SKIN

SKINmages

Membranous Aplasia Cutis Congenita in an Atypical Location

Adarsh Shidhaye, BS[1], Julia Lunt, BS[1], Yash Verma, MS[1], David Lobb, MD[2]

[1] University of South Carolina School of Medicine Greenville, SC, USA
[2] Department of Plastic Surgery, University of South Carolina School of Medicine Greenville

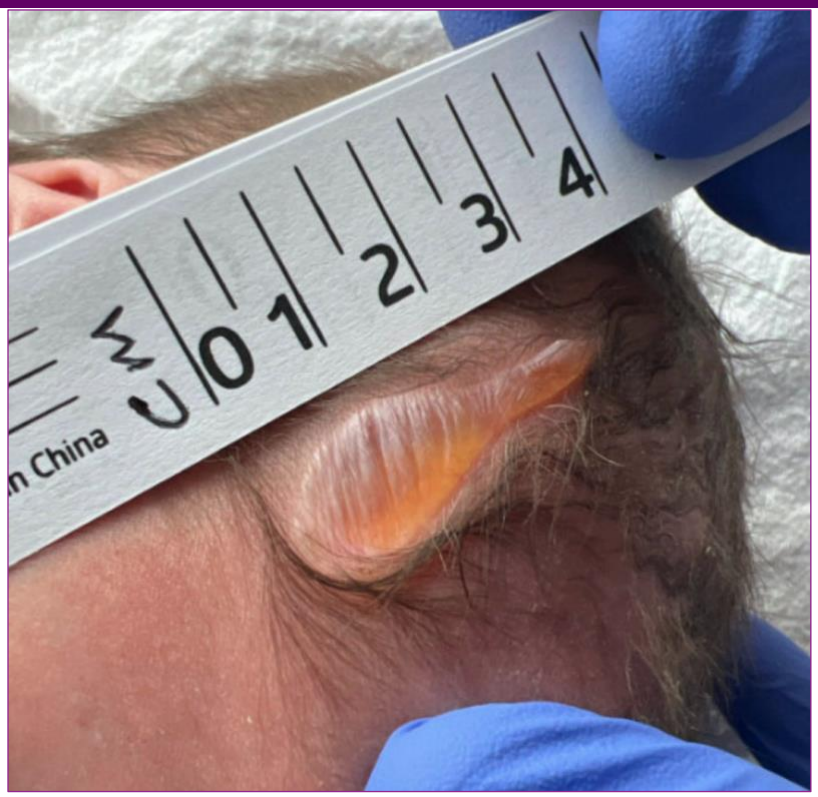

Figure 1. Neonatal female with a glistening 3.5 x 1.5 cm fluid-filled lesion at the right frontoparietal aspect of the scalp.

INTRODUCTION

Aplasia Cutis Congenita (ACC) is a rare congenital defect characterized by the absence of the epidermis and dermis with varying absence of underlying structures such as calvarium and dura. Membranous or bullous aplasia cutis congenita (MACC) is an extremely rare subtype of ACC, presenting as a fluid-filled lesion with a membrane overlying the skin defect, typically located at the vertex of the scalp.[1] We present a case of MACC at the frontoparietal region in an otherwise healthy female.

CASE REPORT

Following an uncomplicated pregnancy and vaginal delivery, a neonatal female presented with a glistening 3.5 x 1.5 cm fluid-filled lesion

at the right frontoparietal aspect of the scalp (**Figure 1**). The lesion was devoid of hair and was not associated with a hair collar sign indicative of a neural tube defect. No other abnormalities were noted on physical examination. A skull x-ray was negative for an associated bony defect. Genetic testing and biopsy were not indicated. The lesion was managed expectantly with simple emollient dressings and close follow-up.

DISCUSSION

The incidence and cause of MACC are not completely understood; however, it is postulated that intrauterine infections, trauma, placental infarctions, ectodermal dysplasia, chromosomal abnormalities, and teratogenic exposure during pregnancy may play a role in pathogenesis.[2] Previous studies indicate that MACC occurs sporadically but may be associated with neural tube defects or cardiac abnormalities.[1] Distinct histologic and trichoscopic features of MACC, including thick distended vasculature, hair collar sign, absence of follicular openings, and bulbs of anlagen hair seen through translucent epidermis, have been reported and may differentiate from other conditions.[4] Histopathology is characterized by fibrovascular or edematous stroma and atrophic epidermis, demonstrating similarity between MACC and encephaloceles and meningoceles and supporting classification as a forme fruste neural tube defect.[5]

Management of MACC is similar to ACC, with non-surgical management recommended for small lesions and operative intervention indicated for large defects or lesions complicated by infection, vascular exposure or bleeding.[3] In suspected cases, workup with neurological imaging, genetic testing, and cardiac assessment can aid in identifying associated comorbid conditions.[3]

CONCLUSION

This case adds to existing extremely limited reports of MACC and documents an uncommon area of presentation. Further pathophysiological studies are required to effectively elucidate the underlying cause of MACC.

Conflict of Interest Disclosures: None

Funding: None

Corresponding Author:
Adarsh Shidhaye, BS
607 Grove Road, Greenville, South Carolina 29605
Email adarsh@email.sc.edu

References:

1. She QY, Zhu HL, Liu ZR, Huang WN. Membranous aplasia cutis congenita: A rare case report highlighting clinical presentation, genetic insights, and the need for comprehensive evaluation. *Heliyon*. 2024;10(13):e33742. Published 2024 Jun 27. doi:10.1016/j.heliyon.2024.e33742
2. Blionas, A., Giakoumettis, D., Antoniades, E., Drosos, E., Mitsios, A., Plakas, S., Sfakianos, G., & Themistocleous, M. S. (2017). Aplasia cutis congenita: Two case reports and discussion of the literature. *Surgical neurology international*, *8*, 273. https://doi.org/10.4103/sni.sni_188_17
3. Harvey G, Solanki NS, Anderson PJ, Carney B, Snell BJ. Management of aplasia cutis congenita of the scalp. *J Craniofac Surg*. 2012;23(6):1662-1664. doi:10.1097/SCS.0b013e31826542de
4. Rout, A., Arora, S., Dabas, R., & Paul, D. (2021). Bullous Aplasia Cutis Congenita-Description of a Novel Dermoscopic Feature. *Dermatology practical & conceptual*, *11*(2), e2021012. https://doi.org/10.5826/dpc.1102a12
5. Colon-Fontanez F, Fallon Friedlander S, Newbury R, Eichenfield LF. Bullous aplasia cutis congenita. *J Am Acad Dermatol*. 2003;48(5 Suppl):S95-S98. doi:10.1067/mjd.2003.150