The 'CHILD' Syndrome (Congenital Hemidysplasia with Ichthyosiform erythroderma and Limb Defects)- a case report

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ABSTRACT

The term 'CHILD' is proposed as an acronym for Congenital Hemidysplasia with Ichthyosiform erythroderma and Limb Defects. The syndrome is characterized by unilateral erythema and scaling, with distinct demarcation in the middle of the trunk. The dermatosis is either present at birth or develops during the first week of life. Ipsilateral limb defects may vary from hypoplasia of some fingers to complete absence of an extremity. A ratio of female to male is 19:1. Apparently, the CHILD Syndrome is genetically determined. Arguments are put forth in favour of the hypothesis that the condition is due to an X-linked dominant gene lethal in homozygous males.

Key Words: CHILD syndrome, congenital hemidysplasia, skin defects, ichthyosiform erythroderma, X chromosome dominant inheritance

INTRODUCTION

Congenital Hemidysplasia with Ichthyosiform erythroderma and limb defects (CHILD Syndrome) is a rare congenital disorder. It is characterized by Hemidysplasia, ipsilateral erythroderma with limb and organ defects. It is caused by an X-linked dominant mutation in the NSDHL gene.[1,2] The mutations are lethal in homozygous male. In 1963, Rossman et al, described a female patient with previous unreported finding of unilateral Ichthyosiform erythroderma in which the patient had hypoplastic limbs and a malformed kidney on the same side of the body as the dermatosis.[3]

CASE REPORT

A two months old girl, the third child of a non-consanguineous marriage, presented with deformity of left lower limb and missing left upper limb along with redness and scaling over the same side of the body since birth. The delivery was normal and the child did not show any signs suggestive of systemic involvement. Her birth weight was 2.5kg. The mother had no history of miscarriage and the elder brother and sister were normal.

Cutaneous examination revealed a strikingly unilateral erythematous dermatosis present over the left side of the trunk to the midline anteriorly and posteriorly. Involvement extended to the left leg and was confluent in all areas. The border between the involved and uninvolved skin could be ascertained by palpation. The affected skin was intensely bright red in colour and blanched minimally on firm pressure. The involved skin was covered with large, thin white scales, free at edges and attached primarily at their centres. The scales did not fall off the skin spontaneously, but could be dislodged without any difficulty by the examining finger. Removal of scales failed to cause any bleeding. The involved skin was dry and felt somewhat rough to touch. There was no tenderness or sensation of increase or decrease temperature in these areas. Mucosal involvement was absent. Routine laboratory investigations were within normal limits. Left upper limb was missing. Lower limb X-ray showed shortening of left femur. Further examination did not show any involvement of other organs such as eyes, brain, heart, lungs or kidneys. Histological examination revealed

marked hyperkeratosis and parakeratosis, a prominent stratum granulosum and a moderate amount of regular acanthosis of the epidermis. Dermal involvement was limited to telangiectasia and mild infiltration of lymphocytes, histiocites and fibroblasts. There was no prominent vacuolization, which is characteristic of congenital bullous Ichthyosiform erythroderma. Based upon the clinical, radiological and histological findings, the diagnosis of CHILD syndrome was made. The patient was managed conservatively with topical steroids and emollients. The patient lost upon follow up.

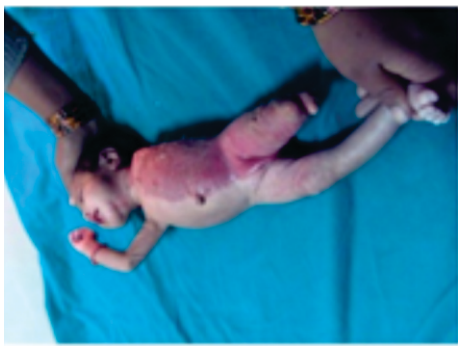

Fig.1. Deformity of left lower limb and missing left upper limb with midline demarcation of erythema and scaling.

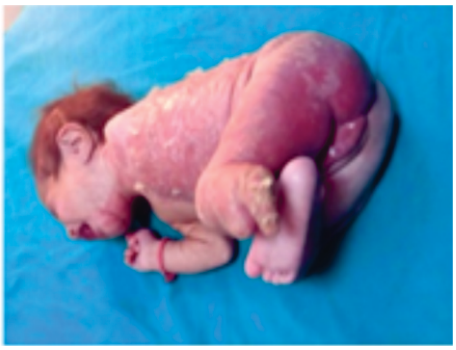

Fig.2. Large, thin white scales, free at edges and attached primarily at their centres.

DISCUSSION

The hallmark of the CHILD syndrome is the strict demarcation of the ichthyosiform erythroderma in the anterior and posterior midline of the trunk. In this patient, the clinical picture of a unilateral erythrodermic dermatosis, associated with ipsilateral limb deformities, is essentially identical to that of the patient reported by Rossman et al.[3] There were some dissimilarities in the salient findings between the patient reported by Rossman et al and this instant case. Though, the dissimilarities are primarily matters of degree. Their patient reported by Rossman was born with a hairlip, a malformed kidney, and more extensive limb aplasia and hypoplasia. The erythroderma extended to the midline posteriorly as in our patient. The cutaneous changes could be considered as unusual type of epithelial nevus. Zeligman and Pomeranz have described Histopathological changes in some epithelial nevi similar to the bullous type of congenital ichthyosiform erythroderma.[4] The erythroderma and desquamation of the skin in our patient does not, however resemble epithelial nevi. An ichthyosiform erythroderma may be noted in many conditions seen in the first week of life. These can be readily excluded, however, on clinical and histopathological ground. This case is being reported here because of its rarity; moreover right side of the body was involved in previously reported case, left side involvement is still rarer.

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