Aplasia Cutis Congenita: A Rare Case with Extensive Symmetrically Distributed Lesions

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Abstract

Aplasia cutis congenita is a rare developmental disorder of the skin of neonates, usually presenting as a solitary lesion over the scalp. We report an interesting presentation of AC along with the histopathological features in a neonate with extensive lesions over scalp as well as in bilaterally symmetrical areas over trunk and thighs; such symmetrical distributions being rarely reported.

Introduction

Aplasia cutis (AC) or congenital absence of skin is a relatively uncommon developmental skin disorder, affecting approximately 3 in 10,000 live births. It usually presents as localized lesions, with scalp being the most commonly described site. We report an unusual presentation of AC along with the histopathological features in a neonate with extensive lesions over scalp as well as in bilaterally symmetrical areas over trunk and thighs; such symmetrical distributions being rarely reported, the first to be reported from India.

Case Report

A 12 days old female neonate, born at term, out of non-consanguineous marriage, with an uneventful natal and postnatal period, was referred to our institution for evaluation of multiple skin lesions present since birth. On examination, the baby weighed 3.1 Kg, had stable vital signs and normal neonatal reflexes. The scalp showed a 2 x 2 cm area of altered skin texture with localised alopecia. The skin in this area was smooth, thin, slightly depressed and had a parchment like consistency with visible underlying capillaries without any tenderness (Figure 1). Similar lesions were seen over abdomen in both the flanks, each measuring about 5 x 6cm; and bilateral thighs, each measuring about 3.5 x 3cm (Figures 2a, 2b, 3). There was minor bleeding from the scalp lesion. The neonate was haemodynamically stable, systemic examination was unremarkable and there were no clinical stigmata of any other congenital abnormality. The mother was a primigravida with no prior bad obstetric history. There was no history of trauma, maternal intake of drugs or maternal infections in the antenatal period and the maternal serological tests for common intrauterine infections like toxoplasma, rubella, cytomegalovirus, herpes and syphilis were negative. There was no history of any such skin lesions in any member of the family.

Investigations showed a total leucocyte count of 12,300 per cumm, haemoglobin of 13 gm/dl and platelet count of 210,000/cumm. The parameters for liver and kidney functions were within normal limits. Biopsy from the abdominal lesion revealed loss of folds in the epidermis with dermis showing mild perivascular chronic inflammatory infiltrate, homogenisation of collagen and absent dermal appendage, consistent with a diagnosis of healing aplasia cutis (Figure 4). Further investigations to search for associated anomalies showed a normal X-Ray skull and ultrasonography of cranium...
Fig. 1: Lesion of Aplasia cutis over scalp, 2 x 2 cm in size, showing smooth, thin, slightly depressed parchment like skin with localised alopecia with visible underlying capillaries.

Fig. 2: (a) Lesion of Aplasia cutis over left flank, measuring about 5 x 6 cm; (b) Lesion of Aplasia cutis over left thigh, measuring about 3.5 x 3 cm.

Fig. 3: Lesions of Aplasia cutis over right flank (5 x 6 cm) and right thigh (3.5 x 3 cm).

Fig. 4: Skin biopsy (Hematoxylin and Eosin stain; 100x) showing loss of folds and absent dermal appendages.

Aplasia cutis is a rare dermatological disorder in newborns, reported infrequently. Most of these cases had solitary lesions over the scalp. Multiple site involvement with symmetrical distribution, as seen in our case, has scarcely been reported. Also, very few have mentioned the histological findings. The disorder is not known to have any racial or sex predilection. The lesions are present since birth and have been described as circular, oval, linear or stellate in shape, ranging from 0.5-1 cm in size.

Scalp lesions may be severe enough to involve the full thickness of the calvarial bone and duramater resulting in sagittal sinus haemorrhage. Isolated cases of AC of trunk associated with biliary atresia, duodenal atresia and intestinal infarcts have been reported. Other rare associations include SCALP syndrome (sebaceous nevus, central nervous system malformations, aplasia cutis congenita, limbal dermoid and pigmented nevus), neurocutaneous melanosis, foetus papyraceus, Adams-Oliver syndrome, Bart’s syndrome and Johanson-Bilzard syndrome.

Several aetiologies have been postulated for the occurrence of AC like hereditary factors, teratogens (methimazole, carbimazole, misoprostol, valproic acid, diclofenac), foetal trauma (intrauterine shearing forces), intrauterine infections, though no single theory can account for all the lesions. Symmetrical lesions of AC have been suggested to be due to a vascular disruption inducing abnormal dermoeipidermal development.

The complications associated with AC include infection, haemorrhage and thrombosis. Treatment modalities depend upon the extent and depth of involvement, with isolated cutaneous involvement, as in our case, requiring a conservative approach. However, involvement of vital organs would warrant early surgery. In case of absence of duramater, temporary closure with a local temporal flap combined with a periosteum patch to protect the underlying exposed structures. Definitive repair can then be performed using scalp flaps, split and full-thickness skin grafts, cultured epithelial autografts, delayed split rib cranioplasty, tissue expansion, and composite cranioplasty. Management difficulties can occur with scar excision of AC which requires complex tissue rearrangement, tissue expansion, or skin grafting.
Conclusion

To conclude, aplasia cutis is a rare dermatological disorder of neonates which, though commonly affects the scalp, may occasionally have an extensive bilaterally symmetrical presentation.

References