

## Case Report

## Aplasia Cutis Congenita in a Newborn of Diabetic Mother: A Case Report and Review of Literature

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**Key Words:**Aplasia cutis congenita,  
Newborn, Skin diseases**ABSTRACT****Background:** Aplasia cutis congenita (ACC) is a rare condition regarded as a congenital absence of the epidermis, dermis, and in some cases, subcutaneous tissues in the newborn. The pathogenic mechanism is unclear, although the condition has been described as a result of the disrupted development or degeneration of skin in utero. ACC may be observed with fetus papyraceous (FP).**Case Presentation:** We report a case of an 8-hour-old newborn female with bilateral symmetrically distributed, stellate type of truncal ACC at birth. She was the survivor twin as the other fetus died at 13wk+3d gestation. This condition describes ACC with FP. Physical examination showed otherwise normal and managed with no other congenital abnormalities. The newborn was treated with antibacterial ointment and antibiotics, and lesions resolved spontaneously within 5 days, leaving scars.**Conclusions:** This report explained a newborn with type V cutis aplasia congenita in whom the detection was approved based on the revision of antenatal history and clinical features. The protocol outcome revealed that the topical and systemic antibiotic and washing with normal saline could be an effective treatment for the healing of ACC lesions. Follow-up after 3 months indicated that the skin lesion completely healed, leaving a very small atrophic scar, and no further lesion management was required.

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## Background

**A**plasia cutis congenita (ACC) is a rare and uncommon congenital abnormality involving various skin layers, mostly as a single lesion involving the midline vertex and, less commonly, the underlying bone and periosteum [1, 2]. The lesions can also be found in the limbs, abdomen, or chest. In 84% of cases, the defect is observed in the scalp; however, the lesion can occur anywhere in the body. Most lesions in the head area are isolated with a size of 0.5-10 cm, and sometimes multiple lesions may be seen. The lesions are round, oval, or jagged, but non-scalp lesions are usually bilaterally symmetric. Individual lesions are often benign but may be associated with other disorders and malformations. The form of the wound can vary from superficial erosion to a deep lesion, with the affected area covered with a transparent, thin membrane [2, 3]. ACC is usually a solitary clinical finding, but it may also happen in several genetic syndromes, including Bart syndrome, Adams-Oliver syndrome, Setleis syndrome, and Patau syndrome [4]. Aplasia cutis is 1 to 3 in 10000 live births [5]. Most presented cases are sporadic, with a few reports of familial occurrence in the form of autosomal dominant and autosomal recessive [6].

Histological evaluation of the wound reveals the lack of normal skin structures, such as sweat glands, sebaceous glands, or hair follicles, with the dermis devoid of collagen fibers [7]. The National Organization for Rare Disorders (NORD) has defined other names for ACC as a congenital lesion of the newborn, scalp defect congenita, and congenital defect of the scalp and skull [8]. Cordon, for the first time, described ACC in 1767. Neither the etiology nor pathogenesis of ACC has been explained; however, genetic, environmental, and exogenous causes have been signified as potential factors. These factors include vascular blood supply, poor blood supply to the skin, fetal and placental ischemia, vascular compromise, placental infarcts, intra-uterine infections, adhesion of the amniotic membrane to fetal skin, amniogenesis, amniotic rupture sequence, syphilis, teratogenic substances, trauma, a sudden arrest of midline embryological development, failure in neural tube closure, ectodermal dysplasia, and maternal intrapartum drug use, such as methimazole, carbimazole, misoprostol, and valproic acid [9-11]. Studies show that the non-scalp ACC is usually large and can be associated with epidermolysis bullosa (EB) [3]. The association of ACC with EB could indicate other congenital anomalies such as renal abnormalities, pyloric or duodenal atresia, craniofacial abnormalities, ureteral stenosis, and nail dystrophy [12].

The diagnosis of ACC is based on clinical findings with a few reports on histopathology depending upon the depth of the defect, which shows a lack of epidermis, dermis, adnexa, and sometimes subcutaneous tissue. An increase in acetylcholinesterase and  $\alpha$ -fetoprotein has also been reported in the amniotic fluid of mothers with ACC fetuses [13]. Regular ultrasonography during prenatal care sometimes contributes to the early diagnosis of ACC. The normal fetal skin generates strong echoes on ultrasound, while in ACC, such echoes are absent [14]. ACC can be associated with fetus papyraceous (FP) and occurs as an isolated defect or with other associated anomalies [2]. It has a bilaterally symmetrical pattern in buttocks, truncal, and thigh lesions, which is related to fetal death in the late first to early second trimester of pregnancy [15]. ACC associated with FP showing symmetrical circumferential scarring encircling the trunk has been less commonly described.

## Differential diagnosis

ACC may be associated with congenital anomalies of the heart, gastrointestinal tract, urinary tract, genital tract (e.g. gastroschisis or omphalocele), and central nervous system (e.g. myelomeningocele or spinal dysraphism). Also, a small percentage of lesions will heal in utero, showing as an atrophic hairless scar, which can be mistaken for an epidermal nevus. Although ACC is often detectable with an exact clinical exam, lesions with similar presentations should be noticed and ruled out. Ultrasound and further workup with MRI can help to make this diagnosis [16].

Herein, we report an 8-hour-old girl with symmetrical circumferential scarring encircling the trunk associated with fetus papyraceus. In this study, one case of non-scalp ACC occurring in the trunk is reported, and a brief literature review is discussed.

## Case Presentation

### Anamnesis and status before treatment

An 8-hour-old girl was admitted to the Neonate Unit of Imam Ali (AS) Hospital in Amol City, Iran. She had a skin lesion on her trunk (flanks), a bilaterally symmetrical, stellate type of truncal aplasia cutis congenita. A relatively broad lesion on the right and left sides of the baby with a width of 3x3 cm and depth of 2 to 3 mm and a clear margin was distinguished, which developed a secondary infection during hospitalization (Figure 1). The patient was born full term (38 wk+4 d) by cesarean section. The baby's Apgar score increased from 9 at 1

**Table 1.** Frieden's classification of Aplasia cutis congenita

1. ACC of the scalp without other abnormalities
2. ACC of the scalp with limb anomalies: Adams–Oliver syndrome, hypoplasia or aplasia of the distal phalanges, nipple and hair abnormalities, fibromas, vascular malformations
3. ACC of the scalp along with epidermal nevi, ophthalmic, and neurological abnormalities
4. ACC with embryologic deformities: leptomenigeal angiomatosis, omphalocele, meningomyelocele, cranial stenosis, pencephaly, spinal dysraphism, or gastroschisis
5. ACC of the trunk or limbs associated with fetus papyraceous, placental infarction
6. ACC and epidermolysis bullosa (butterfly disease) involving the lower extremities
7. ACC of the extremities without epidermolysis bullosa
8. ACC is caused by teratogens such as intrauterine infections by herpes simplex and varicella-zoster virus, and using drugs during pregnancy such as carbimazole or methimazole
9. ACC accompanied with congenital malformations includes Trisomy 13 (Patau syndrome), Wolf–Hirschhorn syndrome (deletion of the short arm of chromosome 4), XY gonadal dysgenesis, Setleis syndrome, Johanson–Blizzard syndrome, and other abnormalities

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minute to 10 after 5 minutes. The anthropometric parameters of the neonate were as follows: Weight, 3000 g; height, 52 cm; and head circumference, 35 cm. The baby was the family's second child, and her parents were not next of kin. There was no positive family history of ACC.

The baby was the survivor of a twin as the other fetus died at a gestational age of 13 weeks and 3 days. The death of an identical twin complicated the pregnancy. This condition indicates congenital aplasia cutis with FP. On physical evaluation, the neonate was alert, well, and otherwise normal. Hemangiomas or other congenital defects were not observed in her body. The infant's vital signs were normal (temperature 36.9°C, heart rate 128 beats per minute, and respiratory rate 48 breaths per minute). The rest of the examination was unremarkable. In particular, there was no evidence of other congenital abnormalities. All blood tests and urinalysis were normal. The blood group of the baby was A+. Culture and smearing of the wound site were performed, and the result was negative. Also, full abdominal and pelvis ultrasound (the liver, gallbladder, pancreas, bile ducts, spleen, kidneys, bladder, uterus, and abdominal aorta) showed normal results. Internal medicine and cardiac consultations were performed, and no abnormal results were reported. A skin consultation was performed.

The mother was 31 years old without a history of infectious diseases or trauma during pregnancy but with a history of drug intake (thyroid and diabetes drugs) during pregnancy. Regarding the baby's mother's history, she suffered from gestational diabetes and received insulin from 16 weeks of pregnancy. The weight and

height of the mother, who was a housewife, were 92 kg and 164 cm, respectively.

### Conservative treatment strategy

#### Antibiotics, infection control, and medical considerations

Conservative treatment was insufficient to ensure survival. The neonate received an intravenous infusion of vancomycin (30 mg in dextrose 10% in water), amikacin (30 mg) for 5 days, as well as 15 g of 2% mupirocin ointment topically two times per day (BID). The wound was washed with a normal saline solution, and the dressing was done with sterile gauze as BID. After 5 days, the skin lesion gradually decreased in size and dried up within 6 days of admission. On the other hand, it was relatively improved with systemic and local antibiotic treatment. Finally, she was discharged after healing the wound as an atrophic area with the following order: Daily wound washing with normal saline solution, topical ointment, dressing with sterile gauze, and consumption of cephalixin syrup (Figure 2).

#### Follow-up

Follow-up after 3 months indicated that the skin lesion completely healed, leaving a very small atrophic scar, and no further lesion management was required.

### Discussion

ACC is a disease characterized by a complete or partial, localized, or widespread absence or scarcity of skin at birth. It can involve large areas of the face, buttocks, and



**Figure 1.** Aplasia cutis distributed over the left and right sides of the abdomen

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trunk as an isolated condition or may be accompanied by other genetic syndromes [1]. The exact mechanism of ACC development is still poorly understood [2]. Mohaddes et al. reported that not all types of these lesions could be justified with one theory because they are phenotypes of different diseases, and more than one mechanism is involved in their occurrence [17].

ACC associated with FP has been reported in numerous studies. In the present case report, the death of a twin occurred at 13 wk+3 d of gestation. Tempark et al. reported that this rare condition is a congenital skin defect and intrauterine death of a fetus with or without a stillborn fetus pressed flat by the growing twin (fetus

papyraceous) at birth. There was a reproducible and distinctive distribution pattern of bilateral symmetrical truncal, thigh, and buttocks lesions related to fetal death in the late first to early second trimester of pregnancy [15]. Although the mechanisms of ACC associated with FP are not clear, vascular ischemia is strongly suggested and could explain the skin's bilateral and symmetrical congenital aplasia [18]. Hypotension and hypovolemia may lead to ischemia of the skin and other organs of the viable twin. Therefore, preventing and treating complications should be considered [19]. Therefore, attention should be paid to the prevention and treatment of complications.



**Figure 2.** Aplasia cutis congenita distributed over the left and right sides of the abdomen at 6 days of admission (discharging time)

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Aplasia lesions are classified into 9 groups according to location, lack of skin, and accompanying anomalies [17]. On the other hand, different classifications based on presentation and or etiologies have been proposed so far. Table 1 presents Frieden's classification of aplasia cutis congenital [2]. According to Frieden's classification of aplasia cutis congenital, the case reported in this research belongs to group 5.

ACC management is related to its pattern, underlying causes, location, and abnormalities [20]. However, according to the literature, most cases of FP-related ACC can be cured with conservative treatment [15, 21, 22]. The lesions can also heal in utero, with scars observed at birth [17]. Small localized lesions can be managed conservatively, while more extensive ones may require surgery [18]. Small areas of aplasia cutis usually heal over time, leaving a hairless scar. Gentle cleaning and special ointments can be used to prevent infection. Antibiotics can be useful in the event of infection, which was also observed in our case. Larger lesions or multiple scalp defects may need surgical repair, and sometimes a skin or bone graft may be needed; moreover, texture improvers could be used [23]. Therefore, ACC's treatment mode can differ depending on the infant's condition. Although the most popular regimen is conservative therapy, reports of surgical treatment of ACC rarely show the use of the scalp as the donor site [15]. Managing ACC with extensive skin defects is problematic because the best treatment has yet to be determined. Skin regeneration with and without grafts and conservative treatments have been performed with different outcomes [22, 24]. Duan et al. showed that using the scalp as the skin donor site for grafting is an effective therapy for large and deep ACC lesions that arise at sites other than the scalp [19]. Also, another study indicated two types of treatment: A conservative approach involving daily antiseptic dressings to allow for epithelialization of the scalp, improving conditions for secondary surgery, and closing the defect with local rotary flaps [25]. Abulezz et al. revealed that healing and recovery are spontaneous in most cases and that no special treatment is needed apart from keeping the wound clean [6]. Magliah et al., in their report of a 45-day-old baby with a flat scalp lesion on the anterior fontanel with a well-defined complex cyst, showed that intravenous infusion of vancomycin (67 mg in dextrose 5% in water) for 3 days did not improve scalp cysts. However, 15 g of 2% mupirocin ointment topically 3 times per day caused a gradual reduction in the size of the head cyst after 2 days, which dried up within 1 week [20]. A limitation of this study is the lack of genetic evaluation.

## Conclusion

We describe an 8-hour-old girl with ACC encircling the trunk in a bilateral symmetrical form. The baby was the survivor of a twin pregnancy; the co-twin died 13 wk+3 d of gestation. To our knowledge, ACC associated with FP showing symmetrical circumferential scarring encircling the trunk has not been previously explained. Also, there is no consensus or guideline for the treatment strategy of ACC, although multiple therapeutic regimens are currently available. The decision between conservative and surgical management depends on the location and size of the lesion. The prompt and effective management of ACC is crucial for preventing fetal complications.

## Key clinical message

ACC is a rare disease described by the absence of skin layers, mostly a lone lesion involving the midline vertex, but the whole body can be applied. We present aplasia of a baby born of a diabetic mother taking diabetes drugs (insulin).

## Ethical Considerations

### Compliance with ethical guidelines

This case study was approved by the Ethics Committee of Babol University of Medical Sciences (Code IR.MUBABOL.REC.1401.161) on February 7, 2023. Permission has been obtained from the neonate's parents to publish the present case report and any photos of the child.

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### Authors contributions

Neonatal clinical management: Alireza Yahyaei Shandasthi; Literature review and writing the original manuscript: Hajar Pasha and Fatemeh Haghshenas; Review and editing: Hajar Pasha, Alireza Yahyaei Shandasthi, and Amir Bahari Bandari; Final approval: All authors.

### Conflicts of interest

The authors declared no conflict of interest.

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