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Case Report

Prenatal Diagnosis of Aplasia Cutis Congenita on the Superior and Inferior Limbs with 2D Ultrasound: Case Report

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Abstract

Background: A section of the skin is absent at birth in Aplasia Cutis Congenital (ACC), a condition that can be localized or generalized. This illness affects 0.1 out of every 100,000 newborns. In 60% of instances, it mostly affects the scalp. Statistics show that of all ACC instances, 12% include the trunk and flanks and 25% entail impairment of the lower limbs. Although the exact reasons are unknown, the wounds are linked to genetic factors, teratogens (including alcohol, cocaine, marijuana, heroin, misoprostol, methimazole, and carbimazole), decreased blood flow to the skin, trauma, amniotic bands, and chromosomal problems (trisomy 13).

Case Description: We discuss a case of a female fetus with Aplasia cutis congenital (ACC) on the superior and inferior limbs, which was discovered by a basic fetal abnormality scan at 23 weeks. Small diverse instances have so far been documented globally. This case is the first comprehensive 2D prenatal diagnosis by using ultrasound, the lesions are identified and believed due to familiarity history.

A female patient was delivered vaginally at term. On both her superior and inferior extremities, she is showing a complete loss of skin integument, affecting about 17% of her body surface. Treatment with regional flaps and cream is appropriate given the size and location of the wound.

Conclusion: Because ACC is so uncommon and there were so few patients in each published series in the literature, standardization of treatment is still in its early stages. Additional research is required to diagnose this condition during pregnancy, particularly with ultrasound to assess therapy options.

Keywords: *Aplasia Cutis Congenital; Lower Limbs; Malformation; Treatment; Prenatal Ultrasound*

Introduction

Aplasia cutis congenital (ACC), a complex collection of rare illnesses characterized by a localized or widespread, total or partial lack of distinct layers of the skin at birth, occasionally spreading to the bone, is the term used to describe localized congenital skin deformities [1,2].

This phenomenon was first described by Cordon in 1767 [3]. The condition can present at birth with scarring that represents lesions that have already healed in utero or with the glistening absence of the skin that manifests as well-demarcated, translucent, ulcerated membranes that can be used to see the underlying structures. Pregnancy-related early-stage skin abnormalities may heal prior to delivery and present as an atrophic, membranous, or parchment-like scar in addition to alopecia. The lesions will eventually heal and leave scars. However, less severe anomalies manifest as ulcerations.

According to the frequency was 0.10 per 100,000 live births estimates [3-5].

Pathophysiology of ACC is not well studied and its exact pathogenesis is unknown. However, there are multiple factors that are probably contributing to the development of ACC according to the literature:

- Chromosomal abnormalities, especially BMS1; a recent study has also implicated the UBA2 gene and the SUMOylation pathway
- Trauma
- Amniotic irregularities
- Intrauterine complications, such as vascular accidents or infection
- Thrombosis, vascular lesions
- Teratogens: such as misoprostol, benzodiazepines, valproic acid cocaine, methotrexate, ACE inhibitors, methimazole [7-10].

According to the location of the skin defect, related malformations, associated syndromes, underlying reasons, and teratogens as causative agents, J Frieden divided aplasia cutis congenital into nine types in 1986 [6]:

(i) Group 1: Aplasia cutis congenita of the scalp without multiple anomalies

(ii) Group 2: Aplasia cutis congenita of the scalp associated with limb anomalies

(iii) Group 3: Aplasia cutis congenita of the scalp associated with epidermal and organoid nevi

(iv) Group 4: Aplasia cutis congenita overlying embryologic malformations

(v) Group 5: Aplasia cutis congenita associated with fetus papyraceus or placental infarcts

(vi) Group 6: Aplasia cutis congenita associated with epidermolysis bullosa

(vii) Group 7: Aplasia cutis congenita of the extremities without blistering

(viii) Group 8: Aplasia cutis congenita caused by specific teratogens

(ix) Group 9: Aplasia cutis congenita associated with malformation syndrome.

Our case is exceptional in that the 2D ultrasound used to diagnose the aplasia cutis congenital during pregnancy revealed typical lesions and symmetrical circumferential scarring surrounding both the superior and inferior limbs. We are aware of no prior reports of this ultrasound-detected in-utero condition. Aplasia cutis congenital is thought to be the outcome of abnormal skin growth or aging in utero, which causes scarring or skin deficiency at birth [11,12].

Case Report

A 34 years patient, G4P3, first degree consanguinity with her partner presented at 23 weeks for her second trimester ultrasound screening.

Obstetrical history: Her first child was born with transparent skin, or skin denudation condition, in the lower limbs, and he passed away from a skin infection at the age of 14 days without any further testing. She also had two healthy children who were free of skin conditions.

The results of the ultrasound testing were as follows: Soft tissue thinning in both lower limbs at the ankle site, more noticeable on the left (Figure 1). It was challenging to locate the calcaneum bone in the left lower limb (Figure 2). When a fetus is moving, the amniotic fluid exhibits a snowflake indication (Figure 3). The pregnancy progresses without any complications at 39 weeks.

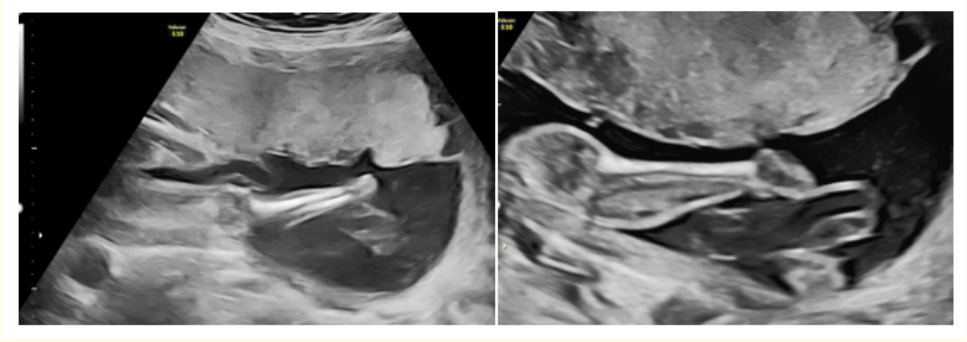

Figure 1: *Ultrasound examination revealed the following findings: Thinning in the soft tissue of both lower limbs at the ankle site, more prominent on the left side.*

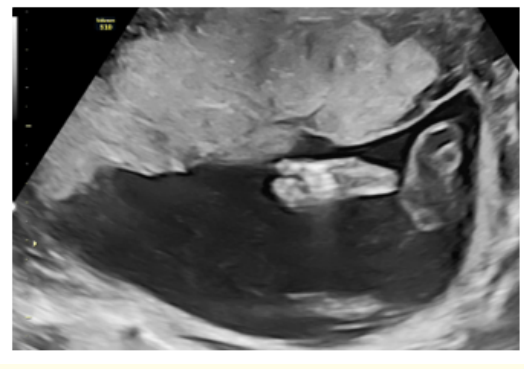

Figure 2: *Calcaneum bone was difficult to identify in the left lower limb.*

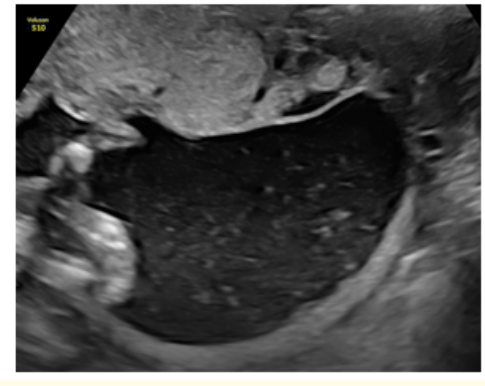

Figure 3: *Snowflake sign in the amniotic fluid.*

A baby girl who was delivered vaginally at full term without any difficulties was referred for congenital skin absence on the left and right lower limbs, primarily at the shin and ankle joint, and for apparent difficulty moving the left leg. The newborn's Apgar scores at 1 and 5 minutes after birth were both 9. Upon examination, it was discovered that the wound was present on the lower limbs. It appeared as bilateral ulcerations with irregularly delineated margins. She developed a significant ulceration on the right lower limb's shin side with erythematous well-defined edges (Figure 4).

Figure 4: *Ulcerations with irregularly delineated margins on the right lower limb's shin side with erythematous well-defined edges, left lower limb appeared severely ulcerated and hypotrophic (absence of calcaneum bone).*

The patient was referred to pediatric care after seven days of life. The patient exhibited a 17% impairment of the body's surface, completely lacking skin integument on the anteromedial face of both legs, and an island of skin on the posterior and plantar regions. The anomaly was the complete absence of all skin and subcutaneous tissue. The lesion had irregularly drawn edges and manifested as ulcerations on the hands and lower limbs, affecting both sides of the legs, feet, and hands (Figure 1).

The pediatric neurologist's neurological examination revealed no signs of sensorimotor neurological dysfunction. Additional neurological testing like electromyography were therefore skipped the calcaneum bone wasn't there, as seen on the radiograph of the left ankle. Results of full blood testing were normal and included serologic examinations for rubella, CMV, and herpes.

Both ulcers received local dressings containing chlorhexidine, antibiotic cream and rehabilitation therapy for hands and limbs has resulted in progressive improvement (Figure 5).

Discussion

Only a few examples of Aplasia cutis congenital on the superior and inferior limbs have been described, and each case had a unique set of clinical characteristics. Nevertheless, our case is the first account of a 2D ultrasonogram with prenatal confirmation and diagnosis.

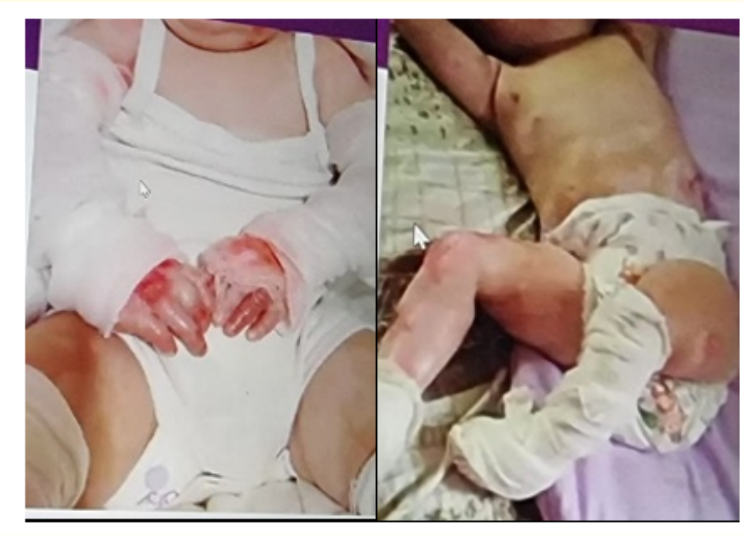

Figure 5: *Local treatment with cream and flaps.*

Skinlessness at birth, a physical characteristic that is unusual, is used to make the clinical diagnosis of ACC. Localized wounds typically have an impact on the scalp as a symptom. Histological characteristics of the wound might range from partial to total absence of skin through impairment of fascia, muscles, and subcutaneous tissue, and eventually impairment of bone and dura mater [13]. Due to its wide dispersion, it rarely spreads to other bodily areas, such as the trunk and limbs.

Like in our patient, ACC frequently manifests as a solitary malformation.

Our research was conducted using a screening ultrasound [11,12]. There are numerous associated single or multiple defects that have been identified that impact the most diverse organ systems (Chart 1) [14,15].

Central Nervous System	Cardiovascular System	Gastrointestinal System	Other systems
Holoprosencephaly	Coarctation of the aorta	Cleft lip and palate	Cutis marmorata
Hydrocephalus	Arteriovenous malformations	Omphalocele	Neural tube closure defects
Spastic paralysis and mental retardation	Patent ductus arteriosus	Tracheoesophageal fistula	

Source: Evers et al.[6].

Chart 1: *Malformations associated with aplasia cutis congenital.*

According to Frieden's classification, the patient under examination appears to fall under category 7, "ACC localized on the extremities, without blistering," which is defined as "ACC involving pretibial areas, the back of the hands and ankles, and wrist extensors [6]".

Depending on the size of the lesion and whether underlying abnormalities are present, several approaches are taken to treat patients with ACC. ACC frequently cures at birth. Small lesions under 4 cm in diameter without other findings should be cleaned daily with a topical antibiotic ointment until full healing has occurred [3,19,20] like our case. The normal healing time for lesions is a few weeks to a few months, leaving an atrophic, hairless scar [3]. Lesions that are larger than 4 cm are more frequently linked to underlying abnormalities and are more vulnerable to consequences such as bleeding, venous thrombosis, and infection [16,17,20]. To prevent these issues, it is advised to have surgery as soon as possible. Considering that certain lesions might be several centimeters in size, skin grafting or flap procedures are frequently used [20].

Protecting the patient from hydration, electrolyte, and caloric shortages, as well as the risk of infection, came before executing the definitive surgical therapy while waiting for the appropriate clinical conditions. Therefore, the patient was under the care of the pediatrics and plastic surgery teams for local care, infection control, and antibiotic administration. For larger wounds, particularly those involving deeper tissues and when vital organs are exposed, local flap treatment is advised (Figure 5).

In cases involving severe wounds, like the one in the present case, adopting a skin transplant appears to be the only option. Most of the similar cases that were documented in the literature experienced the same course of treatment, which led to in complete wound healing [18].

Conclusion

Because this syndrome poses a sizable burden for both the kid and the parents, prenatal diagnosis and family counseling are required. Due to the scarcity of reports on this disease, it may be necessary to describe all characteristics in order to facilitate early diagnosis and appropriate care. Although ACC syndrome is a rare condition, the related impairment necessitates corrective treatment after delivery. Making an informed decision about preparing for their child's impairment and likely orthopedic surgery requires prenatal diagnostics and family counseling. Additionally, since genetic counseling is still unclear in this situation, it must be advised that any future pregnancies have a thorough prenatal ultrasonography.

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