

Type V aplasia cutis congenita with fetus papyraceus



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INTRODUCTION

Aplasia cutis congenita (ACC) is a condition characterized by the congenital absence of all skin layers in a localized or widespread area. This phenomenon was first described by Cordon in 1767.¹ More than 2 centuries later, Frieden² proposed a classification system for clinical subtypes of ACC based on the affected area(s), associated anomalies, and mode of inheritance. This categorization recognized a rare subtype of ACC that is associated with multiple gestation in which there is the in utero demise of a twin with resultant fetus papyraceus (FP), or mummification. FP is thought to develop from compression of the expired twin by the viable fetus as it grows, causing a phenomenon known as *vanishing twin syndrome*. Frieden² designated this subset of cases as type V ACC. ACC with FP is clinically unique in that it is characterized by stellate lesions in a symmetrical distribution over the trunk and extremities, differing from other subtypes of ACC, which are typically localized to the scalp (70%-85% of cases) or extremities. Apart from isolated scalp involvement, there are little epidemiologic data delineating the frequency of other distribution patterns. Here we present a case of type V ACC associated with FP.

CASE PRESENTATION

A twin pregnancy was complicated by the in utero death of twin A at 13 to 14 weeks. Subsequently, amniocentesis results were notable for murky-colored amniotic fluid, suggestive of possible bleeding or infarct. Karyotype and microarray were normal. At 19 weeks, magnetic resonance imaging and ultrasound scan of twin B were reassuring, and no anatomic abnormalities were found. For the remainder of the pregnancy, twin B was monitored

Abbreviations used:

ACC: aplasia cutis congenita
FP: fetal papyraceus

biweekly by a perinatologist. He continued to show consistent growth and development but was classified as having intrauterine growth restriction, for which delivery was induced at 35 weeks. Upon delivery, it was evident that the infant was missing large portions of skin on his trunk and proximal extremities (Fig 1). The plastic surgery department recommended skin grafting, but the decision was ultimately made to use conservative management. This approach consisted of twice-daily dressing changes using petrolatum and an absorbent hydrophilic polyurethane foam dressing. Over the ensuing weeks, the involved areas granulated in steadily with no complications (hemodynamic, infectious, or otherwise). By 3 months, there was complete re-epithelialization (Fig 2). Notably, placental pathology found a smaller-than-expected chorionic plate (13 × 11 cm) and reduced placental thickness (1.7 cm), measurements typically consistent with a pregnancy at 21 to 24 weeks' gestation. Histology found small villi consisting of much denser connective tissue and far fewer blood vessels than would typically be expected.

DISCUSSION

ACC associated with FP is categorized as type V. Although most cases of type V ACC occur in twin pregnancies, they have been described in triplet and even sextuplet gestations.³ Reports clearly show that most fetal deaths in type V ACC occur in the late first or early second trimester. A review of 34 cases of type

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Fig 1. At birth, extensive involvement of the trunk and proximal extremities was observed.

V ACC found a mean gestational age of fetal demise at 13.3 weeks, with an approximately 1:1 male/female ratio.⁴ Interestingly, in all but 2 of these cases, the maternal condition was normal prior to delivery, suggesting that maternal comorbidities may not play a role in the elusive pathogenesis of this condition. However, the placental pathology findings in our case do suggest an underlying issue with diminished maternal blood flow into the placenta.

Most investigators postulate that type V ACC results from a complication of shared blood flow through vessel anastomoses in monochorionic gestations. Some have surmised that the insult stems from emboli secondary to disseminated intravascular coagulation within the dying fetus, whereas others suggest that the lower pressure system of the deceased twin draws blood away from the survivor, leading to hypovolemia and resultant skin necrosis.⁵⁻⁷ The characteristic involvement of the trunk and extremities is thought to represent watershed areas that are farthest from the vascular supply and therefore most susceptible to ischemic insults. Indeed, in the case of our patient, histologic changes noted in the chorionic villi were suggestive of diminished maternal blood flow to the placenta. This attenuated blood supply may have contributed to the loss of twin A, which we theorize resulted in diversion of blood flow to the low-pressure system of the deceased. Such a phenomenon would force the surviving twin to transiently shunt the remaining blood supply inwardly to perfuse his vital organs, resulting in ischemic skin necrosis within watershed



Fig 2. By 12 weeks, there was complete re-epithelization.

areas. Diagnostically, ACC is clinically determined; biopsies are typically not performed. If a biopsy is acquired, the absence of epidermis, dermis, and subcutaneous fat is seen.

The appropriate treatment of type V ACC is not well studied. Like burn patients, babies with ACC are susceptible to desiccation, infection, and electrolyte imbalance. Therefore, Morrow et al⁸ proposed an approach to patients with type V ACC that is derived from the burn literature. They suggest using the antimicrobial agent silver sulfadiazine on exposed areas followed by the application of petroleum gauze, dry gauze, and a self-adherent wrap, respectively. This regimen is designed to minimize fluid losses and microbial exposure. Alternatively, more conservative therapy with exclusively petrolatum and foam gauze has yielded favorable results in numerous patients. Once management is initiated, regular follow-up to evaluate the healing process is critical. During re-epithelialization, contractures can form from scar tissue, creating tight bands across the patient's trunk or extremities. In these instances, Morrow et al⁸ endorse the use of rigorous massage to loosen the tissue, thus avoiding unnecessary surgical release. Similarly, physiotherapy may be beneficial in ensuring that scar contracture does not limit limb movement in any functional way. If conservative management is unsuccessful, if extensive scalp involvement leads to exposure of the dura, or if there are complications of fluid/electrolyte losses or infection, surgical grafting may be warranted and has been successful in several cases.^{4,8,9} However, criteria for the appropriate utilization of allografts and/or autografts has been inconsistent. For example, some investigators suggest the use of surgical grafting for all denuded areas larger than 3 to 4 cm.⁴ Conversely, our patient and others with defects as large as 8 × 12 cm were successfully treated with conservative therapy alone.¹⁰ As surgical grafting carries risks such as hemorrhage, infection, and graft necrosis, it should be avoided when conservative management

is clinically deemed appropriate. Lastly, although serious complications such as sepsis have been reported, most patients with type V ACC suffer few to no sequelae and recover from this condition with complete re-epithelization and scar formation within a few months. In summary, type V ACC is a rare subtype of ACC that is associated with multiple gestation in which there is the in utero death of a twin with resultant fetal papyraceus. As the use of reproductive technologies increases, the incidence of multiple gestations and associated conditions is expected to increase. It is therefore pertinent that clinicians recognize and effectively treat these maladies, yet, limited literature exists regarding the proper management of type V ACC. We therefore present a case of type V ACC that was effectively treated with conservative therapy without the need for surgical intervention.

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