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CASE REPORT

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# Sporadic aplasia cutis congenita

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**Abstract** Aplasia cutis congenita (ACC) is a rare group of disorders characterized by the focal absence of skin at birth. The majority of cases affect the scalp, but involvement of the trunk and extremities has been described. Proposed etiologies for ACC include infection, vascular malformations, amniogenesis, and teratogens, but no unifying theory has been identified. We present the case of a 1-day-old female with large, bilateral posterolateral trunk skin defects noted at birth. The prenatal history was significant for maternal diabetes, fetal papyraceus at 12 weeks' gestation, and a family history of limb defects. The infant was treated non-surgically with local wound care and antibiotics, as well as frequent dressing changes. The areas of absent skin developed a granulation-tissue layer followed by re-epithelialization and mild wound contracture. With early identification of the etiology of the lesions and appropriate investigation and treatment, including conservative wound management, aplastic lesions can heal successfully without affecting growth, but may require cosmetic repair at a later stage.

**Keywords** Aplasia cutis congenita · Fetus papyraceus

## Introduction

Aplasia cutis congenita (ACC) is a heterogeneous group of disorders characterized by well-circumscribed focal absence of epidermis, dermis, and occasionally subcutis at birth. These disorders are relatively rare, with approximately 114 cases reported in the medical literature since a classification system was proposed in 1985. The classification of ACC was based upon Frieden's analysis of 120 publications between 1917 and 1985 [1]. Since 1767, more than 500 cases have been described [1–3]. The majority of cases involve the vertex of the scalp overlying the sagittal sinus and may be associated with other congenital anomalies, while isolated trunk lesions are more rare.

Postulated etiologies of ACC include viral infections, vascular causes such as ischemic/thrombotic events or intrauterine hemangioma involution, amniotic adherence with subsequent skin trauma, and biomechanical forces during embryogenesis [4]. Autosomal dominant and recessive variants have been characterized. Additionally, teratogenic medications such as methimazole [1] and misoprostol [5] may be linked to the occurrence of ACC.

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## Case report

A 1-day-old female was transferred from an outside hospital for evaluation and treatment of skin defects noted at birth. She was born at 38 weeks' gestation by cesarean section for cephalopelvic disproportion to a 37-year-old gravida II, para II diabetic mother. Apgar scores were 8 and 9 at 1 and 5 min, respectively. Birth weight was 3,450 g, and there was no record of birth trauma. The pregnancy was complicated by maternal insulin-dependent diabetes mellitus, as well as the death of a twin fetus at 12 weeks' gestation. The mother was a 20-pack-year cigarette smoker with previously diet-controlled type II diabetes; she became insulin-dependent during the 8th week of gestation. Spotting was noted during the 8th week and a non-viable twin fetus was visualized by ultrasound (US) at 12 weeks' gestation. During the pregnancy, the mother was treated for group B streptococci, a sinus infection, and trichomoniasis. The perinatal history was negative for intrauterine trauma or the use of antithyroid medication or misoprostol.

Examination of the infant's trunk demonstrated a left-sided, full-thickness skin lesion measuring 11.5×3.5 cm (Fig. 1) and a similar right-sided lesion measuring 12.0×5.0 cm. There was complete absence of the epidermis and dermis at these sites, but the lesions were covered by a transparent dry membrane histologically

composed of a single squamocuboidal cell layer. Keratinization was not demonstrable upon cytokeratin staining. The remainder of the examination was unremarkable. There was no evidence of other congenital anomalies following detailed investigation.

Review of the patient's family history revealed a half-sibling (the father's son by a prior relationship) with situs inversus and caudal regression syndrome. A maternal cousin carried the diagnosis of cerebral palsy, but there was no family history of cutaneous or other limb abnormalities. The mother had one healthy teenaged son (also a half-sibling).

The patient was given IV ampicillin and gentamicin upon admission, and wet-to-dry normal saline dressings were initiated to the affected areas. A 12-lead electrocardiogram, an echocardiogram, and a thorough examination by a pediatric cardiologist elicited no cardiac abnormalities. Abdominal US was also normal. A detailed genetic evaluation revealed no evidence of a hereditary form of the disorder. The lesions were managed non-operatively with twice-daily dressing changes, initially normal saline, then silver sulfadiazine (SSD) cream. The patient was discharged home 6 days after admission with daily dressing changes. At 2 weeks, the wounds were noted to be contracting, and granulation tissue covered the defects. At 1 year follow-up, all of the affected areas were covered with a thin, atrophic layer of soft, pliable, keratinized epithelium. There has been some contraction of the defects with minimal distortion of the skin edges. At no time did the lesions become infected or develop erythema.

The lesions continue to be treated with a moisturizing cream and are covered with gauze to prevent chafing. Although further contracture and scarring is expected, she will likely undergo tissue expansion and scar revision at 3–5 years of age for primarily cosmetic reasons. To date, there has been no functional impairment, growth alteration, or limitation of movement. She has achieved normal milestones and development without limb spasticity, and there is no evidence of limb or digit abnormality. The risk of future cutaneous malignancy remains undetermined.

**Fig. 1.** Aplasia cutis congenita. Left posterolateral trunk lesion

## Discussion

ACC is classified into nine groups based on the body area affected, associated anomalies, and inheritance (Table 1). The scalp is the most common site (nearly 90%), with groups 1 to 3 limited to scalp lesions [6]. This case is classified to group 5, with associated fetus papyraceus (''mummified fetus'') or placental infarcts. Affected patients demonstrate multiple symmetric lesions, often with stellate or linear involvement of the scalp, chest, flanks, axillae, and extremities. Associated abnormalities of group 5 include a single umbilical artery (not seen in this case), developmental delay, spastic paralysis, nail dystrophy, clubbed hands and feet, and amniotic bands. Genetically, this is a sporadic event without risk of subsequent transmission.

**Table 1.** Classification for ACC*

| Group | Associated Anomalies | Inheritance |
|---|---|---|
| 1) Scalp ACC without multiple anomalies | Cleft lip and palate, tracheoesophageal fistula, patent ductus arteriosus, omphalocele, mental retardation, polycystic kidneys | Autosomal dominant or sporadic |
| 2) Scalp ACC with limb abnormalities | Limbs reduced, syndactyly, clubfoot, encephalocele, nail dystrophy or absence, persistent cutis marmorata | Autosomal dominant |
| 3) Scalp ACC with skin/organoid nevi | Epidermal nevi, organoid nevi, corneal opacities, scleral dermoids, eyelid colobomas, mental retardation, seizures | Sporadic |
| 4) ACC overlying embryologic malformations | Meningomyeloceles, spinal dysraphia, cranial stenosis, leptomeningeal angiomatosis, gastroschisis, congenital midline porencephaly, ectopia of ear, omphalocele | Depends upon underlying condition |
| 5) ACC with fetus papyraceus or placental infarcts | Single umbilical artery spastic developmental delay, spastic paralysis, clubbed hands and feet, amniotic bands | Sporadic |
| 6) ACC associated with epidermolysis bullosa | Blistering of skin and/or mucous membranes, deformed nails, pyloric or duodenal atresia, abnormal ears and nose, ureteral stenosis, renal anomalies, amniotic bands | Depends upon type of epidermolysis bullosa |
| 7) ACC localized to extremities without blistering | None | Autosomal Dominant or Recessive |
| 8) ACC caused by teratogens | Imperforate anus (methimazole), other signs of intrauterine infection with varicella or herpes simplex | Not inherited |
| 9) ACC associated with congenital syndromes | Trisomy 13, 4p-syndrome, ectodermal dysplasias, focal dermal hypoplasia, amniotic band disruption complex, XY gonadal dysgenesis, Johanson-Blizzard syndrome | Depends upon syndrome |

*Adapted from References 1 and 14

A review of 17 cases by Mannino et al. showed extensive trunk and limb involvement as well as association with fetus papyraceus [7]. A more recent article found elevated alpha-fetoprotein levels and a distinct amniotic fluid acetylcholinesterase band to be markers for ACC [8]. Group 5 also appears to be related to fetal demise in the late first or early second trimester, when disseminated intravascular coagulation may cause selective hypoperfusion of mesodermal tissue of the skin and trunk [9]. Because of these variations, a thorough history and physical examination must be completed with emphasis on cutaneous and limb defects, complications and treatments during pregnancy, and careful examination of the placenta. This child had the following associations: a family history of limb defects (caudal regression syndrome in her half-brother), an associated fetus papyraceus, and maternal diabetes.

Therapeutic options vary with the site of the lesion and the patient's condition. Wound treatment in cases of superficial ulceration is conservative. Extensive and deep lesions require reconstruction or bone and skin grafting. Urgent surgical intervention is recommended for extensive defects overlying the sagittal sinus to prevent potentially lethal infections and hemorrhage [10–12]. Superficial lesions left untreated heal slowly by re-epithelialization and leave a hypertrophic or atrophic scar. Grafting [13], the use of biological dressings [14], and SSD antibiotic dressings while awaiting skin and bony ingrowth [15] have been reported with variable success. Large areas of scarring may require tissue expansion and skin flaps or grafts.

ACC is a relatively rare condition requiring early diagnosis and treatment. A detailed family history as well as a careful physical examination and close developmental follow-up should be conducted. Conservative management, as in this case, can aid in successful healing of aplastic lesions and help to control costs.

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