CASE REPORT

Aplasia Cutis Congenita with Fetus *Papyraceus*: A Rare Case Report

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Port J Pediatr 2020;51:260-3
DOI: https://doi.org/10.25754/pjp.2020.18256

Abstract

Aplasia cutis congenita is a rare disorder, characterized by the absence of skin tissue at birth. It occurs most commonly as a solitary cutaneous defect on the scalp and can be classified according to the Frieden classification system. We report the clinical case of a newborn who was noted upon physical examination to have bilateral, large, symmetrical, and well defined lesions with absent skin on both sides of his thoracic region as well as a similar lesion on the right thigh. The pregnancy was a monochorionic, diamniotic twin pregnancy, with the intrauterine fetal death of one of the twins at 13 weeks gestational age. This illustrates a very rare case (with only around 40 cases reported in the literature), of type V *aplasia cutis congenita* (associated with fetus *papyraceus*), with multiple and large area involvement. Despite the extent of the lesions, surgical therapy was not necessary, and medical treatment was sufficient and effective.

Keywords: Diseases in Twins; Ectodermal Dysplasia/diagnosis; Ectodermal Dysplasia/therapy; Fetal Death; Infant, Newborn

Introduction

Aplasia cutis congenita (ACC) is a rare congenital disorder (reported incidence of approximately 1-3 in 10,000 live births), characterized by localized or widespread areas of absence of all skin layers at birth and, in some cases, of subcutaneous tissue and bone.[1-4]

The first case was reported by Cordon in 1767, and in 2012, there were about 500 cases described in the literature.[5] The lesions are typically in isolated and small areas (< 1 cm), well defined and, in the vast majority, occur on the scalp (about 85%). However, face, torso, and extremities can also be affected.[1,6-9] In addition, ACC can occur as an isolated defect or part of a genetic syndrome.[2,10,11] Several classification systems for AAC have been suggested, but the most widely accepted system was proposed by Frieden in 1986, who classifies *aplasia cutis congenita* into nine groups based on the location, pattern, genetic heritage, and associated malformations (Table 1).[1,4,12,13]

Papyraceus is a word derived from the Latin term *papyrus*, a flattened material similar to paper used in ancient times as scrolls.[1,5] The term fetus *papyraceus* refers to a dead intrauterine twin, with subsequent flattening over time (similar to a scroll) due to compression by the growth of the living fetus.[1,5] The exact etiology of fetus *papyraceus* is still unknown.[1,5,6]

The association of ACC with fetus *papyraceous* is classified as type V of Frieden classification system, and is a very rare condition, with only about 40 cases described in the literature.[2,5]

The majority of type V *aplasia cutis congenita* cases only require conservative treatment, adequate hydration, nutrition, and pain control. However, surgical intervention may occasionally be required.[1-3,5,6,10,12,14]

We report the rare case of extensive ACC with fetus *papyraceus* to illustrate the existence of this congenital malformation, help diagnosis, and suggest the most appropriate therapeutic approach.

Table 1. Frieden classification system

Group	Clinical features
1	Scalp ACC without multiple anomalies
2	Scalp ACC associated with limb abnormalities
3	Scalp ACC associated with epidermal or organoid nevi
4	ACC overlying an embryologic malformation (meningomyelocele, gastroschisis, omphalocele)
5	ACC associated with fetus *papyraceus* or placental infarcts
6	ACC associated with epidermolysis bullosa
7	ACC localized to the extremities without blistering
8	ACC caused by specific teratogens
9	ACC associated with genetic malformation syndromes

ACC - *aplasia cutis congenita*.

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Received: 23/01/2020 | Accepted: 12/06/2020 | Published: 02/10/2020

Case Report

Male, newborn, with two symmetric bilateral lesions on the sides of his thoracic region identified after delivery: around 6 cm, well-delimited stellate-like areas of atrophic skin, with a complete absence of epidermis, dermis, subcutaneous fat, and muscle (Fig. 1). Over these lesions, there was a translucent vascularized fascial plane, under which the abdominal contents could be visualized. Another lesion was also identified on the right thigh, with approximately 0.5 cm, and with a similar appearance to the ones previously described (Fig. 1). No other malformations were reported. There was no family history of ACC, vascular abnormalities, vesiculobullous disease, or any congenital disorders. This was a monochorionic, diamniotic pregnancy, with the death of a monozygotic twin at 13 weeks gestational age. Normal fetal growth of the other twin occurred, and his fetal ultrasound throughout the pregnancy was normal. Spontaneous and uncomplicated vaginal delivery occurred at 38 weeks gestational age. During the first observation, no other malformations were noted apart from those previously mentioned. Abdominal, renal, pelvic, and soft tissue ultrasound, at birth, of the injured areas was normal. He was admitted to the ward together with his mother. During admission, conservative treatment was provided with daily silver sulfadiazine and Aquacel® Ag+ bandages and 0.1% betamethasone cream. No fluid therapy was needed. At 3 days of life, a rectal temperature of 38.2ºC was observed and the bandages seemed to have exudate. He was transferred to the neonatology unit to start empiric intravenous flucloxacillin. Blood tests were normal, blood culture was negative, and cultural exudate examination was later positive for methicillin-susceptible *Staphylococcus aureus*. He was maintained on flucloxacillin for a 10 day course of antibiotic therapy, with bandages changed daily. After 7 days of life, the bandages were changed every two days. The patient was discharged at 16 days of life, with almost total reepithelialization of the lesions (Fig. 2), and the bandages continued to be changed once a week at the pediatric day hospital for one month. Follow-up appointments (at 2, 3, 4, 6, and 8 months of life) showed total reepithelialization of the thoracic wounds (Fig. 3), with a small area of hypertrophic scar on the left. In this hypertrophic scar, 0.1% betamethasone cream continued to be applied for two months, with only moisturizing cream applied to the remaining areas. Growth and development milestones were normal.

No genetic studies were conducted, as the diagnosis of *aplasia cutis congenita* is clinical and type V is associated with fetus *papyraceus* without other genetic malformations, as reported in our case.

Figure 1. Skin lesions observed at birth observed bilaterally on the thoracic region and the right thigh.

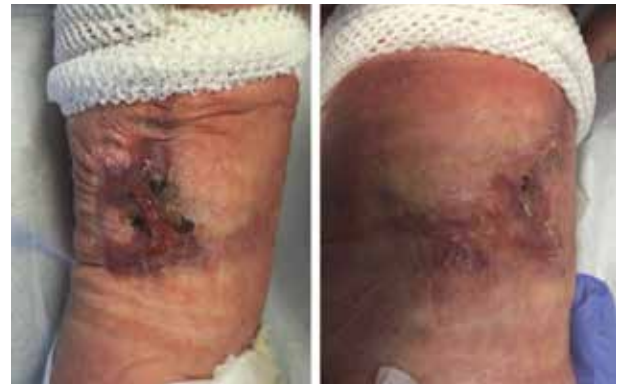

Figure 2. Almost total reepithelialization of the skin wounds at 16 days of age.

Figure 3. Skin wounds in the thoracic region at 6 months of age.

Discussion

About 85% of *aplasia cutis congenita* cases occur on the scalp. Of the remaining cases, only 10% are associated with fetus *papyraceus*.[1,10,12] The exact incidence of ACC combined with fetus *papyraceus* is unknown.[5] It is classified as a very rare condition, with only 40 cases described in the literature at present.[2,5,13] Type V ACC more commonly involves the torso (about 70%), although any region of the body may be affected.[10]

Aplasia cutis congenita lesions are typically small (< 1 cm). However, type V ACC is associated with large affected areas, which may be linear, stellate, and sometimes symmetrical.[1,5] These lesions start early in fetal development and may heal with scarring before birth.[1]

Most cases of *aplasia cutis congenita* (95%) associated with fetus *papyraceus* are observed in monochorionic twin pregnancies.[10,11] Early demise of the fetal twin, usually occurring during the end of the first trimester or the beginning of the second trimester of pregnancy, frequently leads to complete fetal reabsorption, whereas later demise often requires surgical removal of the fetus.[1,3] Curiously, if demise happens before 14 weeks gestational age, the lesions of the other twin are typically on the trunk (which happened in our clinical case), whereas lesions on the extremities are typically associated with demise after 14 weeks of gestation.[1,2]

The exact etiology and pathogenesis of type V ACC is unknown, although studies indicate that it may result from a transient vascular process. [1-3,5,6,10,12,14,15] Several theories have been proposed, of which the two most accepted ones are feto-fetal transfusion with hypovolemia and disseminated intravascular coagulation.[1-3,5,6,10,12,14,15] Other theories have been proposed, such as amniotic membrane adherence, abnormal elastic fiber biomechanical forces and teratogenic drugs.[5] However, these theories are less accepted, as they have fewer scientific studies backing them up.[5]

The feto-fetal transfusion theory is based on doppler ultrasound studies, which show acute transfusion from the living twin to the dying fetus.[5] Decreased blood pressure of the dying twin leads to a blood volume shift from the living fetus, which may cause hypotension and hypovolemia in the living twin, with the resulting ischemia of the skin, and, occasionally, of other organs.[1-3,5,6,10,12,14,15]

The disseminated intravascular coagulation theory suggests that thrombogenic material from the dead fetus to the living twin activates the coagulation cascade, resulting in thrombosis and ischemia of the skin.[1-3,5,6,10,12,14,15]

Prenatal diagnosis of ACC is not possible with the current imaging techniques and is made clinically at birth.[10]

Treatment of type V ACC consists of conservative or surgical interventions, depending on the size and location of the lesions, and the experience of the medical provider.[1,2,5,10,11] Surgery, through flaps or grafts of skin, carry the risk of hemorrhage, infection, possible complications from anesthesia, and necrosis of the grafted area. Medical treatment, successfully used in the majority of cases, consists in healing the skin by secondary intention, using silver sulfadiazine dressings twice a day and saline infusions. This treatment also carries the risk of hemorrhage, infection, skin necrosis, thrombosis, pain during wound tending, and increased epidermal water loss. Control of these key points is essential. Mean duration of reepithelialization, with conservative treatment, is around 28 days, and the scar tissue forms around 3-4 months. When there is severe fluid loss, infection, and delayed epithelialization, then surgical intervention should be considered.[1,2,5]

As both interventions are not without risk, a definitive consensus for treatment has not yet been achieved. The most important factors on deciding between medical or surgical care are the size of the lesion and its location.[1,2,5,10]

It is known that, despite being large in size, thoracic lesions, which are very common in type V ACC, can usually be managed using only conservative treatment, with satisfactory results, although an atrophic or hypertrophic scar can sometimes remain. For the large scalp lesions, surgical treatment is preferred to prevent complications, such as meningitis or severe hemorrhage.[1,2,5,10]

Mortality rates are around 20%-55% in scalp ACC, often secondary to surgical complications, sagittal sinus hemorrhage, infections, or associated congenital defects.[1,10] ACC combined with fetus *papyraceus* has a significantly lower mortality rate, despite the presence of large areas, as these lesions are located in regions with more reduced risk (trunk and extremities).[1] An important prognostic factor is the time of gestation when the twin fetal death occured.[5] If fetal death occurs during the first and early second trimester, the other twin fetal viability is more likely.[5] If the fetal death occurs during the second half of the pregnancy, the other twin fetal viability is less likely, and may develop extensive aplasia cutis and cerebral complications.[5]

The authors present a case of ACC associated with fetus *papyraceus*, a rare condition, where the pregnancy outcome (with the fetal twin demise), and the appearance of the lesions was similar to the few cases described in the literature. Conservative treatment lead to their satisfactory healing.

WHAT THIS CASE REPORT ADDS

- Type V *aplasia cutis congenita* is a rare condition, typically observed in monochorionic twin pregnancies, with the fetal demise of one of the fetuses.
- Diagnosis is clinical, through clinical examination and pregnancy history. In most cases, other diagnostic tests or genetic studies are not required.
- Serious complications are uncommon, and appropriate follow-up is essential.
- Medical treatment is sufficient in most patients, with surgical intervention used in patients with severe fluid loss, infection, and/or delayed epithelialization.

Conflicts of Interest
The authors declare that there were no conflicts of interest in conducting this work.

Funding Sources
There were no external funding sources for the realization of this paper.

Provenance and peer review
Not commissioned; externally peer reviewed

Consent for publication
Consent for publication was obtained.

Confidentiality of data
The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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Aplasia da Cutis Congénita com Feto Papiráceo: Um Caso Clínico Raro

Resumo
A aplasia cútis congénita é uma condição rara, caracterizada pela ausência de tecido cutâneo ao nascimento. Habitualmente ocorre como uma lesão isolada que atinge o couro cabeludo. Pode ser classificada em 9 tipos, de acordo com o sistema de classificação de Frieden. Relata-se um caso clinico de um recém-nascido, que ao exame físico apresentava bilateralmente no abdómen, lesões simétricas de grandes dimensões, bem delimitadas, de ausência de tecido cutâneo, assim como uma lesão semelhante na coxa direita. A destacar gravidez gemelar, monocoriónica, biamniótica, com morte fetal in útero de um dos gémeos às 13semanas de gestação. Ilustra-se assim, um caso muito raro (cerca 40 casos descritos na literatura), de aplasia cútis congénita tipo V (associado a *feto papyraceus*), com envolvimento de múltiplas áreas e de grandes dimensões. Apesar da extensão das lesões, a terapêutica cirúrgica não foi necessária, tendo o tratamento conservador instituído sido suficiente e eficaz.

Palavras-Chave: Displasia Ectodérmica/diagnóstico; Displasia Ectodérmica/tratamento; Doenças em Gémeos; Morte Fetal; Recém-Nascido