous epidermolysis lesions and sometimes other malformations makes managing this syndrome very challenging. Therapeutic management must be carried out in a hospital setting, with careful control of infection risk, anemia, and malnutrition.

Symptoms and severity of congenital bullous epidermolysis can vary significantly depending on the specific type and mutations involved [8]. Treatments generally aim to relieve symptoms, prevent infections, promote wound healing, and improve patients' quality of life. Proper skin care is crucial to prevent lesions and

infections. Creams and ointments containing moisturizing agents, topical antibiotics, and wound-healing agents can be used to protect the skin, prevent infections, and promote wound healing.

It's important to note that the treatment of CBE and ACC is often multidisciplinary and requires an individualized approach based on the specific type of CBE, the severity of symptoms, and associated complications.

CONCLUSION

Bart syndrome presents as a clinical manifestation of congenital cutaneous aplasia associated with congenital bullous epidermolysis, the etiopathogenesis of which remains poorly understood, making it challenging to establish prenatal diagnostic strategies or provide appropriate genetic counseling. The management is multidisciplinary and requires an individualized approach based on the severity of lesions and associated malformations.

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