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CASE REPORT

Aplasia cutis congenita on the scalp: case report and review of literature

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Ectodermal dysplasia, Diseases, Scalp, Infant, Newborn, Diseases.

Abstract

Introduction: Congenital Cutaneous Aplasia is a rare skin lesion, usually restricted to the scalp, characterized by a focal absence of cutaneous tissue, sometimes involving muscles, periosteum and dura mater. There are several etiologies described, such as poor neural tube closure, vascular anomalies, pressure necrosis in the localized embryo, among others. **Method:** We report the case of a newborn at the first physical examination with an annular vesicular lesion, in the occipital region, of 0.8 cm in greater diameter, with hyaline / bloody content and peripheral hyperemia. After a clinical diagnosis was made, we opted for conservative treatment with topical application of hydrogel until complete healing. The patient was then discharged from the hospital, with subsequent return and resolution of the lesion at 30 days of life. **Conclusion:** Congenital cutaneous aplasia is a rare disease, which can affect only the scalp, but can also present with the absence of part of the skullcap, which increases mortality due to the increased risk of complications such as liquor fistula, meningoencephalitis and bleeding.

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INTRODUCTION

Aplasia cutis congenita (ACC) is a rare skin defect that affects the scalp in 85% of the cases and the calvarium in 15% to 30% of the patients[1]. It occurs as a single lesion in around 75% of the patients, but may occur in other parts of the body. It is characterized by a focal absence of skin tissue, with the possibility of involving underlying structures, such as muscles, periosteum and dura mater[2]. Although patients with larger, deeper lesions are at risk of meningitis, bleeding and thrombosis, prognosis is favorable in most cases[3,4].

Reported incidence is approximately 0.5 to 1 case for every 10,000 births[5]. Many cases of ACC occur sporadically, although familial cases have been reported. It is an autosomal recessive or autosomal dominant condition with reduced penetrance (some gene carriers will not develop clinical signs or symptoms)[5].

Lesions appear more frequently close to the fontanelles on the midline, especially along the superior sagittal sinus, although they also occur in the parietal region, retroauricular region, neck, trunk, arms and legs. Approximately 80% of small lesions occur in the vicinity of the parietal whorl. They are present since birth and occur equally in individuals of different ethnicities. In most cases, they affect the scalp and are limited to the dermis, epidermis and sometimes the subcutaneous tissue, measuring 0.5 to 3 cm in diameter[6,7].

Even though ACC is mostly treated by pediatric dermatologists, obstetricians and pediatricians are often the first to see the lesions. Since ACC may be associated with other underlying physical defects or syndromes, it is important to distinguish between the various clinical subtypes of the condition to improve diagnosis, prognosis and further evaluation.

The purpose of this article is to report a case, review current literature on the subject, and discuss diagnosis, etiology, and conditions associated with ACC.

CASE REPORT

E.E.H, a female infant, was born at 38 weeks of gestational age (calculated based on the method described by Capurro) from a cesarean section due to placenta previa. Her mother was 21 years old and was on her third pregnancy; she had had a cesarean section before and an abortion; she was obese and did not have other comorbidities or habits; she went to only one prenatal care visit and an ultrasound (US) scan was performed in the third trimester of pregnancy without showing any changes. She underwent serological screening only in the third trimester and upon admission, all test results were non-reactive. No comorbidities were described during prenatal care.

The newborn (NB) presented no complications during birth. She weighed 3,605g, had a 1-minute Apgar score of 8 and a 5-minute Apgar score of 9, which were appropriate for her gestational age, with a large amount of vernix caseosa on her head and chest.

After a period of observation in the obstetric care center, the mother and the newborn were transferred to rooming-in, where the first complete physical examination of the newborn is carried out and an annular vesicular lesion was observed in the occipital region, with bloody hyaline content and an area of alopecia and hyperemia around it, measuring 0.8 cm in the largest diameter (Figure 1). No other lesions were found and the mother had no other concerns.

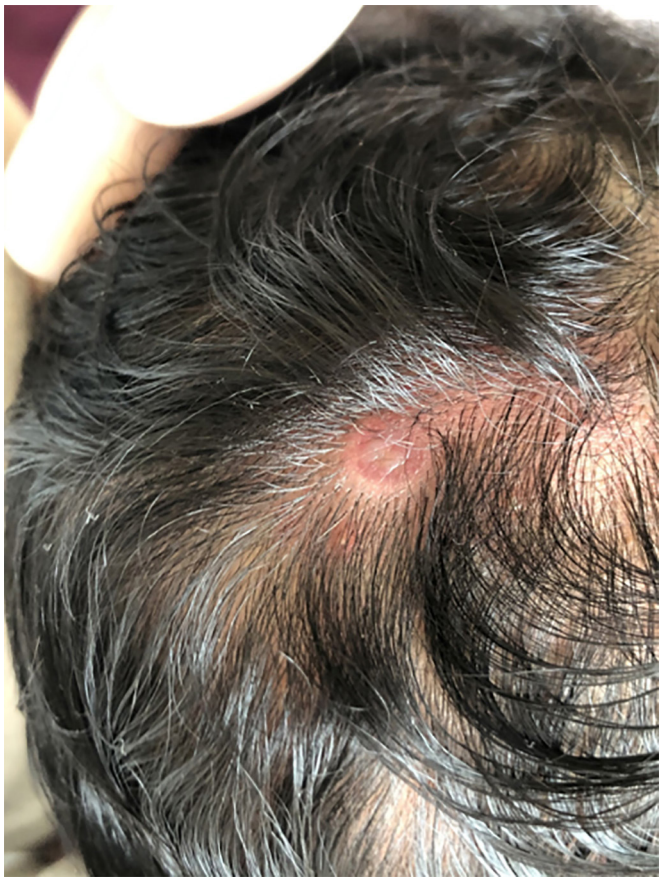

Figure 1. Aplasia cutis congenita. Fonte: The author. Vesicular annular lesion, with bloody hyaline content and an area of alopecia and hyperemia around it, measuring 0.8 cm in the largest diameter.

DISCUSSION

ACC is a rare disease with a mortality rate of around 20%, resulting from an association with meningitis or bleeding in patients with skullcap involvement[4,7].

Although several theories have been proposed for the pathogenesis of ACC, most authors believe that the condition is not tied to one single underlying cause. Rather, it results from a variety of events that occur in utero. There are probably two main pathways for the development of ACC: (a) disruptions or failure in the development of skin layers, including the epidermis, the dermis, and subcutaneous fat, and (b) in utero destruction of skin that might otherwise have developed normally[7].

Frieden[8] categorized the various forms of ACC into nine clinical groups (Table 1) based on associated anomalies, inheritance pattern, and affected body area to improve diagnostic accuracy. Our patient seems to fit best into Frieden's group 1 (Scalp ACC without multiple anomalies) based on clinical findings of skin lesion without associated anomalies.

ACC is diagnosed based on clinical findings, with manifestations identified in several ways. Skin ulcers or erosion are seen on physical examination and may involve deeper structures, reaching muscle and bone tissue. ACC may also resemble an atrophic scar from birth[9,10].

Most patients do not present underlying syndromes, malformations or other abnormalities. However, individuals diagnosed with ACC should undergo further investigation and thorough clinical examination. Cases of ACC among family members should be investigated due to possible inherited and associated conditions[3,4].

There are two main clinical variants of scalp ACC: membranous and non-membranous. Membranous ACC tends to manifest as small, oval or round atrophic plaques with a membrane-like surface. Lesions may be accompanied with the collar sign, usually with darker and longer hair around them[11].

The membranous layer may be filled with fluid or blood, giving it a bullous appearance. Round or oval lesions are more likely to be associated with developmental changes rather than normal tissue destruction[12]. Membranous ACC does not necessarily occur on the vertex of the scalp; It may also develop on the lateral aspects of the scalp and face, suggesting failure to close the embryonic fusion lines[13].

Table 1. Classification proposed by Frieden[17] for aplasia cutis congenita.

Group	Involved body site	Associated alterations	Inheritance
Group 1 Scalp ACC without multiple anomalies	Scalp, more common on the vertex	Cleft lip and palate, tracheoesophageal fistula, patent ductus arteriosus, omphalocele, polycystic kidneys, intellectual disability, cutis marmorata telangiectatica congenita	Autosomal dominant or sporadic
Group 2 Scalp ACC with associated limb abnormalities	Scalp midline	Limb reduction abnormalities, syndactyly, club foot, nail absence or dystrophy, skin tags on toes, persistent cutis marmorata, encephalocele, hemangioma, heart disease, cryptorchidism, postaxial polydactyly	Autosomal dominant
Group 3 Scalp ACC with associated epidermal and organoid nevi	Scalp may be asymmetric	Corneal opacity, scleral dermoid cysts, eyelid colobomas, psychomotor retardation, seizures	Sporadic
Group 4 ACC overlying embryologic malformations	Abdomen, lumbar skin, scalp, any location	Meningomyelocele, spinal dysraphism, cranial stenosis, congenital midline porencephaly, leptomeningeal angiomatosis, ectopic pinna, omphalocele, gastroschisis	Depends on the underlying condition
Group 5 ACC with associated fetal papyraceous or placental infarcts	Multiple symmetric areas, often linear or star-shaped, on the scalp, chest, flanks, axillae, and extremities	Single umbilical artery, developmental delay, spastic paralysis, nail dystrophy, amniotic bands	Sporadic
Group 6 ACC associated with usually localized epidermolysis bullosa, without multiple congenital anomalies	Extremities	Formation of blisters on the skin and/or mucosae, deformed or absent nails, metatarsus varus, renal agenesis (seen in cases of recessive dystrophic EB, dominant dystrophic EB and simple EB)	Depends on EB type: it may be autosomal dominant or recessive
Group 7 ACC localized to extremities without blistering	Pretibial sites, dorsum of the hands and feet, wrist area	None	Autosomal dominant or recessive
Group 8 ACC caused by specific teratogens	Scalp (with methimazole); any site (herpes simplex or herpes zoster infection)	Imperforate anus (methimazole); signs of intrauterine infection by herpes simplex or herpes zoster	Not inherited
Grupo 9 ACC associated with malformation syndromes	Scalp, any location	Trisomy 13; 4-p syndrome, ectodermal dysplasia, Johanson-Blizzard syndrome, focal dermal hypoplasia, XY gonadal dysgenesis	Various, depending on the specific syndrome

ACC = aplasia cutis congenita; EB = epidermolysis bullosa. Source: Frieden[17].

Most scalp lesions are membranous (60%) and may develop from mechanical forces of tension and stretching on the skin and underlying tissue, derived from the rapid growth of the brain and scalp[13]. Growth is greatest at the vertex of the scalp, where many of these lesions are observed[13]. They may also be a form of dysraphism, with impaired closure of the ectoderm over the neural tube[14]. Almost a fifth of the patients may have underlying bone changes, such as cranium bifidum[15].

Treating patients with ACC is not easy, due to the rarity of the condition and the variety of clinical presentations. Lesion repair may range from simple excision to complex reconstruction using bone grafts and muscle flaps.

The size and location of the lesions, along with associated congenital malformations, define prognosis and treatment. Superficial limited lesions often resolve without complications[2]. The recommended treatment for small superficial injuries is conservative, including local care of the wound with dressings and good hygiene. Extensive lesions (more than 6 cm in diameter) rarely heal fully and require long term treatment[1], potentially causing bleeding and infection[15].

Many treatments have been described for ACC, including petroleum jelly or bacitracin, occlusive dressings, moist dressings or saline solution, betadine, silver sulfadiazine, Acticoat and biological or synthetic skin substitutes[9,13-15]. Average healing time is 27.9 days for patients treated conservatively[9]. Hypertrophic scars, atrophic plaques or cicatricial alopecia may remain after wound healing[15].

Our patient was administered conservative treatment due to the size of the lesion (0.8 cm) and other benign characteristics. Daily local applications of Hydrogel and dressings with Calcium Alginate were prescribed for better healing. The patient was discharged with an outpatient follow-up plan, with the lesion resolving within 30 days from birth.

CONCLUSION

Aplasia cutis congenita is a rare disease that may affect only the scalp. Skullcap involvement increases mortality due to the greater risk of complications such as cerebrospinal fluid leak, meningoencephalitis and bleeding. The clinical presentation of ACC may provide clues to the underlying pathophysiology of the condition, which may direct further evaluation and additional treatment.

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