Extensive Aplasia Cutis Congenita and the Risk of Sagittal Sinus Thrombosis

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Critical Situations: Dermatology in the Acute Care Setting

REPORT OF A CASE

A full-term 1-day-old boy weighing 3140 g was transferred to our tertiary care children’s hospital with large areas of skin defects on his scalp, trunk, and extremities. His parents were nonconsanguineous, and the results of prenatal maternal laboratory screening for human immunodeficiency virus, hepatitis B, group B streptococci, and rapid plasma reagin were negative. Intrauterine fetal death of a twin had been reported at 16 to 18 weeks of gestation. No other complications were noted during the pregnancy.

On physical examination, a full-thickness ulcer of the scalp vertex was observed extending from the anterior fontanel to the posterior fontanel in an area overlying the sagittal sinus. The defect measured $5 \times 7$ cm and revealed exposed dura and subcutaneous vessels (Figure 1). Two symmetric, vertically oriented, full-thickness defects, each measuring 3.2 cm, with minimal surrounding erythema, were noted on the lateral aspect of the abdomen (Figure 2). Examination of the knees revealed symmetric full-thickness defects that appeared as yellow-red, indurated ulcers. Symmetric, linear scars were observed bilaterally in the axillary and inguinal creases.

Magnetic resonance imaging of the head and a magnetic resonance venogram revealed patent major intracranial venous sinuses and slight hypoplasia of the left transverse sinus (considered a normal congenital variation). A relative lack of subcutaneous fat over the superior aspect of the skull was also observed, consistent with the clinical presentation.

Concern that the extensive open defect posed a high risk for sagittal sinus thrombosis, hemorrhage, or meningitis prompted plastic surgery and neurosurgery consultations. The scalp defect was repaired by plastic surgery with a skin graft, while the remaining areas of aplasia cutis congenita were allowed to heal by secondary intention. Over the next 3 weeks, the scalp healed well, without complications, as did the other areas of aplasia on the abdomen and knees.

The initial differential diagnosis included aplasia cutis congenita of varying subtypes, focal dermal hypoplasia, and amniotic band defects.1 With the history of fetal death of a co-twin in the early second trimester, and the characteristic extensive areas of symmetric skin defects, the diagnosis of type V aplasia cutis congenita associated with a fetus papyraceus, based on Frieden’s2 classification, was made.

The major challenge in this case hinged on the management of the large skull defect. The mortality rate of patients with extensive scalp aplasia cutis congenita has been estimated to be as high as 20% to 30% and is generally the result of hemorrhage, thrombosis, or infection. Therefore, prompt diagnosis and appropriate management are critical in avoiding adverse outcomes.

In addition to providing an accurate diagnosis, dermatologic consultation provided significant benefit in the management of this case by identifying the risk of existing or future sagittal sinus thrombosis or hemorrhage, the need for vascular imaging, and the high risk of complications without surgical closure.

COMMENT

Aplasia cutis congenita reflects a spectrum of cutaneous and subcutaneous tissue defects. Most frequently, aplasia cutis congenita presents as a small, isolated defect in the scalp of neonates along the lines of embryonic fusion. This limited form of aplasia cutis congenita must be distinguished from traumatic ulceration due to scalp electrode placement in the perinatal period.3

Frieden4 has classified aplasia cutis congenita into 9 subtypes, based on clinical presentation, prognosis, and associated conditions. Type V aplasia cutis congenita is associated with a fetus papyraceus, or mummified co-twin present at birth. The fetus papyraceus is formed after the co-twin dies early in the second trimester. This...
rarely described entity presents with characteristically extensive symmetric areas of cutaneous and subcutaneous skin defects. Most cases of type V aplasia cutis congenita are not associated with other congenital anomalies. Areas of skin aplasia generally heal well, often with scarring. Secondary infection is uncommon except in cases of extensive scalp aplasia cutis congenita.

The etiology of aplasia cutis congenita remains poorly elucidated. Clearly, genetic predisposition plays a role in the development of this disorder, but other factors, including intrauterine infection, amniotic adhesions, intruterine necrosis, and teratogens, may play a role.4-10 Also, some authors have proposed that areas of aplasia result from watershed areas of necrosis due to impaired vascular supply.11 Similarly, scalp aplasia has been proposed as a result of tissue tension in the setting of head growth, leading to a further impaired vascular supply and tissue necrosis.

Type V aplasia cutis congenita associated with fetus papyraceus may result from vascular compromise due to the co-twin’s death in the early second trimester. Whether the vascular compromise leads to embolization of vessels or to a differential supply of oxygenated blood remains unclear. The end result involves areas of full-thickness skin aplasia that may be the product of impaired vascular supply early in the second trimester.12,13 The scars in the groin and axillae in our patient represent areas of aplasia that were able to heal over the ensuing months of gestation between the insult and birth.

Aplasia cutis congenita has been associated with a number of complications. In 1826, Campbell15 described 2 cases of scalp aplasia cutis congenita, one of which was complicated by fatal sagittal sinus hemorrhage 18 days after birth. More recently, the mortality rate of patients with extensive scalp aplasia cutis congenita has been estimated to be as high as 20% to 30%. Death is generally a result of hemorrhage, thrombosis, or infection. Therefore, prompt diagnosis and appropriate management are critical in avoiding adverse outcomes. The surgical literature is divided on the issue of scalp aplasia cutis congenita with regard to aggressive and conservative management, although in cases of exposed dura or sagittal sinus, surgical intervention should probably be undertaken.

In conclusion, prompt intervention should be considered in patients who present with large areas of scalp aplasia cutis congenita. While some evidence exists to support conservative dressing changes and the avoidance of active surgical intervention, several cases of dramatic and often fatal sagittal sinus hemorrhage associated with large full-thickness scalp and skull defects over the sagittal sinuses have been reported. Therefore, consultation with surgical services, including neurosurgery and plastic surgery, is of critical importance in the treatment of these patients. If patients are treated conservatively with dressing changes alone, they should be monitored closely for sagittal sinus hemorrhage and infection, and parents should be informed of the potential for such complications. Fortunately, our patient had an excellent outcome after surgical intervention, and this case highlights the importance of rapid diagnosis, familiarity with potential complications of large scalp defects in aplasia cutis congenita, and close communication and coordination among clinical services in the management of this potentially fatal disorder.

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REFERENCES


**ARCHIVES Web Quiz Winner**

Congratulations to the winner of our February quiz, Yalda Nahidi, MD, first-year resident of dermatology, Division of Dermatology, Mashhad Medical University, Mashhad, Iran. The correct answer to our February challenge was Bazex syndrome. For a complete discussion of this case, see the “Off-Center Fold” section in the March ARCHIVES (Webb KG, Malone JC, Callen JP. Acral psoriasiform eruption in a man with squamous cell carcinoma of the tonsillar pillar. *Arch Dermatol*. 2005;141:389-394).

Be sure to visit the Archives of Dermatology Web site (http://www.archdermatol.com) to try your hand at the Interactive Quiz. We invite visitors to make a diagnosis based on selected information from a case report or other feature scheduled to be published in the following month’s print edition of the ARCHIVES. The first visitor to e-mail our Web editors with the correct answer will be recognized in the print journal and on our Web site and will also receive a free copy of *The Art of JAMA II*.