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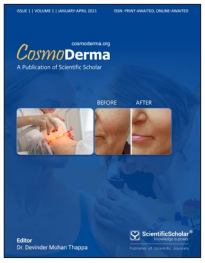

Images/Instrument in Dermatology/Dermatosurgery

Bullous aplasia cutis congenita: A rare entity in the paradigm of congenital skin defect disorder

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A 3-day-old female child, born to a healthy 28-year-old female at term via normal vaginal delivery, presented with a lesion over the scalp which was present since birth. On examination, a solitary soft cystic non-pulsatile dome-shaped swelling of size approximately 1.5 × 2.5 cm and covered with a yellowish semi-translucent membrane was noted over the vertex just lateral to the midline. At the periphery, the swelling showed an erythematous raised border. The surface of the lesion was devoid of any hair. Thin arborizing vessels were noted over the membrane traversing from the peripheral border toward the center of the swelling [Figure 1]. Based on the above clinical findings, a diagnosis of bullous aplasia cutis congenita (ACC) was made. The collar of hair which is ominous for neurological involvement was not seen. The central nervous system examination of the child was normal. Due to loss to follow-up, neither the findings of radiological imaging nor the evolution of the lesion could be ascertained.

ACC is a rare congenital anomaly that occurs generally over the scalp and rarely over the trunk and limbs. It was first described by Cordon in 1767 and later over scalp by Campbell in 1826.[1] It is characterized by localized or generalized congenital absence of skin, which in approximately 20–30% of cases may extend deeper to involve the bone and the dura. The incidence of ACC is 3 in every 10,000 births with a female/male ratio of 7:5.[2] It may be familial or associated with genetic syndromes or malformations.[3]

Bullous ACC or membranous ACC is a rare clinical variant of ACC with <20 cases reported so far. It presents as a solitary or multiple cystic or bullous lesion on the scalp (slightly lateral to midline), which is gradually replaced by an atrophic, flat scar covered by a thin epidermis.[1-3] Vertex is a common site as it is subjected to increased tensile stress during a period of rapid brain growth.[2] The exact etiology is still unknown but multiple factors such as genetic abnormalities, trauma, intrauterine/amniotic defects, vascular compromise, and teratogens are proposed to play a role in the etiopathogenesis.[1] The bullous aplasia cutis congenita (BACC) is proposed to be an incomplete type of neural tube defect supported by collar sign, a marker of cranial neural tube defects.[2] The diagnosis is primarily based on clinical features such as presence since birth, over the scalp, and hair collar sign (collar of dark and coarse hair around the lesion). Lesional and transfontanellar ultrasonography is recommended to rule out underlying defects in the skull or brain.[2,3] Management is usually conservative for smaller defects while early surgical intervention is required for larger defects extending into the dura mater.[2]

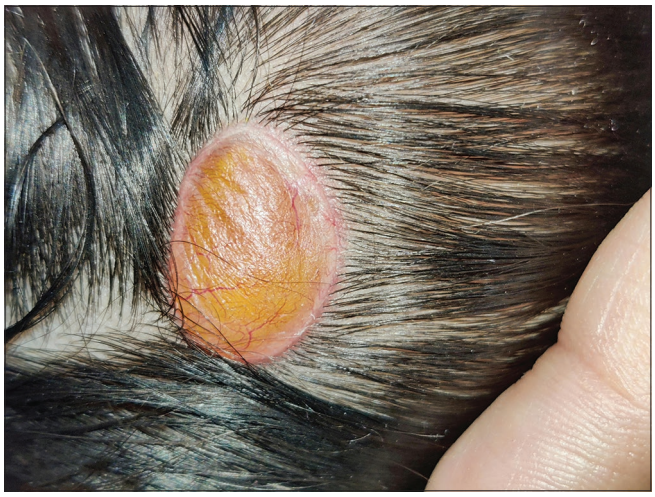

Figure 1: A single soft cystic non-pulsatile non-hairy dome-shaped swelling covered with a yellowish semi-translucent membrane bearing thin arborizing vessels was noted over the vertex just lateral to midline.

In conclusion, BACC is a rare congenital anomaly that is generally diagnosed clinically. It may be misdiagnosed owing to its rarity; hence, awareness among clinicians is required for prompt diagnosis and adequate evaluation to rule out associated cranial and cerebral defects.

Acknowledgement

We have obtained appropriate consent from the parent to use the clinical details and clinical images for publication.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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