Extremely Low Birth Weight Infant Born with Extensive Abdominal Cutis Aplasia

Oren Gordon MD PhD[1], Sinan Abu-Leil MD[2], Yotam Almagor MD[2], Elite Cohen MD[2], Alexander Margulis MD[3], Dan Arbell MD[4], Benjamin Bar-Oz[2] and Smadar Eventov-Friedman MD PhD[2]

Departments of [1]Pediatrics, [2]Neonatology, [3]Plastic Surgery and [4]Pediatric Surgery, Hebrew University-Hadassah Medical Center, Jerusalem, Israel

KEY WORDS: aplasia cutis congenita (ACC), extremely low birth weight, intrauterine growth retardation, prematurity

IMAJ 2015; 17: 719–721

Aplasia cutis congenita (ACC) is a condition characterized by congenital absence of the skin that may involve all layers of the skin, adnexal structures and underlying bone. The pathogenesis is currently unknown but several theories have been suggested, including hypotension, thrombosis, trauma, infection, and exposure to teratogens. Most cases are sporadic, but cases with family association have been described. There are currently at least 500 cases reported in the literature and about 85% of these include solitary lesions of the scalp, although any other area of the body may be involved. For clinical practice, Frieden's classification – which is based on the distribution of the lesion(s), associated anomalies, infections or syndromes – is currently used [1]. Type-V ACC in Frieden's classification is associated with the loss of a monochorionic twin during the late first to early second trimester, otherwise known as fetus papyraceus (FP). At least 44 cases of ACC and FP have been reported and recently summarized in two reviews [2,3] detailing the distinctive bilateral symmetrical lesions over the trunk, buttocks and/or extremities. Almost all the cases describe full- or near-term neonates.

We report here a case of type V ACC in a premature extremely low birth weight infant and highlight some of the unique features of the clinical course and treatment considerations, leading to abdominal skin recovery and his discharge from the neonatal intensive care unit (NICU) without major morbidity.

PATIENT DESCRIPTION

Intrauterine insemination resulted in a monochorionic di-amniotic twin pregnancy in a 26 year old primipara healthy mother. After 12 weeks of gestation spontaneous intrauterine demise of one of the twins was diagnosed. At 28 weeks gestation she was hospitalized due to suspected intrauterine growth retardation and underwent cesarean section at 29+0 weeks gestation for non-reassuring fetal heart rate monitoring. A male infant weighing 720 g was born. Placental pathology revealed a 3.5 g fetus mummified in one placenta, and a focus of extensive avascular villi, suggestive of fetal thrombotic vasculopathy, in the other placenta. Apgar scores of the surviving infant were 6 and 8 at 1 and 5 minutes, respectively. In the delivery room he required respiratory support with continuous positive airway pressure and was transferred to the neonatal intensive care unit (NICU). Physical examination revealed bilateral skin absence of the abdomen from below the thorax to the groins demarcated in a butterfly shape [Figure 1A and B]. The tissue over the abdomen was transparent and the internal organs were visible. No other skin lesion (e.g., scalp lesion) was noted. The chest was small and bell-shaped.

Fluid management, particularly during the initial 2 weeks of life, required extremely high fluid intake, up to 250 ml/kg/day. The highest sodium level reached 144 mEq/L, while the considerable water loss led to renal failure with renal creatinine up to 150 μmol/L and macrohematuria at the age of 1 week. The initial healing process of the abdominal wall, characterized by a fibrin layer [Figure 1C-F], enabled a gradual decrease in fluid requirements and by the second week of life renal function normalized.

Regarding respiratory function, he required 29 days of mechanical ventilation followed by oxygen supplementation for 3 months. The infant developed mild bronchopulmonary dysplasia (requiring 22% O_2 supplementation at 36 weeks post-gestational age) that was treated with diuretics, and he received one course of corticosteroids. He was diagnosed with a patent ductus arteriosus, which later closed spontaneously without treatment, and had no other cardiac anomaly.

His skin defect was treated conservatively with a non-adherent dressing, PolyMem™ and Telfa™. Gradually, a granulation tissue covered the lesion and new skin began to grow [Figure 1G and H]. During this process, a few open ulcers developed with occasional bleeding and the treatment consisted of conservative management only. A purulent discharge that appeared at age 3 months necessitated topical antibiotics. Wound cultures were positive for *Serratia marcescens* and *Pseudomonas aeruginosa*. Blood cultures at that point were sterile.

Closure of the skin defect with scar tissue led to a relatively small abdominal size and most probably increased abdominal pressure that precipitated the development of large bilateral inguinal hernia sacs containing a large amount of intestine [Figure 1E and G]. In order to prevent even higher

Figure 1. Congenital cutis aplasia in a preterm infant born after 29 weeks of gestation who weighed 720 g at birth. Pictures were taken immediately after birth **[A and B]**, at the age of 1 week **[C]**, at 2 weeks **[D]**, 1 month **[E]**, 2 months **[F]**, 3.5 months **[G]** and 6 months **[H]**. Pictures E and G illustrate the huge bilateral inguinal hernia sacs

abdominal pressure due to constipation, he was treated with lactulose to soften feces. At 4.5 months he underwent successful bilateral inguinal hernia repair with reduction of small intestine, cecum and appendix from the hernia sacs. This was followed by a second laparotomy due to intussusception and bowel obstruction that resulted in partial small bowl resection. Pathology workup revealed necrotic small intestine. During his hospitalization he was treated once for sepsis due to *Klebsiella pneumoniae* after the abdominal surgery.

Nutritional management included administration of hyper-alimentation immediately after birth with maximal protein (4 g/kg/day) and lipid (4 g/kg/day) intake, resulting in a gradual increase in his enteral nutrition. He reached full enteral nutrition at the age of 11 days, which enabled the removal of all intravenous lines that might increase the risk of infection in such an infant.

The infant's head ultrasound imaging following birth, during hospitalization and before discharge was normal, as was his neurologic assessment before discharge. Ophthalmic follow-up revealed retinopathy of prematurity at stage 1-2 in both eyes, requiring no intervention. He was discharged at the age of 5 months with no need for medications, weighing 3.37 kg at the corrected age of 49+2 weeks (below the 3rd percentile). At his last follow-up visit at the age of 11 months his developmental milestones were satisfying for his 8 months corrected age.

COMMENT

Most cases of aplasia cutis congenita with fetus papyraceus can be treated with simple non-adherent dressings and topical preparations. The prognosis is usually excellent with wound epithelialization and resolution [2,3]. In severe cases, surgical intervention may be needed; however, no major complications have been reported in term or near-term babies. ACC with FP has been reported previously in low birth weight newborns; one required a few days of ventilatory support and in both cases the

skin healed spontaneously [4,5]. To the best of our knowledge this report describes the smallest newborn with ACC and the first with extremely low birth weight. The main therapeutic challenges in this case included fluid management to overcome the risk of dehydration and electrolyte abnormalities in such a small infant, respiratory issues for his prematurity and the unique structure of his chest and abdomen, prevention of infections for the large area with absence of skin surface barrier, local treatment to enhance skin growth, optimal nutrition management to achieve overall optimal growth, and the reduced abdominal wall space, leading to huge bilateral inguinal hernia sacs requiring early surgical repair and small bowl resection due to bowel obstruction.

The etiology and pathogenesis of ACC is unknown and several theories have been suggested. In ACC associated with FP the most likely mechanism is fetofetal transfusion where acute transfusion from the surviving twin to the dying twin leads to volume depletion and hypotension resulting in ischemia of the skin in presumed vascular border zones. In this particular case, evidence of thrombosis was found in the surviving twin placenta, which could be related to passage of thrombogenic material to the viable twin. There was no other evidence in this case supporting a different etiology such as teratogenic exposure, infection or trauma.

In this era of modern reproductive medicine, with increasing incidence of multiple gestation pregnancies, the importance of fetus papyraceus is probably underappreciated and may be diagnosed in very or extremely low birth weight infants. This case demonstrates that under current advanced neonatal management and multidisciplinary care, successful treatment of extremely low birth weight infants with ACC is possible.

Correspondence

Dr. S. Eventov-Friedman

Dept. of Neonatology, Hadassah University Hospital, P.O. Box 12000, Jerusalem, 91120, Israel

Phone: (972-2) 677-9320

email: smadaref@hadassah.org.il

References

1. Frieden IJ. Aplasia cutis congenita: a clinical review and proposal for classification. *J Am Acad Dermatol* 1986; 14: 646-60.
2. Tempark T, Shwayder TA. Aplasia cutis congenita with fetus papyraceus: report and review of the literature. *Int J Dermatol* 2012; 51: 1419-26.
3. Morrow D, Schelonka R, Krol A, Davies M, Kuang A. Type V aplasia cutis congenita: case report, review of the literature, and proposed treatment algorithm. *Pediatr Dermatol* 2013; 30: e208-13.
4. Visva-Lingam S, Jana A, Murray H, John E. Preterm premature rupture of membranes associated with aplasia cutis congenita and fetus papyraceus. *Aust N Z J Obstet Gynaecol* 1996; 36: 90-1.
5. Wadams S, Garrett-Cox R, Kitteringham L. Aplasia cutis in association with a triplet pregnancy and fetus papyraceus. *Arch Dis Child Fetal Neonatal Ed* 2008; 93: F206.