

## Case Report

# Aplasia cutis congenita with fetus papyraceus: an uncommon case report

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### ABSTRACT

Aplasia cutis congenita is a condition characterized by congenital absence of all skin layers in a localized or widespread area. Frieden's classification recognized a rare subtype, type V, that is associated with multiple gestations in which there is the in-utero demise of a twin with resultant fetus papyraceus or mummification. A twin pregnancy was complicated by in-utero death of one twin at 5 months. On full term birth of the other twin with fetus papyraceus large defects in the skin of knees were noted bilaterally. Conservative management with topical antibiotics and emollients helped in complete re-epithelialization in few months. As the use of reproductive technologies increase the incidence of multiple gestation and associated conditions are expected to increase. This type is clinically unique in that it is characterized by stellate lesions in a symmetrical distribution over the trunk and extremities, differing from other subtypes, which are typically localized to scalp (70%-85% of cases) or extremities. Management ranges from conservative to surgical grafts.

**Keywords:** Aplasia cutis, Fetus papyraceus, Congenital absence of skin

### INTRODUCTION

Aplasia cutis congenita (ACC) is congenital absence of the skin, first described in 1767 by Cordon.<sup>1</sup> The lesions are typically well demarcated and stellate and may have granulating tissue at the margins. Depending on location, there can be variable absence of the epidermis, dermis, subcutaneous fat, adnexal structures, and underlying bone.<sup>1-3</sup> The etiology and pathogenesis are unknown, although several theories exist, including vascular compromise (hypotension or thrombosis), trauma, infection, and teratogens.<sup>4,5</sup> Approximately 85% of the lesions are located on the scalp, with the remainder occurring on the trunk and limbs.<sup>2,6</sup> ACC in general can be isolated lesions or have associated congenital anomalies.<sup>7</sup> It is typically sporadically inherited, but there are reports of familial cases.<sup>5</sup> In an attempt to categorize ACC, Frieden proposed a classification system based on location and associated anomalies.<sup>7</sup> When ACC is

associated with a monochorionic twin pregnancy in which one of the fetuses is lost, referred to as fetus papyraceus, the designation is type V ACC. When the co-twin demise happens before 14 weeks gestation, the lesions are typically on the trunk. If the co-twin demise happens after 14 weeks, the lesions are characteristically on the extremities.<sup>8,9</sup> Affected areas may appear membranous or partially healed with contractures depending on when the insult happened. The etiology of this specific type of ACC is thought to be due to hypotension or disseminated intravascular coagulation associated with the loss of the monozygotic twin.<sup>10-12</sup> There are vascular anastomoses in monochorionic twins, so when the co-twin dies, it is conceivable that the surviving fetus is exposed to emboli and or hypovolemia affecting watershed areas of the surviving twin.<sup>13</sup> We report a case of a neonate with ACC on Knees associated with the loss of a monozygotic twin. The focus of this report and discussion is on conservative management and prognosis of type V ACC.

## CASE REPORT

A twin pregnancy was complicated by in-utero death of one twin at 5 months. For the remainder of pregnancy, another twin was monitored frequently with ultrasound scan. No anatomical abnormalities were noticed. Live fetus didn't have any growth retardation or distress during the rest of the pregnancy. Delivery was at full term at 38 weeks. Live baby was delivered along with mummified dead fetus. On delivery it was noticed that large portion of skin over both knees were missing (Figure 1). There was no history of vesicles or bullae in that area. There was no history of preceding trauma to that area. Vitals of the newborn were normal. Dressing with topical Silver sulfadiazine was given for few weeks after delivery. Any signs of infections were looked for. Adequate fluid balance was maintained to replenish the loss through the lesion. Topical Silver sulfadiazine was stopped once the lesion became dry and then kept opened as the healing ensued. Emollients were used for the scar after healing. Controlled mobilization of knee joints and massaging was done after healing in order to avoid contractures and movement restriction. Re-epithelialization was complete in few months. 4 months after delivery, infant had a well-defined atrophic scar over bilateral anterior knees with less contracture (Figure 2).



**Figure 1: On 3rd day after delivery, lesion over (A) left knee (B) right knee.**



**Figure 2: Atrophic scar over both knees after 4 months of delivery**

## DISCUSSION

There are not clear recommendations for the management of ACC. The literature describes management of giant scalp lesions in conservative and an immediate surgical manner.<sup>14,15</sup> With large scalp lesions and exposed duramater, there is the potential for meningitis, fluid and electrolyte imbalance, and bleeding from the sagittal sinus and adjacent vessels.<sup>14-17</sup> With a large ACC lesion on the trunk or lower extremities, there is the potential for fluid and electrolyte shifts and infection, but there is not the risk of bleeding or meningitis seen with scalp lesions. The treatment and outcomes seen with large scalp ACC offer some insights when treating nonscalp ACC in that lesions on the trunk and extremities have the potential need for urgent surgical coverage, albeit only if conservative management fails or is not an option. The pediatric population presents a unique situation in which children anecdotally are able to heal larger wounds than adults. Depth of nonscalp ACC is difficult to assess. As in this case, there appeared to be only a thin fascial layer keeping the abdominal and thoracic contents in. This film-like layer appeared highly susceptible to desiccation and did not appear adequate to support neoplastic epithelialization. Maintaining a moisture-rich, antimicrobial environment was imperative in our attempt to treat this lesion in a non-surgical manner. Larger and deeper lesions are at increased risk for fluid and electrolyte derangements and subsequent contractures and scarring. Infection is always a risk with exposed soft tissue. Close monitoring of electrolyte and fluid status is essential in the first few weeks of life. The emphasis of our treatment plan is to proceed conservatively and be prepared to address complications if they arise. To minimize fluid loss and infection, we recommend an antimicrobial and near-occlusive dressing. Dressings should be changed twice daily to maintain an adequate moist environment. At the first sign of infection, intravenous or topical antibiotics can be started, depending on the clinical nature of the infection. If there is a cessation of epithelialization, this needs to be assessed to see if local debridement with a different type of dressing care or surgical debridement would facilitate continued skin growth. If there is lack of progress in epithelialization or signs of infection or if fluid losses become difficult to manage, skin grafting becomes an option. Skin grafting or coverage with cellular membranes can only occur when there is no evidence of infection. As wounds epithelialize, they start to contract. As with burns, we recommend vigorous massage for a full year before attempting surgical release of the scars. Scars are typically considered mature after approximately 1 year, and scar revision is typically instituted after this time. Potential options would be dermabrasion and fractionated laser therapy, depending on the appearance of the scar. Another possibility would be continued observation. Even with skin grafting or use of a synthetic bio membrane, scarring is a concern, and there is no clear evidence that either of these offers a better cosmetic outcome. The primary reasons to employ skin grafting or

wound coverage are if the wound fails to epithelialize fully or complications arise from fluid or electrolyte imbalance. There has been some concern in the literature about potential toxicity and side effects from silver sulfadiazine when used over large surface areas. Some of the reported side effects from silver sulfadiazine include anemia, renal insufficiency, seizures, and high liver enzymes. Little is known about dose and duration of therapy that leads to toxicity.<sup>18</sup> A caveat to our treatment algorithm would be that, if there is evidence of side effects from the silver sulfadiazine, it can be discontinued and switched to antibiotic ointment and petrolatum gauze dressings twice daily, but topical antibiotic use can lead to overgrowth of fungal skin elements, and the patient would need to be monitored. Another alternative would be to use petrolatum covered by petrolatum gauze, realizing that a large skin defect such as ACC is at major risk for infection, to close observation would be warranted given the lack of antimicrobial coverage. Giant scalp ACC has been treated with silver sulfadiazine without sequelae.<sup>14</sup> Furthermore, there are large case series of exomphalos major being treated conservatively with silver sulfadiazine also without side effects.<sup>19,20</sup> It remains our practice to use silver sulfadiazine with large ACC lesions.

## CONCLUSION

Diagnosis of ACC is clinical with a few reports on histopathology, which reveal absence of epidermis, dermis, adnexa and sometimes subcutaneous tissue depending upon the depth of the defect. Regular ultrasonography during antenatal checkup sometime helps in early diagnosis of ACC. The normal fetal skin generates strong echoes on ultrasound while in ACC such echoes are absent. Management of type V ACC is going to become increasingly important with increasing use of in vitro fertilization and other fertility treatments. These therapies are known to increase the incidence of multiple gestations. Our conservative approach offers protection from infection and attempts to prevent fluid and electrolyte derangements. Furthermore, it allows the therapy to be adapted as the lesion heals. At any point along the way, the plan can be adapted based on the clinical status of the patient. An additional benefit of a conservative approach is that it may extend lengths of stay in the hospital because patients can be followed closely on an outpatient basis.

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