CASE REPORTS

EPIDERMOLYSIS BULSOSUM SIMPLEX SIMULATING APLASIA CUTIS

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SUMMARY

Raw areas on skin are seen at birth in aplasia cutis congenita as well as epidermolysis bullosum. Histopathology is helpful in distinguishing one from the other. The continued development of blisters after birth is indicative of epidermolysis bullosum and not combination of aplasia cutis congenita and epidermolysis bullosum.

Similarities in clinical manifestations in different diseases often pose diagnostic problems. In such instances one has to look for collateral evidences, clinical or otherwise, to establish the diagnosis. In this paper we are presenting a case which illustrates this point.

CASE REPORT

A four year old male child was admitted to the Department of Dermatology in June 1967 for recurring ulcers on knees. The patient was born normally at full term following an uneventful pregnancy. At birth his mother noticed raw areas on all extremities. These areas were covered with thin transulucent membranes. No other skin lesion was present. All these raw areas healed within 5 months leaving either superficial atrophic scars or hyperpigmented areas. After the child started turning over the healed areas over the knees broke down several times. The one on the right knee never healed completely.

About the age of one the child first developed blisters over the scar and other parts of the body. These healed in 10–15 days leaving superficial pigmented areas. He never had blisters on the mucous membranes. Nails were normal at birth. He lost the right index finger nail at the age of 3 and both big toe nails at the age of 4, following trauma. During infancy he received several broad spectrum antibiotics for the ulcers. He had no other illnesses and his milestones were normal.

FAMILY HISTORY

Patient has two siblings. Both are well. There is no history of similar trouble in the family tree.

PHYSICAL EXAMINATION

Showed a healthy child with large superficial scars over the extensor aspects of the extremities and crusting over the knees (figs. 1 & 2). The right ankle was webbed due to scar infront (fig. 3). The scars were superficial and easily movable over the underlying structures. There were few hyperpigmented macules on the neck and inside the ears. The nails of the right big toe and index fingers were absent

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and the left big toe nail was dystrophic. Teeth were brownish yellow probably due to the antibiotic therapy during infancy.

SYSTEMIC EXAMINATION did not reveal any abnormality. X-rays of lower extremities were normal. No evidence of prophyria was detected.

In view of the history given by the mother and the physical findings we felt this was a case of Aplasia Cutis Congenita associated with blisters. Plastic surgeon was consulted regarding the non-healing ulcer on the right knee.

COURSE IN HOSPITAL

Skin grafting was done, donor site being the thigh. The graft did not take. It was repeated using the skin from the opposite thigh. This time the graft took well and healed rapidly. However, the donor sites took long time to heal under the dressing. The dressing were hence discontinued and the donor sites healed rapidly on open treatment.

The first blister while in hospital was noticed 5 weeks after admission under the blood pressure cuff applied during surgery. Subsequently he developed blisters under the surgical dressings (fig. 4 & 5) and elsewhere on the body apparently due to trauma. At this stage the diagnosis was reconsidered. Biopsy of one of the vesicles showed subepidermal blister with intact basement membrane at the floor of the vesicle (fig. 6). Biopsy of the scar over the ankle showed flattened epithelium with increased collagen, few hair follicles, and an occasional sweat duct in the dermis (fig. 7 & 8).

Based on these histological findings, a diagnosis of epidermolysis bullosum simplex was made on this patient.

COMMENT

Review of literature on aplasia cutis congenita showed that these could be divided into two different groups :

GROUP 1. Showed defects at birth without subsequent development of blisters. Majority in this group presented defects only on the scalp[1-3]. and only a few on the rest of the body [9-11].

GROUP 3. Showed defects at birth and subsequently developed blisters in the defects and elsewhere on the body [12], [13]. The defects were noted mainly on the extremities. In this group some of the cases have been given additional diagnosis of epidermolysis bullosum due to the fact that they continued to develop blisters after birth indistinguishable from usual cases of epidermolysis bullosum. The diagnosis of two genetic disorders in one case become unnecessary if the defects noted at birth are recognised as prenatal lesions of epider molysis bullosum due to prenatal trauma. Further it is important to remember that aplasia cutis congenita is a developmental defect and as such the histological examination of the involved site should not reveal any skin appendages, the presence of which indicates that the defects were post-developmental caused by some type of trauma in utero.

Even though it is unusual to see cases of epidermolysis bullosum born with denuded areas of skin such cases have been reported 14–16. In all these cases blisters were also present at the same time. Our patient was born with large denuded areas on the extremities with no evidence of blister formation either in the denuded areas or elsewhere on the body upto the age of one year. This certainly brings in the possibility of aplasia cutis congenita as a diagnosis. When the patient developed blisters while under our care at sites of minor trauma. We considered the diagnosis of epidermolysis bullosum which was confirmed by histopathology.

The purpose of this paper is to point out the problems in making a diagnosis in a case like ours. We made a diagnosis of aplasia cutis congenita at first on our case based on the history of symmetrical defects of skin on all four extremities at birth. The develoment of blisters made us wonder whether this was a case of aplasia cutis congenita or epidermolysis bullosum with an unusual onset. The histopathological features of the scar and the vesicle favour the latter possibility. The presence of intact basement membrane at the floor of the vesicle makes the simplex variety of epidermolysis bullosum. Like our case, some of the cases reported in the literature as aplasia cutis congenita are probably cases of epidermolysis bullosum.

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