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Congenital Cutis Aplasia: A Case Report from Barranquilla - Colombia

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Abstract: ***Congenital cutis aplasia (CCA) is a rare congenital condition characterized by the absence of areas of skin or deeper tissues. It may present as a single lesion or multiple lesions in one or several locations on the body surface, and it may or may not be associated with other syndromes or pathologies. Although it is a rare condition with limited information available to clarify its pathogenesis and identify risk factors, cases described suggest that it is a multifactorial entity with an important genetic component. A case is presented of a 38-week-old female newborn (Ballard score) with a single lesion on the scalp, which was diagnosed clinically and evaluated by various medical specialties.***

Keywords: Cutis aplasia, congenital malformation, dermatology.

1. Introduction

Congenital anomalies are a major cause of morbidity and mortality worldwide, making the study of these conditions, their clinical presentation, and the identification of maternal and neonatal risk factors crucial for early diagnosis and effective management. Although congenital cutis aplasia (CCA) is a rare and poorly documented pathology, case reports have documented it as a congenital absence of areas of the epidermis, dermis, and subcutaneous tissue, sometimes involving muscle and bone. While it can affect any region of the body, its most common location is the scalp, specifically the vertex area of the skull [1,2].

This anomaly typically presents as a single defect but may also appear as multiple lesions. These lesions are non-inflammatory and well-demarcated. While most of these scalp defects occur sporadically, many familial cases have been described [2]. Treatment of CCA depends on the size and location of the lesion. When the lesions are not extensive, conservative management is preferred, allowing for gradual epithelialization of the skin and the formation of an atrophic scar without hair over several weeks [3].

Congenital anomalies often present a diagnostic challenge due to their low incidence and the limited availability of previous clinical records. Recording these cases allows for the recognition of trends and specific characteristics that help facilitate earlier and more accurate diagnoses.

2. Methodology

This case involves a term newborn with congenital cutis aplasia on the scalp, with significant family history of CCA, as both the mother and maternal grandmother had similar lesions at birth in the same location. In this case, an interdisciplinary approach was employed, involving the services of neurosurgery, dermatology, and plastic surgery.

3. Case Report

A 38-week-old newborn (Ballard score), daughter of a 20-year-old first-time mother, born via cesarean section due to failed labor progression. Birth weight: 2,480 g, birth length: 47 cm, good neonatal adaptation, normal prenatal ultrasounds, and a negative maternal infectious profile. The mother had a history of communication disability (hearing and speech impairment) and cutis aplasia. Family history: maternal line, cutis aplasia (maternal grandmother and maternal aunts). Upon birth, a single lesion was detected on the scalp, approximately 5x4 cm in size, located in the occipital region with absence of skin, without associated dysmorphism (Figure 1). The patient was transferred to the neonatal intensive care unit (NICU) at the Hospital Adelita de Char Mired IPS for interdisciplinary management and follow-up. Upon admission, paraclinical tests were ordered, all within normal limits, and diagnostic imaging was performed, including a simple cranial CT scan, which reported hyperdensity at the interhemispheric fissure towards the posterior aspect, prompting the decision to conduct a follow-up cranial CT scan to rule out a potential hemorrhagic component.

At 24 hours of life, the lesion on the scalp changed color, becoming more hyperemic (Figure 2). The initial treatment consisted of applying fusidic acid to the lesion as infection prophylaxis. A consult was requested with the plastic surgery, dermatology, and pediatric neurosurgery departments.

These specialists respectively recommended wound care with Aquacel dressings every 4 days for 32 days, wet compresses with Domeboro for 20 minutes, fusidic acid, and one drop of vitamin C daily. A follow-up cranial CT scan was performed on day 7 of life, with no pathological findings. Consequently, the neurosurgery service recommended outpatient management and follow-up.

Before discharge, the lesion showed a decrease in size and was in the process of healing, with scabs and less hyperemic areas (Figure 3). The patient did not experience any complications during the hospital stay and was discharged with outpatient management and interdisciplinary follow-up.

Figure 1: Lesion on the scalp in the first hours of life.

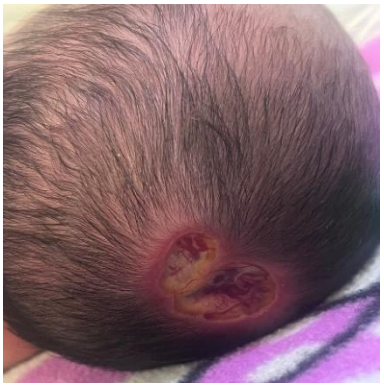

Figure 2: Lesion on the scalp at 24 hours of life.

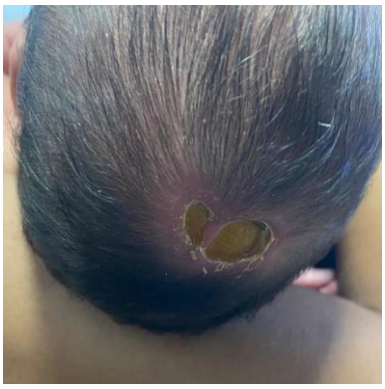

Figure 3: Lesion at the time of patient discharge.

4. Discussion

Congenital cutis aplasia (CCA) is a rare and heterogeneous group of congenital disorders characterized by the absence of skin in focal or generalized areas. Although its true incidence is unknown, it is estimated to occur between 0.5 and 3 cases per 10,000 live births, with some studies suggesting a slight predominance in females. Approximately 85 to 96 percent of CCA cases manifest as isolated lesions on the scalp [4,5,6]. Most cases are sporadic, although familial cases have been documented, such as the one described here, which presents up to the third generation through the maternal line. Inheritance may be autosomal dominant with reduced penetrance (where some gene carriers show no symptoms) or autosomal recessive [7].

Theories regarding CCA pathogenesis include incomplete neural tube closure, embryonic fusion, and intrauterine trauma. In addition, several medications have been associated with the condition, including methimazole, valproic acid, angiotensin-converting enzyme inhibitors, benzodiazepines, misoprostol, and cocaine [8,9]. The phenotype in this case, a cranial defect close to the midline and irregular in shape, is associated with the proposed etiology of incomplete neural tube closure and genetic factors, although the exact cause has not been established.

The clinical presentation of CCA at birth can vary considerably depending on the timing of appearance, the layers of tissue involved, and the degree of intrauterine healing. Generally, CCA presents as a small (≤1 cm), solitary, hairless skin defect located on the upper scalp, covered with atrophic tissue or scab. Larger (>1 cm) and irregularly shaped defects are often associated with underlying bone, dura mater, and intracranial vasculature abnormalities. These characteristics are associated with the non-membranous (star-shaped) form of congenital cutis aplasia of the scalp, which is the type considered in this case, and it may be familial, inherited in an autosomal dominant manner. However, the size and depth of the defects can vary significantly, even within the same family, and small defects on the scalp or areas of alopecia may go unnoticed by some family members. Newborns with large bony defects face a high risk of infections, massive hemorrhages, and thrombosis of the sagittal sinus [10,11].

Diagnosis is usually made clinically. Newborns with large scalp defects or membranous lesions may require neuroimaging studies, including skull radiographs, color Doppler ultrasound, transfontanelar ultrasound, CT scans, and MRI, to assess underlying bone, intracranial vasculature, and brain anomalies [12].

Regarding treatment, there is no consensus or evidence-based guidelines. Treatment decisions for a newborn with CCA should be made on a case-by-case basis, considering the size, depth, and location of the defect, and in consultation with a multidisciplinary team that may include a pediatrician, pediatric dermatologist, pediatric plastic surgeon, and pediatric neurosurgeon, as needed. Treatment options range from no intervention, conservative treatments, surgical treatment, or a combination of surgical and non-surgical interventions [13,14].

In this case, conservative treatment was initiated, which involves the use of adherent or non-adherent dressings with antibacterial agents. Large defects on the scalp and midline cranial defects that expose the sagittal sinus are associated with a high risk of thrombosis or massive hemorrhage, which was ruled out in this patient after two normal cranial CT scans. There is general agreement among experts that these lesions should undergo surgical intervention with skin grafts or local flaps to cover the exposed soft tissue. The choice of surgical technique and timing of surgery depends on the type of defect, the presence of associated anomalies, and the surgeon's experience. For now, the case described shows

notable improvement with conservative treatment, and the need for surgical management will be determined based on outpatient follow-up by plastic surgery [13,14,15].

5. Conclusion

Congenital cutis aplasia is an uncommon and scarcely studied anomaly in newborns that requires a meticulous clinical approach. In this case report, it has been shown that, through accurate diagnosis and multidisciplinary management, a favorable outcome can be achieved in affected patients. The complementary studies conducted showed no significant abnormalities, which enabled timely and appropriate treatment. The patient in question was discharged without neurological or infectious complications and with significant improvement in the lesion, highlighting the importance of an integrated approach.

Ultimately, it is important to emphasize that a patient with congenital cutis aplasia can lead a normal life, provided they do not have associated malformations that compromise their health and quality of life.

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