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Bilateral Patellar Aplasia Cutis Congenita: Clinical Management and Reconstructive Considerations—a Case Report

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Case Report

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Abstract

Background: Aplasia cutis congenita (ACC) is a rare congenital disorder characterized by localized absence of the skin, most commonly affecting the scalp. Bilateral involvement of the patellar region is exceptionally uncommon. We report an isolated case of bilateral patellar ACC in the surviving twin following a monochorionic diamniotic twin pregnancy complicated by selective fetoscopic laser photocoagulation and discuss the successful outcome of conservative management.

Case Presentation: A female neonate was delivered by cesarean section at 38 weeks and 4 days of gestation after a monochorionic diamniotic twin pregnancy in which selective fetoscopic laser photocoagulation with termination of the affected co-twin had been performed at 13 weeks and 4 days of gestation because of multiple severe congenital anomalies. At birth, asymmetric bilateral patellar skin defects were identified. The left knee demonstrated an approximately 3 × 4 cm full-thickness cutaneous defect with exposed patellar periosteum, whereas the right knee exhibited a superficial epidermal defect. No associated congenital anomalies were identified, and the diagnosis of ACC was established clinically following multidisciplinary evaluation. Given the clean, well-vascularized wound bed, absence of exposed joint structures or infection, and preserved tissue viability, conservative treatment consisting of daily non-compressive dressings, topical bacitracin–neomycin, and prophylactic intravenous ampicillin–sulbactam was initiated. Progressive epithelialization was achieved without complications, and complete wound healing with preserved knee function was observed without surgical intervention.

Conclusions: This case demonstrates that selected periarticular ACC lesions, even in the presence of exposed patellar periosteum, can be successfully managed with conservative treatment when careful multidisciplinary assessment confirms favorable wound characteristics. In addition, this report expands the limited literature on bilateral patellar ACC occurring in the setting of a complicated monochorionic diamniotic twin pregnancy.

LEVEL OF EVIDENCE: IV

INTRODUCTION

Aplasia cutis congenita (ACC) is a rare congenital disorder characterized by localized or widespread absence of the epidermis and dermis, with a heterogeneous clinical presentation.[1] Although ACC can occur in any anatomical location, approximately 80% of cases involve the scalp, particularly the vertex and midline region, whereas the lower extremities and trunk account for approximately 25% and 12% of cases, respectively.[2, 3] The estimated incidence of ACC ranges from 0.5 to 3 per 100,000 live births, whereas isolated involvement of the extremities is exceedingly rare.[4]

Although its exact etiology remains unclear, multiple biological, pharmacological, mechanical, and genetic factors have been implicated in its pathogenesis.[5] In certain cases, ACC has been associated with placental infarction or the intrauterine demise of a co-twin, suggesting that vascular disruption may

contribute to lesion development.[2, 6, 7] Several clinical subtypes have been described based on the location of the skin defect, clinical presentation, associated congenital anomalies, and pattern of inheritance.[8] In some patients, ACC may represent a cutaneous manifestation of underlying embryological malformations, chromosomal abnormalities, or ectodermal dysplasias. Associations with inherited disorders such as epidermolysis bullosa, intrauterine infections, and exposure to specific teratogenic agents during pregnancy have also been reported.[9] Extremity involvement, particularly isolated bilateral periarticular involvement, represents one of the rarest clinical presentations of ACC and poses unique diagnostic and therapeutic challenges.[3]

The diagnosis of ACC is established primarily by clinical examination immediately after birth. Approximately 75% of cases present as an isolated lesion.[7] The defect is typically solitary, oval-shaped, well-demarcated, and non-inflammatory, and may manifest as an ulcer of varying depth, granulation tissue, a parchment-like membranous lesion, or an atrophic alopecic scar.[2, 6]

The management of ACC depends on the size and location of the defect as well as the involvement of underlying structures. Early treatment is essential to prevent potentially serious complications, including hemorrhage, infection, fluid and electrolyte imbalance, and excessive heat loss, depending on the anatomical location and severity of the lesion.[6, 10] To the best of our knowledge, isolated bilateral patellar ACC occurring in the surviving twin following selective fetoscopic laser photocoagulation for a monochorionic diamniotic twin pregnancy has not previously been reported. We therefore present this exceptionally rare case and highlight the successful conservative management of a full-thickness periarticular defect with exposed patellar periosteum, resulting in complete healing and preservation of knee function without surgical intervention.

CASE REPORT

We report the case of a female newborn delivered by cesarean section at 38 weeks and 4 days of gestation following a monochorionic diamniotic twin pregnancy. The mother was a 28-year-old primigravida, and the parents were non-consanguineous. During the first trimester, one twin was diagnosed with multiple severe congenital anomalies and underwent selective fetoscopic laser photocoagulation with termination at 13 weeks and 4 days of gestation. The remainder of the pregnancy was uneventful.

The mother had no chronic systemic disease or exposure to alcohol, illicit drugs, radiation, or known teratogens during pregnancy. She had intermittently used flunarizine for migraine before conception but discontinued it during pregnancy. Periconceptional folic acid supplementation was followed by prenatal multivitamins, omega-3, and vitamin D, while vaginal progesterone (500 mg/day) was administered until 32 weeks of gestation. She smoked approximately five cigarettes daily throughout pregnancy. The father was a heavy smoker (2–3 packs/day) with occasional alcohol consumption. There was no family history of ACC, epidermolysis bullosa, or inherited skin disorders, although maternal relatives had congenital heart disease and paternal relatives had multiple gestations and isolated congenital limb anomalies.

Apgar scores were 7, 8, and 9 at 1, 5, and 10 minutes. Birth weight was 2850 g, length 47 cm, and head circumference 35 cm, all appropriate for gestational age.

Physical examination revealed bilateral asymmetric congenital skin defects involving the patellar regions. The right knee showed a superficial epidermal defect, whereas the left knee had an approximately 3 × 4 cm full-thickness skin and subcutaneous tissue defect with a clean, well-vascularized wound bed. The patellar periosteum was exposed without involvement of the joint capsule, tendon, or bone. The defect represented approximately 0.3% of the total body surface area according to the Lund and Browder chart. Mild localized soft tissue contracture was present over the left suprapatellar region. No abnormalities of the scalp, nails, extremities, craniofacial structures, or abdominal wall were identified. Neurological examination, peripheral pulses, systemic examination, and gross placental evaluation were unremarkable.

The patient was admitted to the neonatal intensive care unit and evaluated by neonatology, plastic and reconstructive surgery, and dermatology teams. The diagnosis of ACC was established clinically based on the characteristic appearance of the lesions; skin biopsy was not performed because histopathological confirmation was not expected to influence management.

The differential diagnosis included epidermolysis bullosa, amniotic band sequence, congenital erosive and vesicular dermatosis, birth trauma, and Adams–Oliver syndrome. These conditions were excluded based on the absence of characteristic clinical findings, including skin fragility or blistering, constriction rings, distal limb anomalies, scalp defects, cardiovascular malformations, and delivery-related trauma. The lesions were therefore considered consistent with isolated bilateral patellar ACC.

Conservative treatment was selected because of the limited defect size, viable wound bed, absence of exposed joint capsule, tendon or bone, sterile wound cultures, and the high regenerative potential of neonatal skin. Prophylactic intravenous ampicillin–sulbactam (50 mg/kg every 8 hours) was administered during hospitalization. Daily wound care consisted of paraffin gauze (Bactigras®) covered with topical bacitracin–neomycin cream, applied loosely to maintain tissue perfusion while protecting the wound. The patient was closely monitored for infection, tissue viability, and progressive epithelialization. Figure 1 demonstrates the left knee lesion on the first postnatal day, showing a full-thickness soft tissue defect with a clean, well-vascularized wound bed and exposed patellar periosteum.

Under continued conservative management, progressive epithelialization of both lesions was achieved. As no local or systemic infection developed and the clinical course remained favorable, surgical intervention was not required. The patient was discharged from the neonatal intensive care unit on the sixth postnatal day with instructions for continued outpatient dressing care. The appearance of the lesions at the time of discharge is shown in Fig. 2.

Complete epithelialization was achieved without hypertrophic scar formation, recurrent ulceration, joint contracture, or limitation of knee motion. At the latest follow-up examination, the patient demonstrated full range of motion of both knees, normal lower-extremity function, and age-appropriate growth and

developmental milestones without functional impairment. Because no additional congenital anomalies or dysmorphic features were identified, further genetic investigations were not considered clinically indicated. The appearance of the patient at 1 month follow-up is shown in Fig. 3. Written informed consent for publication of this case report and the accompanying images was obtained from the patient's parents.

This case report received no specific funding or financial support from any public, commercial, or non-profit organization. As this manuscript presents a single case report and does not involve a clinical trial, a clinical trial registration number is not applicable.

DISCUSSION

ACC is a rare congenital disorder characterized by localized absence of the skin.[11] Although its exact pathogenesis remains incompletely understood, it is thought to result from abnormal ectodermal development caused by mechanical, vascular, hereditary, or teratogenic factors.[12] Proposed etiologies include intrauterine exposure to teratogens such as alcohol, cocaine, marijuana, heroin, misoprostol, methimazole, and carbimazole, congenital infections including herpes simplex and varicella-zoster viruses, impaired fetal skin perfusion, amniotic band disruption, intrauterine trauma, and chromosomal abnormalities, particularly trisomy 13.[13]

The present case is unique because it combines three uncommon features: isolated bilateral patellar involvement, occurrence in the surviving twin after selective fetoscopic laser photocoagulation in a monochorionic diamniotic pregnancy, and successful conservative treatment despite exposed patellar periosteum. Although a causal relationship cannot be established, this case expands the recognized clinical spectrum of ACC and supports individualized management based on wound characteristics rather than periosteal exposure alone.

The temporal association with selective fetoscopic laser photocoagulation raises the possibility of vascular or mechanical mechanisms contributing to lesion development. Altered placental perfusion following laser photocoagulation could theoretically produce transient regional ischemia during a critical stage of fetal skin development. In addition, the retained demised co-twin may have exerted localized mechanical compression on the surviving fetus, particularly over the lower extremities, potentially impairing cutaneous development. However, these mechanisms remain speculative, and additional clinical reports are required to determine whether this association represents a true pathogenic mechanism or a coincidental finding.

ACC has been reported in association with numerous congenital anomalies involving multiple organ systems, including central nervous system abnormalities, cardiovascular malformations, gastrointestinal anomalies, epidermolysis bullosa, limb defects, renal anomalies, and cutis marmorata.[2, 4] Consequently, every patient with ACC should undergo careful clinical evaluation to exclude associated systemic abnormalities.

Frieden proposed the most widely accepted classification system, dividing ACC into nine subgroups according to lesion location, associated anomalies, and inheritance pattern. Our patient was classified as Group 7, which includes ACC localized to the extremities without associated epidermolysis bullosa or blistering.[8] The absence of additional congenital anomalies or hereditary skin disorders further supports this classification. Table 1 summarizes Frieden's classification of ACC.

Although prenatal ultrasonography may occasionally suggest the diagnosis, ACC is usually recognized during the initial neonatal examination.[14] Skin biopsy is reserved for diagnostically uncertain cases because histopathological findings are largely nonspecific, typically demonstrating absent or markedly thinned epidermis, dermal thinning with loosely arranged collagen bundles, dermal atrophy, reduced skin appendages, and thinning of the subcutaneous tissue.[2] Histopathological evaluation is particularly useful when differentiating ACC from conditions such as nevus sebaceus, traumatic scar, congenital dermoid cyst, cutaneous meningioma, or heterotopic brain tissue, or when surgical treatment is planned.

In the present case, epidermolysis bullosa–associated ACC variants, including EB type VI and Bart syndrome, were excluded by the absence of skin fragility, blistering, nail dystrophy, and mucosal involvement during both hospitalization and follow-up.[15] Likewise, Adams–Oliver syndrome was considered unlikely because scalp defects, transverse limb anomalies, nail abnormalities, and central nervous system malformations were absent.[16] Despite the history of monochorionic diamniotic twin pregnancy complicated by selective fetoscopic laser photocoagulation, no clinical evidence suggested a syndromic form of ACC or another inherited skin disorder.

Although syndromic associations were excluded, ACC may still be associated with substantial morbidity and mortality depending on lesion size, location, and associated anomalies. Approximately 70–90% of lesions occur on the scalp, and calvarial defects are present in nearly one-third of these patients. Extensive scalp lesions may lead to life-threatening complications, including hemorrhage, sagittal sinus thrombosis, and central nervous system infections, with mortality rates reported as high as 50% in selected series.[2, 6] Therefore, management should include not only local wound care but also close surveillance for systemic complications, together with meticulous neonatal supportive care, including infection control, appropriate antibiotic therapy when indicated, fluid and electrolyte management, nutritional support, prevention of heat loss, and adequate hydration.

There is currently no universally accepted treatment algorithm for ACC.[17] Management is primarily determined by defect size and the presence of underlying osseous involvement.[10] In general, lesions smaller than 2 cm^2 without bony defects are managed conservatively with secondary intention healing.[6] Owing to the remarkable regenerative capacity of neonatal skin, satisfactory healing is often achieved while avoiding anesthesia and surgical morbidity. Primary closure following excision may be considered for selected defects measuring approximately 2–4 cm or in cases with limited bone exposure.[6, 18]

In contrast, defects larger than 4 cm or those associated with significant bony abnormalities generally require surgical reconstruction because secondary healing may increase the risk of complications,

prolonged hospitalization, and poor cosmetic outcomes.[6] Available reconstructive options include split- or full-thickness skin grafts, dermal substitutes, local or regional flaps, and tissue expansion techniques.[18, 19] Multidisciplinary assessment involving plastic and orthopedic surgeons is particularly important for periarticular lesions.[20] Regardless of treatment strategy, potential complications include wound infection, hypertrophic scar formation, keloids, and scarring alopecia.

In our patient, despite exposure of the patellar periosteum, conservative treatment was chosen because the wound was well vascularized, the joint capsule and extensor mechanism remained intact, and no evidence of infection was present. Daily wound care resulted in complete epithelialization with preservation of knee function, avoiding neonatal reconstructive surgery. This favorable outcome suggests that carefully selected periarticular ACC lesions with exposed periosteum may heal successfully with meticulous conservative management when deeper structures remain intact.

This report has several limitations. As a single case report, no causal relationship can be established between selective fetoscopic laser photocoagulation and the development of ACC. Furthermore, genetic testing was not performed because no clinical findings suggested an inherited syndrome. Additional case reports and multicenter studies are needed to clarify the potential relationship between monochorionic twin interventions and ACC.

Abbreviations

ACC: Aplasia cutis congenita., EB = epidermolysis bullosa.

Declarations

Ethics approval and consent to participate

This case report was conducted in accordance with the ethical principles of the Declaration of Helsinki. Ethics committee approval was not required for this single case report.

Consent for publication

Written informed consent for publication of the patient’s clinical details and clinical photographs was obtained from the patient’s parents/legal guardians.

Availability of data and materials

All data supporting the findings of this case report are contained within the manuscript. Additional information related to the case is available from the corresponding author on reasonable request, subject to patient confidentiality and applicable privacy regulations.

Competing Interests

The authors declare that they have no conflicts of interest to disclose concerning the research, authorship, and publication of this article.

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Authors' contributions

A.Ö. designed the case report, conducted the literature review, and wrote the main manuscript text. Y.N.E. and H.T.A. evaluated and followed the patient and contributed to the clinical assessment and interpretation of the findings. H.T.A. supervised the clinical management and the preparation of the manuscript. All authors contributed substantially to the work, reviewed and critically revised the manuscript, approved the final version, and agreed to be accountable for all aspects of the work.

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Additional Statement

This manuscript has not been presented at any scientific meeting or conference and is not under consideration for publication elsewhere. All data supporting the findings of this case report are included in the manuscript. Additional case-related information is available from the corresponding author on reasonable request, subject to patient confidentiality and applicable privacy regulations.

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Table

TABLE 1: Classification proposed by Frieden in 1986 for the types of ACC.

Category	Body Area Impaired	Associated Changes	Heritage
Group 1 ACC on scalp, without multiple anomalies	Scalp, usually at the apex	Cleft lip and palate, tracheoesophageal fistula, patent ductus arteriosus, omphalocele, polycystic kidneys, mental retardation, cutis marmorata telangiectatica congenita	Autosomal dominant or sporadic
Group 2 ACC on the scalp, associated with limb malformations	Midline of the scalp	Limb reduction anomalies, syndactyly, concave foot, dystrophy or absence of nails, dermal appendages on the fingers, persistent cutis marmorata, encephalocele, hemangioma, cardiac disease, cryptorchidism, and postaxial polydactyly	Autosomal dominant
Group 3 ACC associated with epidermal or organoid nevus	Scalp can be asymmetrical	Corneal opacities, scleral dermoids, eyelid colobomas, psychomotor retardation, and seizures	Sporadic
Group 4 ACC involving areas of embryological malformations	Abdomen, lumbar skin, scalp, or any area	Meningomyelocele, spinal dysraphism, cranial stenosis, midline congenital porencephaly, leptomeningeal angiomatosis, ear ectopy, omphalocele, and gastroschisis	Dependent on baseline conditions
Group 5 ACC associated with papyraceous fetus or placental infarction	Multiple and symmetrical areas, often linear or stellate, on the scalp, thorax, flanks, underarms, and extremities	Single umbilical artery, developmental delay, spastic paralysis, nail dystrophy, and amniotic bands	Sporadic
Group 6 ACC associated with EB, usually localized without multiple congenital anomalies	Extremities	Formation of skin and/or mucous membrane blistering, deformed or missing nails, metatarsus varus, congenital absence of kidney (observed in recessive dystrophic, dominant dystrophic, and simplex EB)	Dependent on the type of EB; autosomal dominant or recessive
Group 7 ACC localized on the extremities, without blistering	Pretibial areas, back of the hands and feet, and wrist extensors	None	Autosomal dominant or recessive
Group 8 ACC caused by specific teratogenic substances	Scalp (with methimazole); any area (herpes simplex/herpes zoster infection)	Imperforate anus (methimazole); signs of intrauterine herpes simplex or herpes zoster infection	Not hereditary

Group 9 ACC associated with genetic malformation syndromes	Scalp or any area	Trisomy 13, 4p syndrome, ectodermal dysplasias, Johnson–Blizzard syndrome, focal dermal hypoplasia, and XY gonadal dysgenesis	Diverse and dependent on each specific syndrome

Abbreviations: ACC = aplasia cutis congenita; EB = epidermolysis bullosa.

Figures



Figure 1

Clinical appearance of the left patellar lesion on the first postnatal day demonstrating a full-thickness cutaneous defect with exposed viable patellar periosteum and a clean, well-vascularized wound bed.



Figure 2

Clinical photograph obtained before hospital discharge (postnatal day 6) demonstrating progressive epithelialization of the bilateral patellar lesions without evidence of infection or wound deterioration following conservative wound management.



Figure 3

Clinical appearance at the 1-month follow-up demonstrating complete epithelialization of the bilateral patellar lesions with satisfactory scar formation following conservative treatment.