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A Case Report of Aplasia Cutis Congenita in Northeastern Iran

Zahra Yazdani[1], Hamide Kamali[2], Haniye Ghasri[2], Fatemeh Rabani[3], Azam Saedikia[1, 4]*, Zobide Rafat[1,5]

[1] Instructor, Shirvan School of Nursing, North Khorasan University of Medical Sciences, Bojnord, Iran.
[2] Nursing student, Student Research Committee, Shirvan School of Nursing, North Khorasan University of Medical Sciences, Bojnord, Iran.
[3] Bachelor of Nursing, Nursing Department, Bent Al-Hoda Hospital, North Khorasan University of Medical Sciences, Bojnord, Iran.
*[4] PhD student, Student Research Committee, Nursing Department, Faculty of Nursing and Midwifery, Zahedan University of Medical Sciences, Zahedan, Iran. **(Corresponding author)***

5 PhD student, Student Research Committee, Nursing Department, Faculty of Nursing and Midwifery, Mashhad University of Medical Sciences, Mashhad, Iran.

Abstract

Aplasia Cutis Congenita (ACC) is a rare and astonishing congenital disorder characterized by localized or widespread absence of skin, particularly in the head region. ACC has an estimated incidence of 0.5 to 1 in every 10,000 newborns. This disorder can damage various depths of the skin and, in some cases, is associated with bone abnormalities and cranial defects. While the exact pathophysiology of this disease is not yet fully understood, genetic factors, intrauterine infections, and vascular insufficiency have been proposed as its potential causes.

In a specific case, a baby girl was born with a lesion measuring 3 × 3 centimeters in the anterior fontanel area, and brain ultrasound confirmed the diagnosis of Aplasia Cutis, so the infant was transferred to the intensive care unit. Treatment included regular dressing changes and antibiotic administration, and fortunately, the infant health improved after a few days and was discharged.

The prognosis for small lesions is usually positive, but this disorder can lead to complications such as infection and bleeding. Given the possible connection between placental type and the occurrence of Aplasia Cutis, further research in this area is essential to gain a deeper understanding of the factors influencing this disorder and to improve treatment methods.

Key word: Aplasia Cutis Congenita, pathophysiology, skin.

Corresponding author: Azam Saedikia, ORCID ID: 0000-0002-9631-6549. Email: a.saedikia@nkums.ac.ir
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Introduction

Various congenital disorders that are associated with localized skin defects have an unknown genetic basis. Although these disorders are not common, they can provide valuable information and insights into the main biological mechanisms of the skin [1]. Aplasia Cutis Congenita (ACC) is a rare congenital disorder characterized by localized or widespread absence of skin [2]. The most common site of involvement is the scalp, but it may also be seen on the face, torso and limbs. In these patients, large defects in the scalp accompanied by skull involvement are not common but can be observed [3-4].

This disease was first described by Cordon in 1767. However, by 2013, only 500 cases of this disease have been reported. This defect can be observed at various depths of the skin, including the epidermis, dermis, and even subcutaneous tissues or bony tissues [5]. Other names have also been attributed to this disorder by the National Organization for Rare Disorders, which include congenital scalp and skull defect, congenital neonatal scalp ulcer, and congenital scalp defect [2,6].

ACC is classified into 9 different types based on the location and pattern of skin loss, the presence of associated anomalies, and the method of inheritance [2]. The pathophysiology of ACC is unknown, but it appears to be multifactorial in some cases, as both genetic and environmental/exogenous factors can be involved in its occurrence. Various causes recognized as factors leading to intrauterine skin growth disorders include intrauterine infections, vascular skin insufficiency due to placental insufficiency, placental infarction and thrombosis, chromosomal abnormalities and genetic syndromes, amniotic bands, neural tube defects, and the use of certain teratogenic substances such as benzodiazepines, misoprostol, angiotensin-converting enzyme (ACE) inhibitors, methimazole and valproic acid [2,6].

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The management of aplasia cutis congenita (ACC) remains controversial and should be tailored to each patient's specific condition. The decision between conservative and surgical management depends on factors such as lesion depth, involvement of the underlying dura or sagittal sinus, risk of infection or hemorrhage, and the overall clinical stability of the newborn. Although surgical repair has traditionally been preferred for extensive or deep lesions, recent literature increasingly supports conservative approaches when the dura is intact and there is no exposure of critical vascular structures. Conservative management—comprising gentle wound cleansing, moisture-preserving dressings, and infection control—can promote spontaneous epithelialization and satisfactory healing outcomes. Therefore, treatment choice should be individualized, guided by clinical judgment and the specific characteristics of each lesion rather than a fixed size threshold. [6- 8] This defect can also lead to complications such as infection, thrombosis, and bleeding. [6]

Case presentation

The patient was a female infant weighing 3,180 grams, measuring 50 cm in height, with a head circumference of 36 cm. She was delivered vaginally in cephalic position at 39 weeks of gestation in Imam Khomeini Hospital, Shirvan, North Khorasan Province, with a velamentous placenta. Vital signs at birth were stable (pulse 130 bpm, temperature 36.6°C, respiration 50 breaths/min). Apgar scores were 9 and 10 at one and five minutes, respectively.

According to the parents, pre-pregnancy screenings were normal. There was no consanguinity between the parents and this was their first child. They had no history of miscarriage or previous infertility. The mother was 17 years old, had hypothyroidism and was taking 100 mg of levothyroxine daily.

On initial examination of the infant, a 3 x 3 cm circular lesion was observed in the

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anterior forehead area without hair in that area (Figure 1). Brain ultrasound showed no abnormalities. The clinical diagnosis was aplasia cutis. The infant was transferred to the neonatal intensive care unit (NICU), where wound care was performed with saline and Mupirocin ointment three times daily. Broad-spectrum antibiotics, including vancomycin, were started because of the risk of meningitis.

A brain CT scan demonstrated a focal absence of the cranial bone beneath the scalp defect, consistent with aplasia cutis congenita. Given the need for specialized management and the lack of advanced facilities for newborns in the neonatal ward, the infant was referred to Ghaem Hospital in Mashhad for further evaluation. Following stabilization, comprehensive medical assessments were performed, and the newborn was discharged within a few days in stable condition. According to the attending neurosurgeon, surgical intervention was not indicated at that stage, as spontaneous ossification of the skull was anticipated during normal growth. After 32 days of follow-up with the mother, the lesion had markedly decreased in size to approximately 2 × 1 cm, with progressive epithelialization and no signs of infection or neurological complication (Figure 2).

This case emphasizes the importance of prompt recognition and management of Aplasia Cutis to prevent complications such as infection or meningitis in newborns. Informed consent was obtained from the infant's parents for the publication of photographs and information about this anomaly.

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Fig 1: A lesion measuring 3 × 3 centimeters in the anterior fontanel area of the head in birth.

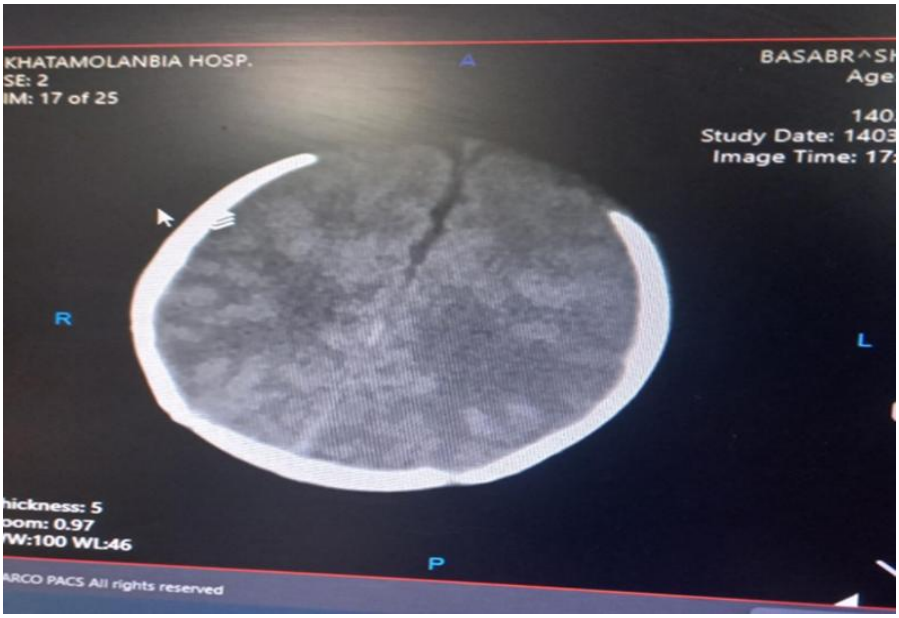

Fig 2: An image of scan CT Brain in the anterior fontanel area of the head.

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Discussion

Aplasia Cutis is a rare congenital disorder with a reported incidence of 0.5 to 1 per 10,000 newborns. It is characterized by localized or widespread skin loss, and presents with diverse manifestations in terms of extent and location [8-9].

The presentation of ACC is diverse, and several classification systems have been proposed for it. ACC can be associated with physical defects or syndromes, which may assist in the diagnosis, prognosis, and further evaluation of the patient.

The exact pathogenesis of ACC is unknown. Although several theories have been proposed in this regard, including neural tube defects, vascular damage due to placental insufficiency, intrauterine infections, genetic mutations, and teratogens.

About 86% of ACC cases involve the scalp, primarily the vertex [8]. ACC at the vertex can be partly explained by the presence of maximum tensile force during the rapid growth of the brain in that area, which occurs during weeks 10 to 15 of pregnancy [10].

Bone abnormalities in ACC occur in approximately 15-20% of cases as the majority of the remaining scalp is involved. Skull defects in ACC lead to exposure of brain tissue and the sagittal sinus, which significantly increases the risk of bleeding, infection, and sagittal sinus thrombosis [9].

The diagnosis of ACC is primarily clinical and currently there is no consensus regarding the therapeutic management of ACC, due to its rarity and variability in location and extent. Management approaches include conservative treatment, which involves dressing the aplastic skin until complete re-epithelialization, and surgical management, which includes excision and closure, skin grafting, local flaps, and tissue expansion [11]. Small lesions without exposed limbs usually heal spontaneously within a few weeks, even when the bone is involved. The prognosis for small self-healing scalp lesions is very

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good [12]. In a retrospective study conducted by Mesrati et al. (2015), 22 infants with aplasia cutis were investigated, including14 were girls and 8 were boys. The lesion shape was oval in 20 cases, triangular in one case, and linear in another case. Conservative treatment included wound dressing with vaseline in 6 cases, and bone reconstruction was carried out in two cases. Also, follow-up and no treatment was recommended for 14 cases. [12]. The shape of the lesion in our report was circular, and the treatment for this type of lesion was the use of topical and IV antibiotics along with sterile dressings

In a study, Magliah and Alghamdi. (2018) reported a case report in which a 45-day-old boy presented to the dermatology outpatient clinic at King Abdulaziz Medical City, Jeddah, Saudi Arabia with a flat scalp lesion in the anterior fontanel space. There was a positive family history of such a condition on the scalp of his older brother, which resolved spontaneously within 3 weeks of delivery without any medical intervention. There was no hemangioma or other congenital malformation in his body. The case was hospitalized.

For 3 days, received intravenous vancomycin, but no improvement was observed in the scalp cyst. Therefore, 15 g of 2% mupirocin ointment was prescribed. received intravenous vancomycin infusion, but no improvement in the scalp cyst was observed. So, 15 g of 2% Mupirocin ointment was administered topically three times a day. After 2 days, the size of the scalp cyst gradually decreased and dried up within 1 week. A 3-month follow-up showed that the scalp lesion had completely healed, leaving a very small atrophic scar, and no further management of the lesion was required. The results of this case show that congenital aplasia of the scalp is a rare disease of unknown etiology, but consanguinity may play a role in this regard. Its management depends on the pattern, location, underlying causes, and associated abnormalities [13].

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In the reported case, the patient was born with a velamentous placenta, which may be associated with certain skin anomalies such as ACC. Reduced blood flow to the fetus due to vasa previa can also affect the normal growth and development of the skin, leading to the occurrence of Aplasia Cutis. Further research is suggested to investigate the relationship between the type of placenta and ACC.

Conclusion

This study aimed to report a case of ACC. Aplasia cutis, as a rare congenital disorder, presents unique medical and scientific challenges. It also provides valuable insights into biological and genetic processes. Given the low prevalence and diversity of this disorder, each case requires thorough examination and personalized management. Recent advancements in the diagnosis and treatment of this condition raise hopes for a brighter future for affected infants. Additionally, the potential link between placental type and the occurrence of aplasia cutis highlights the need for further research in this area. Ultimately, this disorder presents an opportunity to enhance medical awareness and improve treatment methods, which can lead to better quality of life for infants and their families.

Acknowledgments

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Conflict of Interest

The authors declared no conflicts of interest.

Ethical considerations

Informed consent was obtained from the infants' parents to report illness without mentioning their personal identity.

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