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RESEARCH ARTICLE

A CASE REPORT OF APLASIA CUTIS CONGENITA TYPE VI: BART'S SYNDROME

Ahmed Mead¹, Yordanos Alem², Omar Adam Sheikh³ and Layla Ibrahim Hussein⁴

1. Faculty of Medicine, Department of Basic Medical Science, Salaam University, Mogadishu, Somalia.
2. College of Health Science, School of Medicine, Addis Ababa University, Addis Ababa, Ethiopia.
3. Faculty of Medicine, Department of Basic Medical Science, Somali National University, Mogadishu, Somalia.
4. Banadir Maternity and Children Hospital, Mogadishu, Somalia.

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Abstract

Aplasia cutis congenita type VI (Bart's syndrome), is a rare genetic disorder characterized by congenital localized absence of skin, formation of blisters (epidermolysis bullosa), and nail abnormalities. In this report, we present a rare case of aplasia cutis congenita type VI (Bart's syndrome) in a newborn male baby with the absence of a skin layer over the anterior right leg, slightly below the patella (kneecap) and around the ankle joint. On the second day, the affected areas developed blisters characterizing epidermolysis bullosa. Laboratory investigations were all normal. The patient's wound was managed conservatively with dressing and topical antibiotic ointments.

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Introduction:-

Aplasia cutis congenita (ACC) is an inborn skin defect that typically presents as isolated or multiple lesions characterized by the absence of skin layers and in some cases underlying structures such as subcutaneous tissue and bone. In almost 85% of the cases, the defect is most commonly seen on the scalp localized to the scalp vertex; however, it can also affect other body parts like the face, trunk, and limbs [1, 2].

The reported incidence of ACC is 3 per 10,000 births with a predisposition to female patients [2]. The typical lesion is non-inflammatory, small in size, and well-circumscribed, with a variety of configurations. The lesions may be linear, oval, circular, or stellate, membranous, or non-membranous [3]. The underlying etiology of ACC is multifactorial and not well understood; genetics and other factors like vascular accidents, exogenous teratogenic substances likely play a role in the etiology [2].

A classification system was proposed by Frieden in 1986, which classified ACC into nine main types on the basis of location and pattern of the skin defect, and presence of associated anomalies as shown in Table 1 [4].

Table 1:- Frieden's classification of ACC [4].

Type	Clinical Features	Body area affected
I	Scalp ACC without multiple anomalies	Scalp, usually vertex
II	Scalp ACC with associated limb abnormalities	Midline scalp
III	Scalp ACC with associated epidermal and organoid nevi, corneal opacities, and psychomotor retardation	Scalp, may be asymmetric

Corresponding Author:- Ahmed Mead

Address:- Faculty of Medicine, Department of Basic Medical Science, Salaam University, Mogadishu, Somalia.

IV	ACC of any site with overlying embryologic malformations such as meningomyeloceles, spinal dysgraphia, omphalocele, and gastroschisis	Any site, more commonly on the abdomen, lumbar skin, scalp
V	ACC with associated fetus papyraceus or placental infarcts	Multiple, symmetric areas, often stellate or linear, on scalp, chest, flanks, axillae, and extremities
VI	ACC of the extremities associated with epidermolysis bullosa	Extremities
VII	ACC localized to extremities without blistering	Pretibial areas; dorsal aspects of hands and feet; extensor areas of wrists
VIII	ACC caused by specific teratogens such as varicella and herpes simplex infections or in association with methimazole treatment	Scalp (with methimazole); any area (with varicella and herpes simplex infections)
IX	ACC associated with malformation syndromes such as Trisomy 13, ectodermal dysplasias; and amniotic band disruption complex	Any site, more commonly scalp

ACC may also occur as one of the key characterizing features of other multisystem disorders, including Adams-Oliver Syndrome, Bart's syndrome, and Setleis syndrome. Adams-Oliver Syndrome is a multisystem disorder of unknown etiology characterized by the combination of congenital scalp defects (ACC) and terminal transverse limb defects [5]. Bart's syndrome, also known as aplasia cutis congenita type VI, is a rare genetic disorder characterized by ACC with epidermolysis bullosa [6]. Setleis syndrome is one of the ectodermal dysplasias that are characterized by ACC with facial skin abnormalities [7].

The Pathophysiology of ACC is not well known; however, there are multiple factors that contribute to the cause and progression of the disease:

1. Genetic (chromosomal abnormalities): BMS1 gene
2. Physical trauma
3. Intrauterine infections and vascular incidents
4. Thrombosis
5. Exposure to teratogenic substances such as antithyroid drugs, methotrexate, valproic acid, cocaine, and alcohol during pregnancy [1, 8].

In this report, we present a rare case of aplasia cutis congenita type VI (Bart's syndrome) in a newborn male baby with the absence of a skin layer over the anterior right leg, slightly below the patella (kneecap) and around the ankle joint.

Case Report

We obtained informed permission from the patient's mother and family in the Department of Obstetrics and Gynecology, Banadir Maternity and Children Hospital, Mogadishu, Somalia. A newborn male baby was born to a 35-year-old gravida 7, para 5, plus 2 (G7P5+2). The baby was born at 38 gestational weeks through normal vaginal delivery without any obstetric complications. The Apgar score was 10. The parents were not relatives, but there was a family history of skin abnormalities. The newborn's older brother, two of his cousins, two of his aunts, and his grandfather had a history of absent skin in the lower extremities.

Weight, length, head circumference, and vital signs were normal and the baby was admitted to the neonatal intensive care unit (NICU) of the hospital due to skin lesions. On physical examination, the baby had an absence of a skin layer over the anterior right leg, slightly below the patella (kneecap) and around the ankle joint as shown in Figure 1. The affected areas are seen to have red thin mucous membranes. The scalp, mucous membranes, and systemic examination were normal. On the second day, the affected areas developed blisters characterizing epidermolysis bullosa. Results of the laboratory investigations such as complete blood count (CBC), electrolytes, liver and renal function tests were all normal. The baby's wound was treated conservatively with dressing and topical antibiotic ointment. After five days the baby was discharged and the mother was informed to complete conservative wound treatment at home.



Figure 1:- Aplasia cutis lesions in the right leg.

Discussion:-

According to the classification proposed by Frieden in 1986, ACC type VI also known as Bart's syndrome is a rare genetic disorder characterized by ACC with epidermolysis bullosa [7]. ACC can be unilateral or bilateral [9]. In our patient, there was a congenital absence of skin layer in the right leg with epidermolysis bullosa. In some patients, pyloric atresia, ureteral stenosis, and renal abnormalities can be seen [10]. In this case, these congenital anomalies were not present.

The best pathophysiologic explanation for ACC states that the skin defect is a result of rapid brain growth which induces tension in the fetal skin and subcutaneous tissue resulting in skin disruption at around 10–15 weeks of gestation, with the scalp being the most tensioned area. Other less relevant hypothesis state amniotic irregularities as a possible cause for ACC [1]. In some cases, the scalp is not affected [11]. Similarly, in our patient, the scalp was not affected.

Clinical presentation is the basis for the diagnosis of ACC, although it can be diagnosed earlier using prenatal ultrasound, it is usually discovered after delivery as the lesion is visible and presents as a thin layer or scab with the absence of hair follicles [12].

Management of ACC ranges from conservative management to immediate surgical interventions based on the size, location, and extent of involvement [2]. Conservative treatments are for smaller-sized defects (less than 2 cm) with no additional findings that spontaneously close through epithelialization and approximation from the edges thus daily cleaning of the lesion with antimicrobial emollient is recommended until healing is complete which takes from few weeks to months leaving an atrophic cicatrix behind. On the other hand, larger defects (greater than 4 cm) are more commonly associated with underlying defects, thus early surgical repair with either a skin graft or a flap is recommended. Both conservative and surgical management are options to treat defects between 2 and 4 cm [8, 12].

In cases of lesions with bony involvement, the defect is managed with flaps or grafts until 2 years of age, as the reconstruction of bone cannot be attempted till then. Occasionally, the bony defect might spontaneously resolve as a

result of the osteogenesis. In addition, coverage of the defect using local flaps and reconstruction of hair patterns through tissue expansion of the scalp might be necessary for aesthetic purposes at a later age [12].

Large defects that involve the bone especially around the vertex pose a great risk of developing fatal complications like sagittal sinus hemorrhage or thrombosis, CNS infection, meningitis, and brain herniation. The mortality rate of ACC is strongly related to the size, location, and depth of the defect. For extensive scalp defects, it can get as high as 55%. The cause of death is mainly attributable to sagittal sinus hemorrhage [2].

Isolated ACCs without any other underlying defect have a relatively good prognosis with benign outcomes. However, when there is an underlying defect or an associated anomaly, the prognosis and mortality become entirely dependent on the course and complication of the underlying defect or associated disorder [8].

Conclusion:-

ACC is an inborn skin defect that typically presents as isolated or multiple lesions characterized by the absence of skin layers. The typical lesion is non-inflammatory, small in size, and well-circumscribed. The underlying etiology of ACC is multifactorial and not well understood. Genetics and other factors play a role in the etiology. Management of ACC ranges from conservative management to immediate surgical interventions based on the size, location, and extent of involvement. The prognosis depends on the course and complication of the underlying defect or associated disorder.

Conflict of Interests

The authors declare no conflict of interests.

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