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APLASIA CUTIS CONGENITA: A RARE CONGENITAL ABSENCE OF SKIN

Dermatology

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ABSTRACT

Aplasia Cutis Congenita is a rare developmental anomaly characterized by the congenital absence of skin, commonly affecting the scalp, can also involve the trunk and limbs. The lesion may range from superficial absence of epidermis to full-thickness defects exposing underlying structures such as bone or dura. Several genes have been implicated depending on the type and associated syndrome. Isolated Aplasia Cutis Congenita is associated with BMS1, KCTD1 or TERT variants which mainly affect skin morphogenesis and follicular development. One day old female baby, born via caesarian section at term gestation presented with absence of skin over both legs and feet since birth which was noted post baby wrapping. No history of oozing from the site of lesions, fever or associated systemic illness in the neonate. No history of infection, trauma, teratogenic drug intake during pregnancy or significant comorbidities in mother. The neonate was managed with wet sterile gauze covering over the lesions and topical treatment with fusidic acid cream in the morning and night. Collagen dressing was done on alternate days. Few well demarcated erythematous areas of absent skin exposing subcutaneous tissue present over the anterior aspect of both legs and dorsal aspect of feet, with the base appearing smooth showing areas of thin epithelial covering and surrounding shiny, atrophic skin. Histopathology was not performed for ethical concerns. Aplasia Cutis Congenita can occur as an isolated defect or as part of syndrome like Bart syndrome. The lesion may vary from a superficial ulcer to a deep defect involving bone or dura. Most isolated cases heal spontaneously with atrophic scarring, while extensive lesions may require surgical intervention. Early recognition and appropriate management are essential to prevent morbidity and optimize cosmetic outcomes. Multidisciplinary care ensures comprehensive evaluation and treatment.

KEYWORDS

Aplasia Cutis Congenita, Developmental anomaly, Neonatal skin defect

INTRODUCTION

Aplasia Cutis Congenita (ACC) is a rare congenital disorder characterized by the localized or widespread absence of skin and sometimes underlying structures such as bone or dura. The scalp is the most frequently involved site. The etiology is heterogeneous, with both genetic and environmental factors implicated. The lesion may range from superficial absence of epidermis to full-thickness defects exposing underlying structures such as bone or dura. Several genes have been implicated depending on the type and associated syndrome. Isolated Aplasia Cutis Congenita is associated with BMS1, KCTD1 or TERT variants which mainly affect skin morphogenesis and follicular development.

Here we report a one day old female baby with absence of skin over both legs and feet since birth which is very rarely reported in the literature.

CASE REPORT

One day old female baby, born via caesarian section at term gestation presented with absence of skin over both legs and feet since birth which was noted post baby wrapping. No history of oozing from the site of lesions, fever or associated systemic illness in the neonate. No history of infection, trauma, teratogenic drug intake during pregnancy or significant comorbidities in mother.

The neonate was managed with wet sterile gauze covering over the lesions and topical treatment with fusidic acid cream in the morning and night. Collagen dressing was done on alternate days.

ON EXAMINATION :

Few well demarcated erythematous areas of absent skin exposing subcutaneous tissue present over the anterior aspect of both legs and dorsal aspect of feet, with the base appearing smooth showing areas of thin epithelial covering and surrounding shiny, atrophic skin.
Histopathology was not performed for ethical concerns.

Figure 1&2 shows solitary well demarcated erythematous areas of absent skin exposing subcutaneous tissue present over the anterior aspect of lower part of both legs and dorsal aspect of feet approximately measuring 1x2cm on right side and 2x3cm on left side.

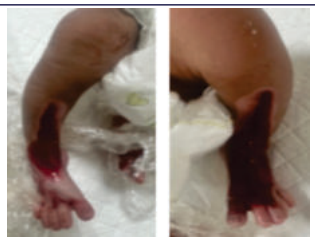


Figure 3 & 4 show solitary well defined erythematous area of absent skin with exposed subcutaneous tissue over the medial aspect of plantar surface of both feet mainly over the great toe and adjacent region of soles of both feet.

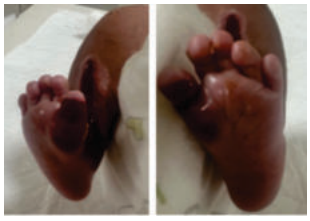
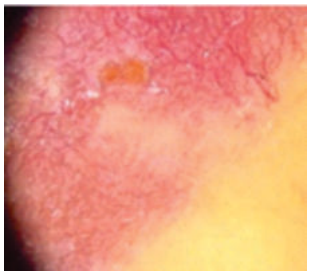


Figure 5 shows dermoscopic findings of visible vascular network denoted by erythema and white streaks centrally with telangiectasia at the borders.



DISCUSSION

An uncommon congenital lack of skin, Aplasia Cutis Congenita

(ACC) affects the epidermis, dermis, and sometimes subcutaneous tissue. Numerous factors have been implicated in the syndrome, including teratogens, trauma, genetic alterations, amniotic band sequencing, vascular impairment, and disturbance of embryonic skin development. Risk factors include chromosomal abnormalities, maternal drug exposure, and intrauterine infections. Defective morphogenesis or the degradation of skin that has already developed during the first trimester is part of the pathogenesis. 1

Aplasia Cutis Congenita is a visible sign of aberrant intrauterine skin development and has no known underlying cause. There are probably a variety of causes for the disease. Genetic variables, placental infarcts, trauma, teratogens as methimazole, intrauterine infections, and neural tube abnormalities (NTDs) are among the hypothesised aetiologies. Until recently, no particular genetic target had been found. The BMS1 gene, however, has been suggested as a potential contribution by recent data. 2

In 1986, Frieden put up a classification scheme for Aplasia Cutis Congenita that is still in use today, creating nine groups based on the location and number of lesions as well as the existence or lack of related abnormalities. This particular case belongs to Group 7 which describes Aplasia Cutis Congenita located to extremites without blistering. 2

Cutaneous defects noted at birth may initially be mistaken for obstetric trauma. The most generally recognised pathophysiologic model explains how tension during foetal development interferes with appropriate skin approximation. Ingested teratogens, foetal or placental ischaemia, intrauterine infections, and NTDs are examples of exogenous factors that may contribute. Dominant-negative mutations in the KCTD1 and KCTD15 genes have been found in recent studies to produce Aplasia Cutis Congenita via impairing function.3

This is a largely clinical diagnosis. For large, high-risk flaws, management options include surgical closure and conservative wound care. Isolated, superficial lesions often have a good prognosis, but complex cases with underlying structural abnormalities have higher morbidity and fatality rates. Aplasia Cutis Congenita is mainly diagnosed clinically, though histopathologic examination may be helpful in some patients. Keeping the patient's age in mind, many physicians steer clear of lesional biopsy.4

The extent of the lesion and the existence or lack of related abnormalities determine the management strategy for Aplasia Cutis Congenita. Treatment for tiny lesions (less than 4 cm) with no other abnormalities usually consists of topical antibacterial ointment and daily cleaning until the lesions heal completely. The main goals of management are to lessen the chance of fluid loss or organ exposure, as well as to restore the skin's mechanical and immunological integrity. Small lesions typically leave an atrophic, hairless scar after healing in a few weeks to months.5

CONCLUSIONS

This case highlights a rare and atypical presentation of Aplasia Cutis Congenita, with lesions confined to the extremities—a site infrequently involved in this condition. Although the scalp is the most commonly affected areas, clinicians should be aware of such unusual presentations to avoid misdiagnosis.

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