

Single Case

Aplasia Cutis Congenita: A Case Report

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Keywords

Aplasia cutis congenita · Skin diseases · Management

Abstract

Background: Aplasia cutis congenita is characterized by congenital focal absence of skin in a newborn. **Case Report:** A 45-day-old boy presented to the outpatient clinic of dermatology at King Abdul Aziz Medical City, Jeddah, Saudi Arabia, with a flat scalp lesion on the space of the anterior fontanel. There was a positive family history of such condition in his older brother's scalp, which resolved spontaneously within 3 weeks after labor, without any medical intervention. There were no associated hemangiomas or other congenital defects in his body. The case was admitted to the hospital. Ultrasound of the head showed bullae over the anterior fontanel, well-defined complex cyst, and an isolated subcutaneous lesion, with no evidence of intracranial extension. The baby was started on intravenous infusion of vancomycin (67 mg in dextrose 5% in water) for 3 days, but no improvement occurred to the scalp cyst. The baby started to receive 15 g of 2% mupirocin ointment, topically three times daily. After 2 days, the scalp cyst gradually decreased in size and became dry within 1 week. Follow-up after 3 months showed that the scalp lesion completely healed, leaving a very small atrophic scar and no further management of the lesion was needed. **Conclusions:** Aplasia cutis congenita is a rare condition of uncertain etiology, but consanguinity may play a role. Its management depends on its pattern, location, underlying causes, and associated anomalies.

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Introduction

Aplasia cutis congenita is a disease that is characterized by a localized or widespread, complete or partial absence or scarcity of skin at birth. The disease may result from the disrupted development or degeneration of skin in utero [1].

The exact mechanism for developing aplasia cutis congenita is still not completely understood. Several factors have been suspected as possible causes, e.g., placental infarcts, genetics, teratogenic substances, intrauterine infections and trauma, ectodermal dysplasia, imperfect neural tube closure, and maternal intrapartum drug use [2].

The disease has been classified into several groups according to its pattern, location, underlying causes, and associated anomalies [3]. Clinically, its lesions generally appear as well-demarcated, translucent, ulcerated membranes, through which it is possible to visualize the underlying structures [4]. Its management depends on the condition. Small localized lesions may be managed conservatively, while bigger wide-spread lesions may necessitate surgery [5].

This article aims to report a rare case of aplasia cutis congenita in an infant aged 45 days.

Case Report

A 45-day-old boy presented to the Outpatient Clinic of Dermatology at King Abdul Aziz Medical City, Jeddah, Saudi Arabia, with a flat scalp lesion on the space of the anterior fontanel (Fig. 1). He was a product of a full-term, spontaneous vaginal delivery, after an uncomplicated pregnancy. His parents were second-degree relatives. He received all his vaccines. The mother was negative for group B streptococcus when investigated during pregnancy.

There was a positive family history of such condition in his older brother's scalp, which resolved spontaneously within 3 weeks after labor, without any medical intervention. There was no previous history of hospitalization to the child.

At delivery, the baby's scalp lesion was 0.5 × 0.5 cm. During the first 25 days of his life, the lesion kept on raising and growing to become an isolated yellowish cyst 2 × 3 cm, after which it stopped growing (Fig. 1). There were no associated hemangiomas or other congenital defects in his body.

On examination, the baby looked well and alert. He was in the 25th percentile for both height-for-age and weight-for-age. Culture from the site of the lesion showed Gram-positive cocci, consistent with *Staphylococcus aureus*. Ultrasound of the head showed bullae over the anterior fontanel, well-defined complex cyst, and an isolated subcutaneous lesion, with no evidence of intracranial extension. During the whole time of hospital admission, the baby was vitally stable.

The baby was started on intravenous infusion of vancomycin (67 mg in dextrose 5% in water) for 3 days. Blood culture became negative, but no improvement occurred to the scalp cyst.

The baby started to receive 15 g of 2% mupirocin ointment, three times daily, a small amount on the lesion. After 2 days of applying mupirocin ointment, the scalp cyst gradually decreased in size and became dry (Fig. 2) within 1 week.

The baby was discharged and his mother was advised to continue applying the mupirocin ointment 3 times daily for 1 month. Follow-up after 3 months showed that the scalp lesion completely healed leaving a very small atrophic scar and no further management of the lesion was needed.

Discussion

Aplasia cutis congenita is a rare condition [6], with an estimated incidence of 0.5/10,000 to 1/10,000 births [7]. Brzezinski et al. [8] stated that the exact mechanism for developing this condition is not completely understood, but recently, several factors have been incriminated, e.g., chromosomal abnormalities, traumatic mechanism, amniotic defects, and intrauterine problems.

In this report, we presented a case with aplasia cutis congenita, localized in the scalp of a male baby aged 45 days. Despite the rare nature of aplasia cutis congenita, his older brother also had the same condition.

Garcia-Romero et al. [5] noted that cases of aplasia cutis congenita may be widespread, associated with dysgenic hydrocephaly, extensive unilateral linear epidermal nevus, hemangiomas, and multiple defects (with less familial incidence) or present as a localized congenital defect with familial incidence (usually on the scalp). The positive family history of aplasia cutis congenita in our patient may be explained by the fact that his parents were second-degree relatives.

The impact of consanguinity on the incidence of congenital malformations was described by Mosayebi and Movahedian [9], who reported that the rate of congenital malformation was 2% among neonates from non-consanguineous marriages compared with 7% from consanguineous marriages. Moreover, a history of congenital malformation was more common in siblings of consanguineous than non-consanguineous marriages.

The study of Al-Abdulkareem and Ballal [10], in Saudi Arabia, reported a lower mean birth weight of the offspring of consanguineous couples. This may explain the finding that both weight-for-age and height-for-age of our case were at the 25th percentiles.

There was a positive family history of such condition in his older brother's scalp, which resolved spontaneously within 3 weeks after labor, without any medical intervention. There was no previous history of hospitalization to the child.

The lesion in our case was relatively small (2 × 3 cm). It could be successfully managed after applying mupirocin ointment topically, which completely healed leaving a very small atrophic scar.

Garcia-Romero et al. [5] stated that management remains controversial. The lesion's location, size, and depth must be considered. Good results are obtained with conservative treatment in a defect up to 4 × 7 cm in size, while larger defects that extend into the dura need early surgery.

In conclusion, aplasia cutis congenita is a rare condition. Its etiology is still not completely understood, but consanguinity may play a role. Its management depends on several factors, i.e., its pattern, location, underlying causes, and associated anomalies.

Statement of Ethics

The authors confirm that caregivers of their patients were fully informed and they agree to report his case.

Disclosure Statement

The authors have no conflicts of interest to disclose.

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Fig. 1. Tense bulla over the vertex of the neonate.



Fig. 2. Healed ruptured bulla with no residual complication.