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BART SYNDROME

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An infant presenting with extensive aplasia cutis on lower extremities later developed blisters on skin and mucous membrane. Clinical features and histopathological examination of skin favoured the diagnosis of Bart syndrome.

Key Word : Bart syndrome

Introduction

Bart et al[1] in 1966 described a kinship of 104 members over 5 generations, in whom 26 were affected with any one or a combination of the following traits : 1.congenital localised absence of skin, affecting the lower extremities, 2. blistering of skin or mucous membranes or both with no residual scarring, and 3. congenital absence or deformity of nails or both. It was also concluded that this represents a new syndrome which is different from other congenital localised aplasia of skin and from the various forms of epidermolysis bullosa.[1] Cases of aplasia cutis with blistering of skin are occasionally reported in Indian literature.[2,3] We encountered an infant having all the features of Bart syndrome.

Case Report

A 2-week-old-male child was brought for extensive ulcerative lesions on the lower extremities. The child was fourth issue born with full term normal delivery. Absence of skin on lower extremities was noticed by mother at birth of the child. Local practitioner advised systemic ampicillin and framycetin for topical use. There was no response to the treatment and on seventh day the child started developing blisters on sacral and scapular region and on both lips for which the child was brought to this hospital. Examination of the child revealed otherwise healthy child having normal neonatal reflexes. The skull was normal and all the nails were dystrophic. There

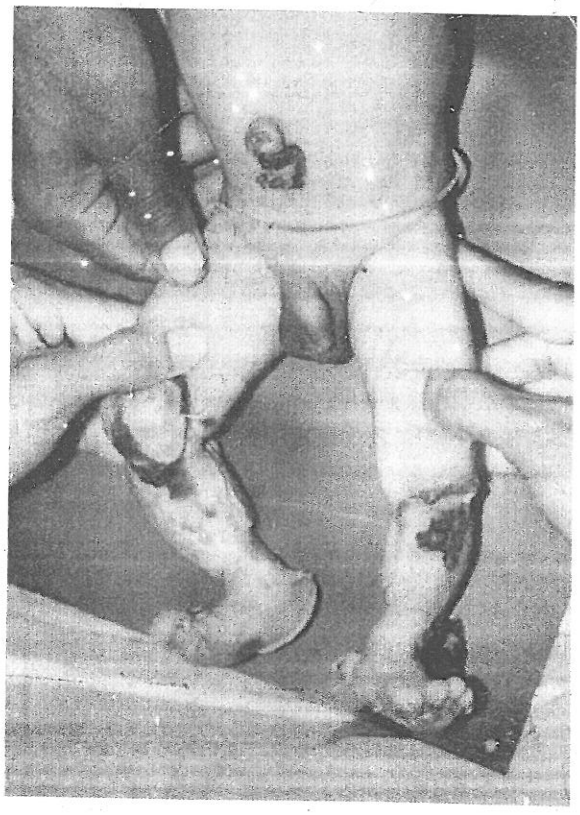

Fig. 1. Aplasia cutis on lower extremities

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was syndactyly of first 2 toes on left side. Local examination revealed symmetrically distributed aplasia cutis on the lower extremities. The surface of the ulcerative area was covered with inflammatory exudate and crusts (Fig. 1). There was clear demarcation between the normal and diseased skin. Blisters were present on the sacral area (Fig. 2), scapular

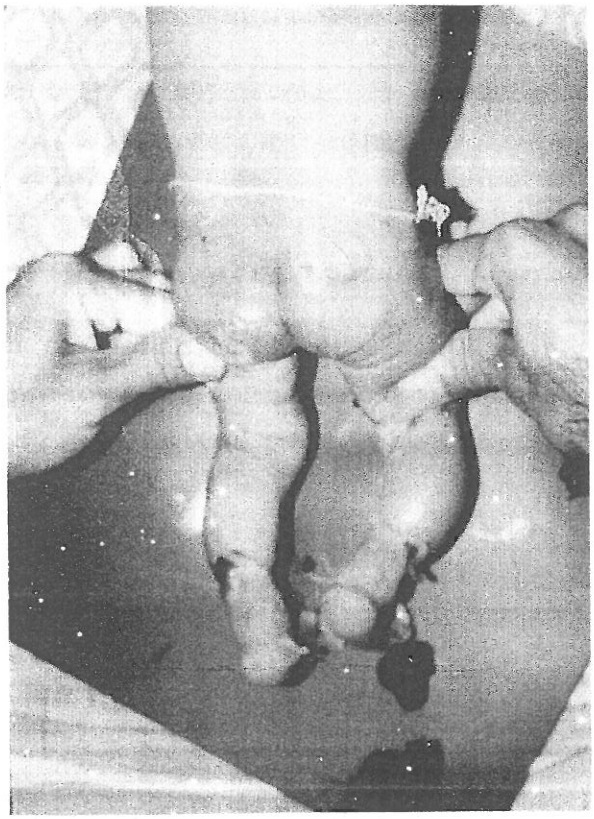

Fig. 2. Aplasia cutis and blisters in the sacral area

region and on both lips. Examination of oral cavity revealed superficial ulcerative lesions on buccal mucosa on both sides. There was no congenital anomaly and systemic examination was normal. Patient was the fourth issue of consanguinous parents. The first 2 female children were normal and alive. The third child was also born with similar skin lesions on lower extremities, the parents were not sure about the blisters on skin, the child died at the age of 6 weeks. Father and mothe of the patient had normal nails and there was no history of blisters on skin. Haemogram, urinalysis, kidney functio tests, liver function tests, X-ray chest and lower extremities were normal. Staph. *aureus* was grown from the ulcerative lesions on legs. Histopathological examination of skin from the centre o the lesion revealed absence of epidermis. There were fufts of sweat glands and hair follicles were absent. The patient was given inj. gentamycin, oral vitamin E and topical sisomycin. Ten days later the vesicles regressed and the infection was controlled, however there was no tendency for healing of the ulcerative lesions. Fresh blisters appeared on the sacral, scapular and scalp area. On 15th hospital day the child became suddenly dyspnoeic, developed tachycardia, became unconscious and died. Autopsy was not permitted by the parents.

Comments

The mode of inheritence of Bart syndrome is that of an autosomal dominent gene, showing full penetrance and variable expressivity.[1] Mild form of blisters without residual scarring following blistering, frequently seen nail abnormalities, blistering of mucous membranes, localised aplasia cutis and autosomal dominent inheritence separates this new syndrome from other types of epidermolysis bullosa.[1] In the second group of aplasia cutis the defect is common on extremities and blisters develop subsequently, the aplasia cutis in this group is indicative of post-developmental defect caused by some trauma in uterus.[2] In a patient with extensive aplasia cutis it may be a feature

of Bart syndrome.[1,3,4] It seems that Bart syndrome is not a rarity but in the initial stages it may be confused with aplasia cutis congenita, susequent development of blisters unfolds the diagnosis. There was no response to the drug therapy in our patient. Keeping the wound clean and immobilisation of the parts to avoid trauma was partially effective: Cause of death in the patient could not be ascertained, however mild type of blisters in the present case rules out the possibility of epidermolysis bullosa letalis.

References

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