Giant Aplasia Cutis Congenita of the Scalp: Between the Risk of Infection and the Risk of Bleeding, Case Report

Khaled Sadeq Al-Shaibari, MD, Ped-SB, ABP, JBP*

ABSTRACT

The scalp (70%) is the most prevalent site of a lesion. ACC has an estimated incidence of about 3 in 10,000 newborns, with roughly 500 recorded instances in the literature.

A full-term male product for primigravida mother, born in a small general hospital by cesarean section due to the concern of oligohydramnios. He cried immediately after birth with APGAR score 8& 9 at 1 and 5 minutes respectively. After delivery, they noted to have extensive tissue defect of the scalp. Family history showed non-consanguineous healthy parents, no similar history in their families.

INTRODUCTION

Aplasia cutis congenita (ACC) is a varied group of illnesses first described by Cordon in 1767 that are characterized by the loss of epidermis, dermis, and, in rare cases, subcutaneous tissues or even bone tissue in several body regions. The scalp (70%) is the most prevalent site of a lesion. ACC has an estimated incidence of about 3 in 10,000 newborns, with roughly 500 recorded instances in the literature[1]. The biggest risk factor described in the literature is the antithyroid medication methimazole, which is classified as type 8 ACC by Frieden. However, because of the extremely low incidence, cases of ACC are extremely limited, making it impossible to derive completely accurate epidemiological data. The actual cause of ACC is unknown; however, it is most likely caused by poor prenatal skin development. The disease manifests as single or multiple lesions on any area of the body, while 70% to 90% of lesions are localized to the scalp's vertex. ACC is divided into six categories, some of which are linked to congenital dermatologic disorders[2]. Although most lesions heal on their own, certain locations and clinical characteristics should prompt a more thorough workup to rule out underlying soft tissue anomalies that could be fatal. ACC can be caused by a variety of factors, including familial cases, epidermolysis bullosa (EB), and imperfect neural tube closure[3]. At birth, ulcerations can be shallow or deep, with complete absence of all layers of skin, extending to the dura or bone in some cases. In other circumstances, due to in utero healing, a healed scar may be the only discovery. Because of the varied nature of ACC, knowing the pathophysiology is critical in the diagnosis and therapy of these patients. Most ACC heals on its own, but deadly consequences include sagittal sinus bleeding, meningitis, and brain herniation. The therapy of ACC is contentious. Surgical treatment involves debridement and suturing, local flap transfer, free flap transfer, and skin grafting. Anesthesia-related problems, intraoperative hemorrhage, postoperative infection, and flap necrosis are all hazards[4]. Conservative treatment consists primarily of regular dressing changes to promote spontaneous epithelialization, and its dangers include bleeding, longer healing period, and wound infection.

CASE REPORT

A full-term male product for primigravida mother, born in a small general hospital by cesarean section due to the concern of oligohydramnios. He cried immediately after birth with APGAR score 8& 9 at 1 and 5 minutes respectively. After delivery, they noted to have extensive tissue defect of the scalp. Family history showed non-consanguineous healthy parents, no similar history in their families.

PHYSICAL CONDITION

His physical examination revealed an active, conscious, alert, well boy, not in respiratory distress with stable vital signs and normal growth parameters. There was a giant aplasia cutis (measuring 10*9 cm), in most of the vertex area, no other dysmorphic features or skin defects and no hemangiomas or skin stigmata and no limbs defects or anomalies. The chest was clear; the cardiovascular system revealed normal heart sounds with no murmur and normal peripheral pulses. The abdomen was soft, with no organomegaly and normal male genitalia. There was no neurological deficit with normal neonatal reflexes (Figure 1).

Figure 1: Physical Condition

MANAGEMENT

He was admitted to Neonatal Intensive Care Unit (NICU), stayed for three days where skull X-ray showed a large area of bone defect. He was transferred to another hospital upon father request for financial issues. Neurosurgeon decided to do daily dressing of the defect and referred him to a plastic surgeon. In the second hospital, they reported findings of the black area around and in the area of scalp defect with yellow discharges. They managed him as a case of sepsis, and the culture of wound showed growth of Klebsiella organisms. Plastic surgeon deferred operation due to this infection and advised to transfer him to a higher center. The wounds were managed conservatively with

* King Abdulaziz Hospital
Jeddah
Saudi Arabia.
E-mail: kald22ye@gmail.com

antibiotic systemic and ointments, covered by occlusive dressings that were regularly changed. Patient has been discharged after treating the infection. Two days later, patient was admitted to King Abdulaziz Hospital- Jeddah, as parents noted reappearing of blackish and yellowish discoloration of the defect area. The patient was admitted to ICU; septic workup sent and was covered with intravenous antibiotics. He was quiet in the room air, not in respiratory distress, active and feeding well. Wound on vertex area showed improvement in color and became drier (Figure 2).

Figure 2: Radiology Imaging

OUTCOME

While the patient was on management, one day at early morning, cardiac monitored showed an increased in heart rates, and his blood pressure started to show dropping of hypotension range, much bleeding was noted from scalp defect as covered gauze was soaked with blood. Resuscitation started, but unfortunately, the patienthas been lost due to mostly hypovolemic shock. There was no bleeding from other sites and patient temperatures last days were normal. Unfortunately, chromosomal analysis and neuro imaging not done for this patient.

DISCUSSION

This case of the giant aplasia cutis congenita of the scalp represents a rare large size of aplasia cutis among published cases of the literature[1]. As they ranged the size of the scalp defects from 0.5 cm[12] to as large as 100 cm[2]. Depending on Frieden et al. classification[1] this case belonged to group one: Scalp aplasia cutis congenita without multiple anomalies. ACC is often diagnosed purely through clinical examination. Given the patient's age demographics and typical scalp involvement, many doctors refuse to undertake a lesional biopsy. When a biopsy is required to aid in the diagnosis, a thorough workup is required, including evaluating the lesion using ultrasound (US) or MRI to ensure there are no underlying abnormalities that could be injured during the biopsy procedure. Non-healed lesions have a missing epidermis and/or dermis, as well as a proliferation of blood vessels, according to histopathologic results. A thin translucent membrane covers a form of ACC known as membranous type ACC. On pathology, lesions that have already healed with a scar will have a thin or flattened epidermis, no adnexal structures, and extensive, dermal fibrosis.

Bleeding and infection are the most complications from large defects as reported to lead to death in 20 to 30% of patients[2]. Lethal hemorrhage, as happened in our case, was reported by Kim et al. as sagittal sinus hemorrhage[3].

Although the lesions are noninflammatory and well defined, there is much debate about how to treat ACC, and there has been a lot of scientific interest because of the extremely high mortality rates, which range from 20 to 55%. Sagittal sinus hemorrhage, subsequent local infection, meningitis, sagittal sinus thrombosis, or other significant birth abnormalities associated with ACC cause high mortality/morbidity rates.

Conservative therapy entails regular wound cleansing and dressing application, as well as the administration of systemic antibiotics. This comprises physiological saline, continuous saline drips, betadine solution, bacitracin ointment, and silver sulfadiazine dressings, which are used to keep moisture in the skin, avoid desiccation, and allow for spontaneous epithelialization.

There have been numerous accounts of specialized adherent wound dressings that are supposed to promote faster wound healing, as well as recent reports of innovative dressing materials such as fatty gauzes.

Surgical management, on the other hand, encompasses a variety of procedures. Primary wound closure, skin grafting (autologous or allografts), local scalp flaps with or without tissue expansion, free flaps, 26 muscle flaps, full-thickness or split-thickness skin grafts, and cranial vault reconstruction with bone grafts are all standard surgical procedures. Specialized surgical procedures, such as bipedicle opposing local flaps, rotational flaps, or L-shaped flaps, have been utilized successfully.

Unless there are underlying problems, aplasia cutis congenita can be easily managed. Complications can emerge, necessitating the assistance of an interprofessional team of healthcare specialists to diagnose and manage these instances. With complications, the morbidity and mortality of ACC are extremely high, emphasizing the importance of effective communication among all physicians involved.

CONCLUSION

It is critical to understand each of the causes and manifestations of ACC in order to perform appropriate diagnosis and management. It should be mentioned that, due to the lack of treatment algorithms and standards for ACC care, as well as the requirement for specific treatment decisions, such cases should be addressed in pediatric neurosurgical centers with sufficient experience and skill.

Authorship Contribution: All authors share equal effort contribution towards (1) substantial contributions to conception and design, acquisition, analysis and interpretation of data; (2) drafting the article and revising it critically for important intellectual content; and (3) final approval of the manuscript version to be published. Yes.

Potential Conflicts of Interest: None

Competing Interest: None

Acceptance Date: 06-05-2023

References

1. Frieden IJ. Aplasia cutis congenita: A clinical review and proposal for classification. J American Academy Dermatology. 1986.
2. Humphrey SR, Hu X, Adamson K, et al. A practical approach to the evaluation and treatment of an infant with aplasia cutis congenita. J Perinatol 2018;38(2):110-7.
3. Note C. Scalp Aplasia Cutis Congenita Presenting With Sagittal Sinus Hemorrhage. Neonatal Surg 2012;1(1): 8.
4. Başterzi Y1, Bağdatoğlu C, Sari A, et al. Aplasia cutis congenita of the scalp and calvarium. J Craniofac Surg 2007;18:427-9.
5. Beekmans SJA, Haumann TJ, Vandertop WP, et al. Aplasia cutis congenita in 4 infants. Ned Tijdschr Geneeskd 2002;146:1842-5

6. Blionas A, Giakoumettis D, Antoniades E, et al. Aplasia cutis congenita: Two case reports and discussion of literature. Surg Neurol Int 2017;8:273.
7. Puvabanditsin S, February M, Garrow E, et al. Our experience with a severe case of aplasia cutis congenita with a large skull defect. Int J Dermatol 2016;55(10): 1151-3.
8. Perry BM, Maughan CB, Crosby MS, et al. Aplasia cutis congenita type V: a case report and review of the literature. Int J Dermatol 2017;56(6): e118-21.
9. Humphrey SR, Hu X, Adamson K, et al. A practical approach to the evaluation and treatment of an infant with aplasia cutis congenita. J Perinatol 2018;38(2):110-7.
10. Sachs C, Tebacher-Alt M, Mark M, et al. Aplasia cutis congenita and antithyroid drugs during pregnancy: Case series and literature review. Ann Dermatol Venereol 2016;143(7):423-35.
11. Marneros AG. BMS1 is mutated in aplasia cutis congenita. PLoS Genet 2013;9(6):e1003573
12. E Rogvi R, Sommerlund M, Vestergaard ET. Aplasia cutis congenita is a rare and possibly overlooked congenital anomaly. Ugeskr Laeger 2014;176(48).
13. O'Neill JK, Carter M, Warr RP. Aplasia cutis congenita. A case of scalp defect repair using two opposing bipedicled local flaps. J Plast Reconstr Aesthet Surg 2010;63(3):e242-4.
14. Martinez-Regueira S, Vazquez-Lopez ME, Somoza-Rubio C, et al. Aplasia cutis congenita in a defined population from northwest Spain. Pediatr Dermatol 2006;23(6):528-32.