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Case Report

Aplasia cutis congenita in two brothers – a rare occurrence

Amer Ejaz*, Naseem Raza, Majid Suhail***

* Department of Dermatology, Combined Military Hospital, Kharian.
** Department of Dermatology, Combined Military Hospital, Abbottabad.

Abstract Aplasia cutis congenita (ACC) is characterized by the absence of a portion of skin in a localized or widespread area at birth. It is a rare disorder with a complicated pattern of inheritance. No unifying theory can account for all lesions of ACC. Apart from the isolated finding of aplasia cutis; several reports have associated it with a large number of developmental anomalies. Therefore, a complete physical examination should be performed to search for associated physical anomalies or recognizable malformation syndromes. We document two brothers aged, 6 years and I year, who presented with aplasia cutis congenita at almost the same location on scalp.

Key words Aplasia cutis congenita, familial.

Introduction

Aplasia cutis congenita (ACC) is characterized by the absence of a portion of skin in a localized or widespread area at birth. More than 500 cases have been reported since it was first described, but because of significant underreporting of this generally benign disorder, the precise frequency is unknown. It most commonly presents as a solitary (70%) defect on the scalp, but sometimes it may occur as multiple lesions.[1] The lesions are noninflammatory and well demarcated, and range in size from 0.5 cm to as large as 10 cm. ACC may be circular, oval, linear, or stellate in configuration. At birth, the lesions may have already healed with scarring or remain superficially eroded to deeply ulcerated, occasionally involving the dura or the meninges.

The hallmark of ACC in the neonatal period is the presence of a solitary well-demarcated, punch-out lesion which must be differentiated from traumatic cutaneous lesions (fetal monitor injuries). Aplasia cutis congenita may be isolated or it may occur with trisomy 13, 4p- syndrome, ectodermal dysplasia, and amniotic bands. Aplasia cutis in the hair whorl area is usually a benign finding. Neonates with ACC in the craniofacial and lumbosacral regions should have an MRI or ultrasound to evaluate the central nervous system structures below it. Isolated aplasia cutis congenita is usually sporadic.

We report two brothers aged, 6 years and 1 year, who presented with aplasia cutis congenita at almost the same location on scalp.

Address for correspondence
Dr. Amer Ejaz,
Consultant Dermatologist,
CMH, Kharian, Pakistan.
E mail: amer_ejaz@yahoo.com

Case report

Our patient, a one-year-old boy was brought to us for a bald patch at the vertex of his scalp since birth. The lesion was non-itchy and did not increase in size. On examination, he was found to be a healthy infant. There was a circular patch of hair loss on the vertex (**Figure 1**). There were no signs of inflammation or scaling. His dentition, rest of the hair growth, nails and skin were normal and hair collar sign was negative. He had achieved all the developmental milestones for his age and was in good health. He was accompanied by his elder brother, a six-year-old boy who also exhibited the same kind of patch, at almost the same location at the present since birth (**Figure 2**). The lesion was asymptomatic without scaling or signs of inflammation and a negative hair collar sign. He was also in good health with no anomalies of skin, teeth, nails and rest of hair. The mother had normal deliveries conducted at a general hospital in both instances. She had one other daughter who was asymptomatic. She did not give any history of drug intake during her pregnancies. The boys are being followed up at regular intervals.

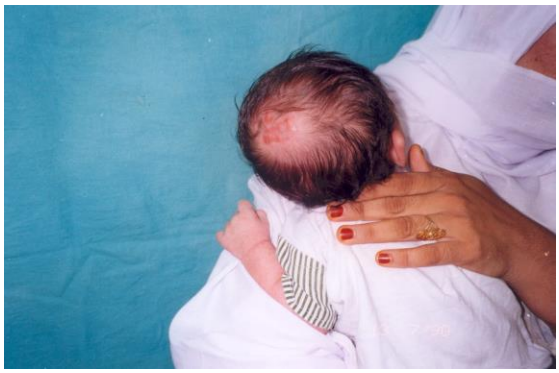

Figure 1 A circular patch of alopecia on the vertex representing aplasia cutis.

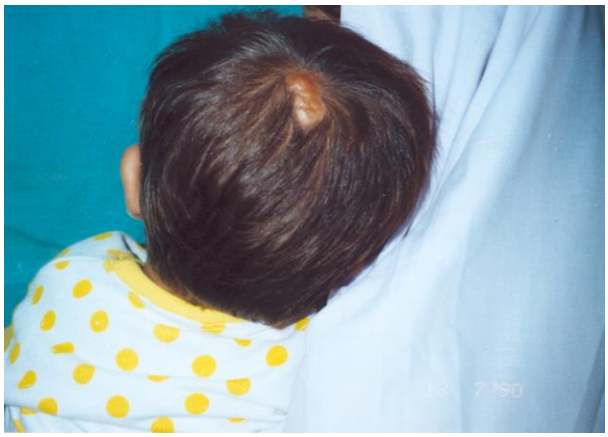

Figure 2 A patch of alopecia on vertex region in the elder brother.

Discussion

Aplasia cutis congenita is a rare disorder with a complicated pattern of inheritance. No unifying theory can account for all lesions of ACC. Because this condition is the phenotypic result of more than one disease processes, it is likely that more than one mechanisms are involved and these include genetic factors,[2] teratogens (methimazole), [3] compromised vasculature of the skin, and trauma. Of particular note is the association of fetus papyraceous with bilaterally symmetric ACC.[4]

The lesions are noninflammatory and well-demarcated. The appearance of lesions varies, depending on when they occur during intrauterine development. Early lesions have healed by the time of delivery and appear as scar tissue, while

later lesions are less mature and may present as ulceration. The involvement may be limited to the epidermis and superficial dermis; however, it can extend deeper, through the subcutaneous tissue, periosteum, bone and may rarely reach dura mater.[5] Distorted hair growth around a scalp lesion, known as the hair collar sign, is a marker for underlying defects. A complete physical examination should be performed to search for associated physical anomalies or recognizable malformation syndromes.

Several morphological types of aplasia cutis have

been described in the literature. The one presented by us is the familial type. Other reports include giant aplasia cutis of scalp without associated anomalies,[6] bilateral abdominal aplasia cutis without skull defect,[7] aplasia cutis of distal forearm with radial dysplasia,[8] and isolated, unilateral involvement of a leg.[9]

Apart from the isolated finding of aplasia cutis, several reports have associated it with a large number of developmental anomalies. Most of these belong to the ectodermal dysplasias group of developmental anomalies. The complete list of all those syndromes is exhaustive, ever increasing and beyond the scope of this article. However, a complete physical examination should be performed in every case of ACC to search for associated physical anomalies or recognizable malformation syndromes. Scanty publications are available on the subject in Pakistani literature.[10]

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