Dermatology Reports
https://www.pagepress.org/journals/index.php/dr/index

eISSN 2036-7406

Please cite this article as:

Poddine G, Salvagno S, Bellinato F, et al. Membranous aplasia cutis congenita. *Dermatol Rep 2025 [Epub Ahead of Print] doi: 10.4081/dr.2025.10544*

Submitted 17/07/25 - Accepted 29/10/25

Membranous aplasia cutis congenita

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Key words: aplasia cutis congenita; congenital skin defects; newborn skin defects.

Conflict of interest: the authors have no conflict of interest to declare.

Ethics approval and consent to participate: ethical approval was not required. Informed consent was obtained from the patient's relatives.

Availability of data and materials: the datasets used and/or analyzed during the current study are available upon reasonable request from the corresponding author.

Dear Editor,

Aplasia cutis congenita (ACC) is a rare congenital malformation characterized by the focal absence of skin visible at birth, most frequently involving the scalp, although any part of the body may be affected.[1] The estimated incidence is approximately 1 to 3 per 10,000 live births. The pathogenesis is heterogeneous and yet not fully understood. Proposed etiologies include genetic factors, teratogenic insult exposure, vascular disruptions, and mechanical trauma in utero.[2] Clinically, ACC presents with variable morphology. The most frequent presentation is a well-demarcated area of ulceration or atrophic scarring on the scalp.[1,2] ACC can rarely manifest as a bullous or membranous lesion with a cystic, translucent appearance, occasionally associated with deeper tissue involvement.[3,4] This article describes the case of a 9-month-old female patient with a rare form of membranous ACC.

The patient was referred for the evaluation of a solitary and asymptomatic bullous lesion on the scalp. The lesion had been present since birth and remained stable over time. No associated systemic symptoms or concomitant developmental anomalies were reported. There was no relevant family or prenatal history. Physical examination revealed a 5 mm bullous lesion on the right parasagittal scalp region (Figure 1A). The lesion appeared translucent, round-shaped, and filled with clear fluid. Dermoscopy demonstrated fine arborizing vessels overlying a translucent background, allowing visualization of hair roots and bulbs aligned in an arch-like distribution at the periphery of the lesion (Figure 1B). The remainder of the skin examination was unremarkable. Color Doppler ultrasonography revealed a well-defined hypoechoic area measuring 5×15 mm, with no signs of increased vascularity or underlying bone involvement. The lesion was diagnosed as a membranous variant of ACC, and a conservative approach with regular clinical follow-up was adopted.

ACC encompasses a spectrum of conditions characterized by the congenital focal absence of the skin, with or without involvement of deeper structure.[1] ACC most commonly affects the scalp, particularly along the midline, but can occur anywhere on the body and can present in various forms. Frieden's classification system categorized ACC into nine groups based on lesion morphology, inheritance patterns, associated anomalies and underlying etiology (genetic, vascular, or teratogenic). This classification reflects the distinction between isolated and syndromic forms of ACC, but also facilitates a more precise diagnostic interpretation of the condition.[1]

Membranous ACC is particularly rare and may be easily mistaken for other neonatal scalp lesions. Zhang *et al.*, in a case series, reported that the majority of cases presented as a solitary lesion mostly located in the parietal region of the scalp, with a higher prevalence observed among female patients.[5] This variant typically presents as an alopecic area surmounted by a soft, fluid-filled lesion coated by a thin translucent and vascularized layer of epidermis.[6,7] The underlying causes of membranous ACC

remain incompletely understood, however, recent studies emphasize a possible association with chromosomal abnormalities, particularly trisomy 13 and 18.[8,9] Among the leading hypotheses, membranous ACC is thought to represent an incomplete variant of a neural tube defect.

Usually, it is visible a characteristic collarette of dark hair ("Hair Ring Sign") along the peripheral edge of the lesion, which may suggest a possible neural tube defect.[10] Among the distinctive dermoscopic features of membranous ACC are the central absence of hair follicles and the presence of a translucent epidermis, through which hair roots and bulbs are visible and arranged in an arch-like configuration along the lesion margin, a finding referred to as the "translucency sign".[6] In addition, the hair collar sign, as previously described, is also considered a characteristic finding.

In most patients membranous ACC is an isolated lesion but still requires further evaluation to exclude any associated anomaly, in particular neural tube defects or midline development anomalies.[9] Ultrasound with color doppler is typically the first line imaging modality in neonates, allowing non-invasive assessment of intracranial structure.[3,7] Of note, the literature includes two cases in which prenatal imaging raised suspicion of membranous ACC, which was later confirmed postnatally. In selected cases, especially when ultrasound is inconclusive, brain magnetic resonance should be considered.[7] Histological analysis is not routinely required, but if performed shows an atrophic epidermis with a loose fibrovascular stroma and edematous dermis.[6]

The differential diagnosis of membranous ACC includes dysraphism, epidermal nevi, dermoid inclusion cyst, epidermolysis bullosa, congenital infection and perinatal trauma.[3,5] Syndromic association, including Adams-Oliver syndrome, trisomy 13 and 18, should also be considered.[8,9]

Management of membranous ACC is primarily conservative. Typically, an isolated lesion of the scalp requires only topical intervention with non-adherent dressing and antibiotic ointment.[6] Surgical procedures may be required for extensive defects involving the dura mater or venous sinuses, which warrant early specialistic evaluation. In the absence of associated anomalies, the prognosis is generally favorable, with acceptable cosmetic outcomes.[3]

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Figure 1. A) Scalp solitary bullous lesion filled with clear liquid. **B**) Dermoscopy shows fine arborizing vessels over a translucent background within which is possible to observe hair roots and hair bulbs arranged in an arch shape along the margins of the lesion.