

# Aplasia Cutis Congenita Type V: a case report and review of the literature

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## Abstract

Aplasia cutis congenita is a relatively rare congenital anomaly that most commonly occurs as a solitary cutaneous defect on the scalp. Depth of involvement varies and involvement of deeper calvarium and dural structures can be seen in more severe cases. Multiple classification systems have been devised with the Frieden Classification System being the most widely adopted. Using this system, we describe a patient that developed Type V aplasia cutis congenita (ACC) with associated fetal papyraceous. The child healed remarkably well with application of petrolatum impregnated gauze and topical silver sulfadiazine twice daily for approximately 4 weeks. The child was noted to have no significant contractures or complications at 6 months and 1 year follow-up exams.

## Case

A 37w4d male was born to a G8P5 mother after a complicated, monochorionic twin pregnancy. At 16 weeks gestation mother experienced fetal demise of one fetus. The surviving fetus developed hydrops fetalis and severe anemia requiring a fetal blood transfusion. At the time of delivery the male infant was noted to have significant absence of skin on the lateral torso and vertex scalp (Image 1). There was also apparent stellate scarring on the elbows, knees and hips. This was presumed to have been areas of aplasia cutis that had begun the healing process in utero. The remaining open wounds on the torso were initially dressed with petrolatum impregnated gauze and then a regimen of topical silver sulfadiazine was implemented (Image 2). The parents applied these dressings twice daily for approximately 4 weeks. At two weeks postpartum the infant developed a fever of unknown origin. Bacterial cultures from the healing wounds were taken and returned negative. At 4-8 weeks postpartum the areas of involvement demonstrated significant healing (Image 3). No significant contractures or complications were noted with routine exams at 6 months, 12 months and 2 years (Image 4).



Image 1- Large defect on the lateral torso

Image 2- Healing at 1 week old with conservative wound care



Image 3- At 2 months old, defect has nearly closed

Image 4- At 2 years old, well formed scar without contracture or restrictions in mobility.

## Background

Aplasia cutis congenita (ACC) involves the congenital absence of a localized or widespread area of skin occurring in approximately 1-3 out of every 10,000 live births. It is most commonly observed in the scalp (84% of cases,<sup>1</sup> 86% of solitary lesions<sup>2</sup>) but can affect any part of the body. It was first described by Cordon in 1767 and later by Campbell in 1826.<sup>3,4,5</sup> Typically a clinical diagnosis with findings of single or multiple circular, oval, linear, stellate or rhomboidal defects with varied depth from upper dermis down to dura in 15-30% of cases.<sup>6</sup> Mortality rates ranging from 20% to 55% have been reported in association with large areas of scalp involvement, often secondary to sagittal sinus hemorrhage, surgical complications, or associated congenital defects.<sup>1</sup> ACC has been reported to occur in approximately 1-3 out of every 10,000 live births.<sup>7</sup> The most widely used system to classify ACC was developed by Frieden (Table 1).<sup>2</sup> Using this system, the patient discussed above was diagnosed with type V aplasia cutis congenita with fetus papyraceus.

Group/Subtype	Clinical Features
1	Scalp aplasia cutis congenita without multiple anomalies
2	Scalp aplasia cutis congenita with associated limb abnormalities
3	Scalp aplasia cutis congenita with associated epidermal and organoid nevi
4	Aplasia cutis congenita overlying embryologic malformation
5	Aplasia cutis congenita w/ associated fetus papyraceus or placental infarcts
6	Aplasia cutis congenita associated with epidermolysis bullosa
7	Aplasia cutis congenita localized to extremities without blistering
8	Aplasia cutis congenita caused by specific teratogens
9	Aplasia cutis congenita associated with malformation syndromes

Table 1- Freiden Classification for Aplasia Cutis Congenita<sup>2</sup>

## Discussion

In contrast to the other types of Aplasia cutis congenita, type V most commonly affects the trunk and is often symmetric (Table 2).<sup>1,8</sup> ACC with fetus papyraceus is typically observed in association with monochorionic twin pregnancies (95% of cases).<sup>1</sup> It has been noted that if demise occurs prior to 14 weeks gestation, ACC typically will develop on the trunk versus the extremities with fetal demise after 14 weeks.<sup>8,9</sup> Following twin fetal demise, some believe that an ensuing transient hypovolemia leads to ischemia of watershed areas of the skin<sup>4,10</sup> while others have suggested that thrombi formation in the setting of disseminated intravascular coagulation of the dying fetus embolizes to the healthy twin.<sup>9</sup> An exact pathogenesis has not been proven but it is almost certain that a transient vascular process is responsible for the clinical findings observed in type V ACC.

Body Area	Percent Affected
Flank	70%
Buttock/Thighs	60%
Abdomen	33%
Scalp	26%
Axilla/arms	21%
Back	16%

Table 2- Type V ACC distribution<sup>1,8</sup>

One of the greatest risks in type V ACC, is neonatal development of infection. As a result, early intervention is directed towards minimizing this risk. Depending on the size of the defect, treatment using surgery (skin grafts or flaps) or conservative wound care is often employed. Surgical risks include potentially fatal hemorrhage, infection, and anesthesia complications. Supportive wound care carries risk of hemorrhage, sagittal sinus thrombosis, wound bed necrosis, and infection. Because both carry significant risk of complications a definitive consensus for treatment has yet to be achieved. Most experts agree that conservative wound care is appropriate in the majority of cases with size of defect and location the most important factors to consider. In general, skin grafting is often reserved for defects larger than 2-4 cm; particularly with scalp defects.<sup>4,11</sup> One review of 11 cases of type V ACC associated with twin loss demonstrated successful reepithelialization and later scar formation in 10 of the cases.<sup>9</sup> In the remaining case, skin graft became necessary due to the development of bacteremia.<sup>9</sup> Conservative wound care has found similar success in other cases including our patient.

As a result, we feel that any treatment algorithm for type V ACC should rely on conservative wound care and infection prevention with more invasive methods utilized if complications arise. A basic regimen could include sequential application of silver sulfadiazine, petrolatum gauze, dry gauze and a self-adherent wrap with dressing changes twice daily.<sup>9</sup> Use of antibiotic ointment can be considered in place of silver sulfadiazine for any concern of toxicity with close monitoring for fungal overgrowth.<sup>9</sup> Antibiotics (topical or IV) should be reserved for signs that are suggestive of infection. Due to the risk for infection or significant electrolyte disturbances with conservative care, it is imperative to monitor closely until reepithelialization has occurred. Surgical intervention should primarily be considered in cases of refractory fluid loss, stalled epithelialization, and infection.<sup>9</sup>

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