

Case Report

Aplasia cutis congenita with two completely different presentations

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ABSTRACT

Aplasia cutis congenita is a rare disorder, which presents with congenital absence of skin. Besides isolated presentation, it may manifest with other developmental malformation of cardiovascular, gastrointestinal, genitourinary and central nervous systems, and malformation syndromes such as chromosomal abnormalities, Adams-Oliver syndrome, Bart's syndrome, and Johanson-Bilzard syndrome. Here, we present two cases of aplasia cutis congenita, which represent two different types of the disease. The first case presented as non-syndromic aplasia cutis congenita of scalp, and the other case presented as a part of Bart's syndrome.

Keywords: Aplasia cutis, bart's syndrome, collar sign

Introduction

Aplasia cutis congenita is a disorder, which commonly involves the skin of the scalp and other areas but may involve the subcutaneous tissue, bone, and dura mater. Frieden's classification has described it in 9 types.^[1] As per this classification, our cases belong to type1 [non-syndromic aplasia cutis congenita of the scalp] and type 6 or Bart's Syndrome [congenital absence of skin with epidermolysis bullosa], which is a very rare disorder. It represent antenatal loss of skin most commonly in association with dominant dystrophic type epidermolysis bullosa and also junctional and recessive epidermolysis bullosa simplex.^[2]

Case Reports

Case 1

A 7-day-old female baby presented with a solitary circular erythematous ulcer over scalp since birth [Figure 1]. Underlying bone was not affected. She had no other clinical sign and symptom suggestive of other abnormalities. No significant family history neither any history of

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drug exposure nor any infection in maternal side during pregnancy was present. Mother was primi-gravida, and there was no history of consanguinity. The child was born by cesarian section. Laboratory investigations showed normal complete hemogram, but echo-cardiography on day 3 showed atrial septal defect (ASD), ventricular septal defect (VSD), tricuspid regurgitation (TR), and pulmonary hypertension. Serous discharge from the lesion was sent for culture and sensitivity, but it did not showed growth of any organism. Skin biopsy was suggested, but parents refused the procedure. The baby has been treated with topical antibiotic, and on day 20, the lesions started to re-epithelialize. The parents have been asked to consult plastic surgeon later on.

Case 2

A 15-day-old, pre-term, low birth weight female baby presented with large area of denuded skin involving the anterior aspect of right leg and knee and also chest, since birth [Figure 2]. Birth was by normal vaginal delivery. She was the 2nd child of a couple. No history of consanguinity was present. The other sibling was not affected. Her nails were normal. During presentation, the major part of the lesion had healed leaving behind atrophic scarring. Two bullous lesions were present, one over right great toe and another over right middle finger [Figure 3]. Other clinical examination findings were normal. History of recurrent bullous lesion, which healed by scarring over acral part, was

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present in mother [Figure 4], but no history of drug exposure and infection during her pregnancy could be elicited. The parents refused skin biopsy. The baby was treated with topical antibiotic, and proper care of the wound was advised.

Discussion

Aplasia cutis congenita (ACC) is a multifactorial disease. It was first described by Cordon in 1767.^[3] Some genetic disorders are found to be associated with aplasia cutis. Frieden's classification has described it in 9 types. Type 1 is non-syndromic aplasia cutis congenita of the scalp, type 2 is ACC with limb reduction abnormalities, type 3 is ACC with epidermal nevi, type 4 is congenital absence of skin overlying developmental malformations, type 5 is ACC with fetus papyraceous, type 6 is associated with epidermolysis bullosa, type 7 is isolated congenital absence of skin localized to the extremities, type 8 is caused by teratogens or intrauterine infection, type 9 occurs as a feature of malformation syndrome. Though the classification has described most of the factors, some authors proposed it as an incomplete type of neural tube defect due to presence of the collar sign, which is characteristic of neural tube defect.^[4]

Type 1, which is our first case, usually presents as single lesion. Most of the lesions appear over scalp, but involvement of trunk and extremities may occur.^[5] Autosomal-dominant inheritance has been demonstrated. In autosomal-dominant ACCA, heterozygous Arg-to-His missense mutation in the ribosomal GTPase BMS1 is identified in ACC that is associated with a delay in 18S rRNA maturation, consistent with a role of BMS1 in processing of pre — rRNAs of the small ribosomal subunit.^[6] The increased incidence over vertex point toward its occurrence over the area of maximum tensile strength during the period of rapid brain growth.^[5,7] Dilated scalp vein also indicates some vascular pathology. The lesions may be superficial or deep, ulcer or bulla, which heal with scarring. It should be distinguished from birth trauma, which occurs due to scalp electrode.^[8]

The second case we are discussing is Bart's syndrome, which is an exceedingly rare disorder described in very few literature.^[9] Bart *et al.* reported it as a dominantly inherited disorder characterized by wide spread blistering over skin and oral mucosa, congenital absence of skin over lower legs and nail dystrophy.^[10] It is now considered as a non-specific feature of not only the dystrophic epidermolysis bullosa type but also junctional type with pyloric atresia and epidermolysis bullosa simplex variety.



Figure 1: Solitary erythematous ulcer over the scalp of the new born



Figure 2: Denuded area of skin over anterior aspect of leg, knee, and chest of the neonate



Figure 3: A bulla present over the great toe of the neonate



Figure 4: Healed scar over dorsal surface of mother's hand

The ultrastructural studies revealed the substitution of glycine in type VII collagen. Zelickson analyzed Bart's kindred and demonstrated poorly formed anchoring fibrils and cleavage below the lamina densa on ultrastructural analysis. Lesions along the Blaschko's line have also been described.^[11]

Histopathological study of the skin shows the absence of epidermis and sometimes the dermis. Dermis, if present, usually lacks appendages and connective tissue. Where re-epithelialization occurs, epidermis appears flat, dermis is devoid of appendages, and hypertrophic scarring may also occur.^[12] In familial cases, genetic counseling is needed. Prenatal diagnosis through amniocentesis and chorionic villus sampling can be helpful to rule out junctional and dystrophic epidermolysis bullosa.

Lesions of ACC usually heal with conservative management. Skin grafting or other reconstructive surgery is needed when lesion is extensive. The two cases were managed conservatively showing good result.

We have described two cases of the same fascinating and rare neonatal disease with diverse manifestations.

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