

Case Report

Aplasia Cutis Congenita of the Scalp with a Familial Pattern

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Aplasia Cutis Congenita (ACC) is a condition characterized by congenital absence of skin, usually on the scalp. ACC can occur as an isolated condition or in the presence of other congenital anomalies. Here we describe a case of a 16-day-old baby girl with an isolated ACC of the scalp. Her elder two siblings have been diagnosed with ACC with concomitant cardiac or limb anomalies. The patient was managed conservatively until the defect has formed scar tissue 6 months later.

1. Introduction

Cutis aplasia or Aplasia Cutis Congenita (ACC) is an uncommon and rare congenital abnormality involving variant layers of the skin, mostly as a solitary lesion involving the midline over the skull vertex and, less commonly, underlying periosteum and bone [1]. It may occur in other sites as well such as the chest, abdomen, or limbs [2, 3]. Of lesions on the scalp, 20% involve the cranium, exposing the underlying dura membrane. ACC could also be found in other congenital anomalies; since it was first described in 1767 by Cordon, around 500 similar cases have been reported so far [4]. Frieden classified the different anomalies into 9 groups based on the number and the presence or absence of other anomalies (Table 1) [5]. The lesions in those cases are quite variable, ranging from only local absence of skin to a complete absence of epidermis, subcutaneous tissue, bone, or in some cases the dura [6–8]. The incidence of ACC is estimated as 1 per 10,000 live births [1]. This failure of formation is frequently more observed in females. The etiology remains unclear so far; however, both genetic and environmental causes have been implicated, including vascular blood supply, a sudden arrest of midline embryological development, and failure in neural tube closure, and syphilis has at one time contributed as the cause [1, 9]. Rupture of amniotic membrane in an early time, forming amniotic bands, may also be from the cause [5]. A number of teratogenic drugs such as Methimazole,

a Thiomidazole derivative used as an antithyroid agent, have shown to be involved [10–13]. There are similar cases, classified as being of an autosomal-dominant inheritance [14]. Establishing a diagnosis is usually based on the findings of the clinical examination, typically presenting as a hairless, smooth skin defect covered up by atrophic tissue or a dark-colored eschar. Superficial defects presenting as an ulcer are usually treated conservatively. Extensive or deep defects may require reconstruction of the scalp area or the use of bone transplants. However, hairless or scarcely haired scars mandate excision of the lesion and covering it with local flap from the scalp [9, 15–20].

2. Case Report

A 16-day-old newborn Female from Saudi Arabia presented to the clinic with a skin defect localized to the scalp since birth. The baby did not suffer from any ailments, and her medical history was unremarkable. Her mother, 32 years old, denied any history of illnesses during her pregnancy, infection, or drug intake taking including Nonsteroidal Anti-Inflammatory Drugs (NSAID) or Methimazole. She completed 38 weeks of gestation and delivered her baby via a normal vaginal delivery. The newborn did not sustain any birth injury and did not suffer from any other abnormalities or feeding difficulties. She did not require any intensive care and went home from hospital with her mother. Upon

TABLE 1: Classification for ACC.

| | Group | Associated anomalies | Inheritance |
|---|---|---|--|
| 1 | Scalp ACC without multiple anomalies | Cleft lip and palate, tracheoesophageal fistula, patent ductus arteriosus, omphalocele, mental retardation, polycystic kidneys | Autosomal dominant or sporadic |
| 2 | Scalp ACC with limb abnormalities | Limbs reduced, syndactyly, clubfoot, encephalocele, nail dystrophy or absence, persistent cutis marmorata | Autosomal dominant |
| 3 | Scalp ACC with skin/organoid nevi | Epidermal nevi, organoid nevi, corneal opacities, scleral dermoids, eyelid colobomas, mental retardation, seizures | Sporadic |
| 4 | ACC overlying embryologic malformations | Meningomyelocele, spinal dysraphia, cranial stenosis, leptomenigeal angiomas, gastroschisis, congenital midline porencephaly, ectopia of ear, omphalocele | Depends upon underlying condition |
| 5 | ACC with fetus papyraceus or placental infarcts | Single umbilical artery spastic developmental delay, spastic paralysis, clubbed hands and feet, amniotic bands | Sporadic |
| 6 | ACC associated with epidermolysis bullosa | Blistering of skin and/or mucous membranes, deformed nails, pyloric or duodenal atresia, abnormal ears and nose, ureteral stenosis, renal anomalies, amniotic bands | Depends upon type of epidermolysis bullosa |
| 7 | ACC localized to extremities without blistering | None | Autosomal dominant or recessive |
| 8 | ACC caused by teratogens | Imperforate anus (methimazole), other signs of intrauterine infection with varicella or herpes simplex | Not inherited |
| 9 | ACC associated with congenital syndromes | Trisomy 13, 4p-syndrome, ectodermal dysplasia, focal dermal hypoplasia, amniotic band disruption complex, XY gonadal dysgenesis, Johanson-Blizzard syndrome | Depends upon syndrome |



FIGURE 1: The newborn presented with skin defect of the scalp with an overlying crust.



FIGURE 2: Six-month follow-up shows scar formation over the affected area.

local examination, the defect was solitary, localized with an irregular shape and approximately 6×6 cm in size (Figure 1). The lesion involved the epidermis and the upper dermis only. Neurosurgical team was involved in the care of this patient. A CT scan of the head was performed, and no deep tissue involvement was noted. Reconstruction solutions were offered to the parents but they insisted on nonsurgical intervention. Therefore, the patient was treated with noninvasive debridement of the lesion and local therapy, including gentle water cleansing and the application of topical antibiotic ointment. Six months later, the patient has returned for a follow-up. Scar tissue has formed over the defect (Figure 2). Family history revealed that none of her parents had the

same condition; however, two of her sisters did and were diagnosed with cutis aplasia. The elder one is currently 4 years of age, with right unilateral terminal reduction of the first and second toes (Figure 3). The other sister was born prematurely and died shortly after birth due to cardiac anomalies.

3. Discussion

ACC occurs as a solitary defect; it can happen alone or in the presence of syndromic congenital anomalies. The involvement of the scalp area may lead to the understanding of the etiology. Upon our review to the literature available, cases were often characterized by an entire absence of skin



FIGURE 3: Unilateral terminal reduction of the right first and second toe.

and subcutaneous tissues. Histologically, we found that most of the lacking tissues belonged to epithelial ectoderm. The condition could be associated with chromosomal defects [21]. Some researches show the association with gestational conditions such as an intrauterine vascular ischemia, amniotic adhesions, and viral infections [22, 23]. A rise of alpha-fetoprotein levels and a distinct amniotic fluid acetylcholinesterase band were found in recent article as markers for ACC [24]. Also, a number of drugs have been linked to ACC. For example, the use of cocaine during pregnancy can lead to vasoconstriction of the placenta or disruption of the fetus vascularity, causing the cranial defects and anomalies of the central nervous system (CNS) [25]. Methimazole, a drug used for the treatment of hyperthyroidism, may show some skin affection. Benzodiazepines use is also linked with ACC as described by Martínez-Lage et al. [7]. Surgical treatment requires careful preoperative planning [26]. Minimal superficial lesions are treated conservatively to heal gradually by reepithelialization and result with a hypertrophic or atrophic scar. Tissue expander insertion may be necessary in extensive lesions reaching the scalp, whereas the one deep enough to reach the brain, bone, and meningeal transplants may be indicated [9, 15, 20, 27]. Deep defects overlying the sagittal sinus are indicators for urgent surgical intervention to prevent potentially lethal infections or hemorrhage [28–30]. Grafting [31] and temporary use of biological dressings [32] and silver sulfadiazine dressings while waiting for the processes of skin and bony ingrowth [3] have been published with variable degree of success.

4. Conclusion

Aplasia Cutis Congenita is a rare congenital disorder characterized by absence of skin, most frequently overlying the scalp. ACC can be associated with many anomalies. We report a case of a newborn with isolated scalp ACC, which was treated conservatively. Although neither of the parents had ACC, two of the patient's siblings have ACC of the scalp with concomitant cardiac or limb anomalies.

Competing Interests

The authors declare no competing interests.

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