

Epidermolysis bullosa with congenital localised absence of the skin

This newborn baby was referred to us for the congenital absence of the skin. He is the second child of a consanguineous couple. Pregnancy and delivery were normal. Both the parents were healthy with no history of blistering skin lesions in the family. On examination, there was an absence of the skin on the anterior and lateral aspects of the right leg and dorsum of the foot, extending from the knee up to the lateral half of sole involving digits and interdigital spaces (Fig. 1). The right leg seemed to be thinner with varus deformity of the knee and decreased interdigital spaces.

On day2, the child developed multiple blisters over buttock, under surface of the scrotum, medial surface of the left leg and cheeks (Fig. 2). He also developed multiple flaccid blisters over the digits and forearm which ruptured subsequently (Fig. 2). There was no oral lesion. Systemic examinations were normal.

The child was managed in newborn nursery with aseptic handling, prophylactic antibiotics, avoidance of friction and breastfeeding. Investigations revealed normal blood count, negative skin swab culture and septic workup. Magnetic resonance imaging (MRI) and skin biopsy could not be done as the baby developed lethargy on day 5 which progressed to shock and in spite of supportive management succumbed to death on day 6.

Congenital localised absence of the skin may be a manifestation of *in-utero* friction in epidermolysis bullosa (EB). Bart's syndrome was described in a large family in 1966

with the affection of 26 members and consisted of any one or a combination of the three characteristics; congenital absence of skin, blistering and associated nail abnormalities¹. The incidence of aplasia cutis congenita is 1-2 per 10,000 births but the exact incidence of Bart's syndrome is not known². Several isolated cases have been reported in the literature^{3,4}.

The latest international consensus meeting on diagnosis and classification of EB had proposed to eliminate the eponym in this subtype of EB and substituted with the descriptive term 'EB with congenital absence of the skin'⁵. The electron microscopy and genetic analysis were not possible in our case; however, the involvement of buttocks and paronychia inflammation as described is seen frequently in junctional EB⁶. DNA based prenatal diagnosis using chorionic villous sampling or amniocentesis is available for junctional and dystrophic forms of EB⁷. Genetic counselling for this rare disorder is important for affected families.

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Figure 1: Absence of the skin on the right leg and sole with thinning and decreased interdigital spaces

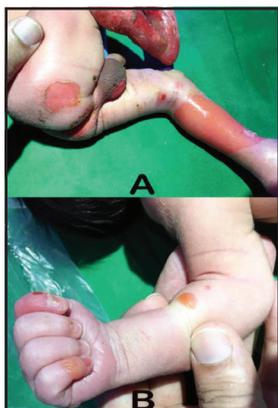


Figure 2: Blistering with erosion on buttock, scrotum and the leftleg (A); over the hands and digits (B).

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