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Case Report

A Rare Case of Abdominal Skin Malformation

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Abstract

Case description: We describe the clinical case of a female newborn, result of a monitored, full-term pregnancy, who presented at birth with an atrophic lesion on the abdominal wall, measuring around 5cm and with visualization of the underlying vasculature. There were no other alterations on objective examination. Aplasia cutis congenita (ACC) was hypothesized and corroborated by dermatology. Abdominal, transfontanellar ultrasound and echocardiogram were requested with no associated anomalies. No family history of similar skin lesions. The approach was conservative with the application of an emollient and there was a favorable evolution, with gradual regression of the lesion. **Conclusion:** ACC is a rare entity characterized by focal or generalized absence of skin. Since mild cases may not be reported, its incidence may be underestimated. The appropriate approach to this entity depends on its characteristics and severity, and a careful evaluation should be carried out to identify and guide associated malformations at an early stage. Because of the possible implications in terms of counseling, a thorough family history is important. Small, isolated lesions usually resolve spontaneously, and complications are rare.

Keywords: Aplasia Cutis Congenita; Neonatology; Dermatopathology; Congenita Malformation

Case presentation

A female neonate, result of a monitored, full-term pregnancy, delivered at term, presented at birth an incidental discovery of a mid-abdominal atrophic plaque with a parchment-like texture, measuring around 5 cm, that allowed visualization of the subjacent vasculature (Figure 1). Dermatological evaluation promptly made the diagnosis of aplasia cutis congenita (ACC). To ensure a thorough examination and rule out any concurrent malformations, renovesical, transfontanellar, and cardiac ultrasounds were immediately requested. No associated anomalies were found. A conservative management approach based on supportive care and close monitoring was implemented. Emollients were the only treatments prescribed, allowing the natural healing process to unfold. Closed follow-up by pediatric dermatology confirmed a satisfactory progression. The lesion underwent progressive healing, as evidenced by a consistent reduction in size. (Figure 2), which led to the patient's discharge from dermatological care. Nowadays, the child continued monitoring in the neonatology department ensuring ongoing observation of the patient's long-term well-being.

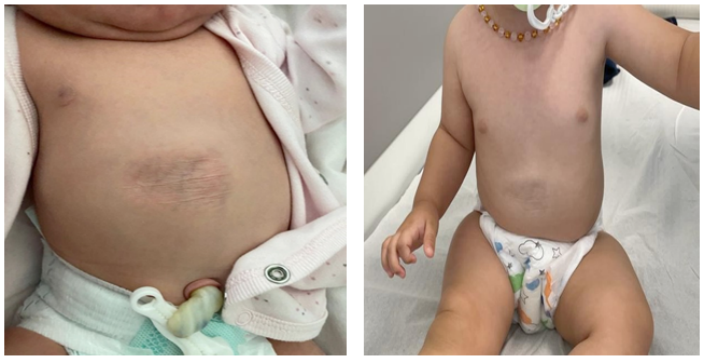

Figure 1, 2: Abdominal Skin Malformation.

Discussion

ACC is a rare skin disorder, characterized by the absence or thinning of skin layers, primarily affecting the scalp, limbs, or abdominal wall [1]. It presents as localized areas of missing skin, ranging from small lesions to large defects. In most cases, the diagnosis of ACC is clinical, and abdominal wall involvement is rare, occurring predominantly in the mid-abdominal region [1,5]. The etiology of aplasia cutis remains uncertain and is believed to result from a combination of genetic and environmental factors [1]. Although it is an isolated finding in most cases, it can be associated with a few genetic syndromes and congenital anomalies (Including abdominal wall defects, limb abnormalities, cleft deformities, and fetus papyraceus). Therefore, it is important to differentiate aplasia cutis from other skin disorders, as the underlying cause may dictate the prognosis and management strategies. In the majority of cases, as in ours, there is no previous history of ACC in the family. The management of this condition depends on various factors and the treatment options range from conservative measures to surgical interventions. In this neonatal case, the early detection of the condition through dermatological examination enabled prompt consultations with relevant specialists to rule out any associated internal malformations and conservative measures were immediately initiated. With this report we explore the serendipitous discovery of aplasia cutis in an otherwise healthy female newborn, focusing on the rare abdominal wall manifestation, reviling the natural healing potential of aplasia cutis, which may lead to new therapeutic interventions and targeted approaches to promote healing and tissue regeneration.

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