ICTHYOSIS BULLOSA OF SIEMENS A CASE REPORT

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Abstract:

A case of icthyosis bullosa, an extremely rare variant of epidermolytic hyperkeratosis is presented. The case is notable for it's characteristic clinical features and lack of response to treatment.

Introduction:

Icthyosis Bullosa of Siemens (IBS) is a rare inherited disorder of keratinization first described by Siemens in 1937[1]. However, the entity fell in to oblivion till it was rediscovered in 1986 by Traupe[2] who described a family affected by the condition. It has now been recognised as a extremely rare variant of bullous Icthyosiform erythroderma (epidermolytic hyperkeratosis, EHK), with certain characteristic distinguishing features[2,3]. We describe a patient with this rare condition, which, to the best of our knowledge, is the first case from the gulf region.

Case Report:

A male boy A.K. aged one and a half years, presented to the skin clinic at Nahdha Hospital, the main tertiary care referral centre in the Sultanate of Oman, with history of recurrent blisters, of 15 months duration. He was born of full term normal delivery to parents of consanguinous marriage (first cousins). The bullae first started at 3 months of age and occurred on legs, thighs, buttocks and back. The lesions were recurrent, and were precipitated by trauma, though at times occurred spontaneously. There was no history of preceeding erythema. The lesions healed with mild pigmentation, but without any scarring. There were no systemic complaints. There was no significant family history.

Examination showed a moderately built child 1 1/2 years with weight 12 kgs and height 60 cms. General examination was within normal limits. Cutaneous examination showed multiple bullae and erosions on buttocks, back, upper thighs, without any erythema of surrounding skin. There were areas with rumpled, brownish, hyperkeratosis on the buttocks thighs and knees (mauserung phenomenon) (Fig-1). There were few patches of pigmentation on back and buttocks, but no scar formation. Palm, soles, scalp, hairs, nails and mucosae were all within normal limits. At this stage, clinical differential diagnosis of epidermolysis bullosa and epidermolytic hyperkeratosis were considered.

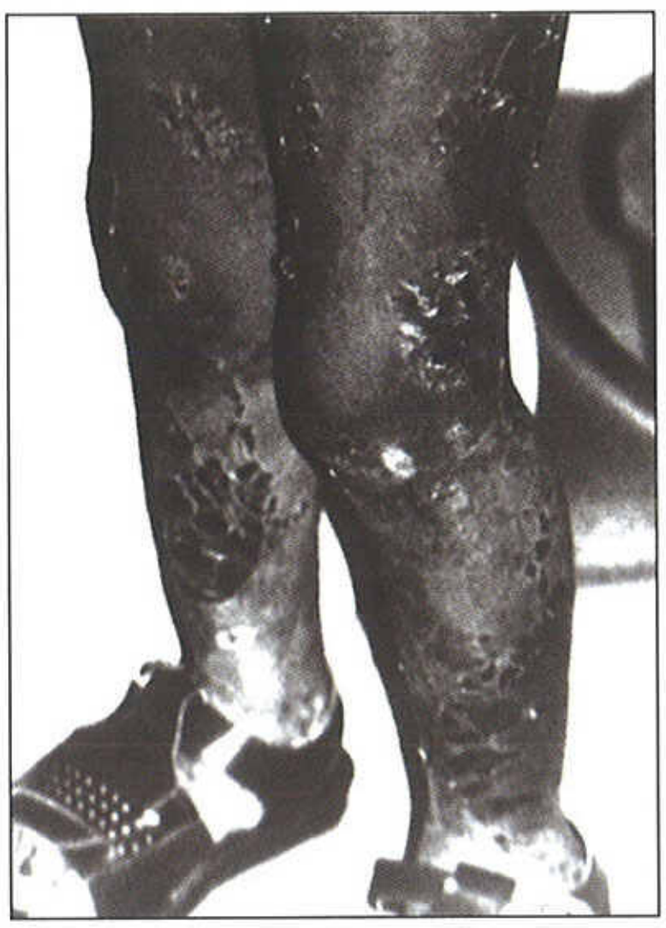

Fig. 1: Clinical photograph of patient showing mauserung (maulting) phenomenon on thigh and knees

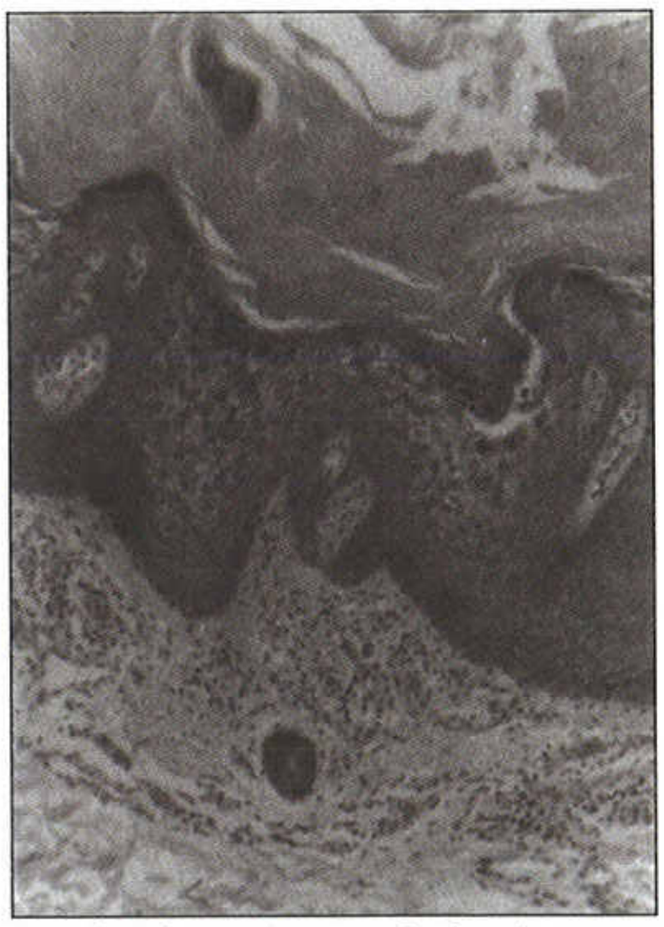

Fig. 2: Photomicrograph showing changes of hyperkeratosis, hypergranulosis, vacuolar degeneration in str.malphigi (20X)

Fig. 3: Electron micrograph showing clumping of tonofilaments in basal layer (24,000X)

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Routine investigations were within normal limits. Biopsy from lesion on buttock showed hyperkeratosis, focal vertical parakeratosis and intracorneal vesicles. There was mild hypergranulosis with large keratohyaline granules. Str.malphighi showed vacuolar degeneration (fig.2). Dermis showed a mild superficial perivascular infiltrate of round cells. These histological features were suggestive of epidermolytic hyperkeratosis.

Electron microscopic examination showed clumping of tonofilaments in stratum malphigi, with many intracellular vacuoles, confirming the diagnosis of epidermolytic hyperkeratosis (fig.3).

In view of the above mentioned clinical features of mild blistering with onset in infancy, absence of severe erythema, presence of rumpled hyperkeratosis over buttocks typical of mauserung phenomenon, histological evidence of epidermolytic hyperkeratosis, the case was diagnosed as Icthyosis Bullosa of Siemens. Patients was managed initially with etretinate 0.5 mg/kg for three months. There was no significant benefit and hence the dosage was raised to 1 mg/kg for three months. However, the drug did not bring about any improvement in the patient's condition and was discontinued.

The child has been under follow up for 3 years. The condition has remained mild, though there is an exacerbation in summer and improvement in winter.

Discussion:

Icthyosis Bullosa of Siemens, first described by Siemens[1] and subsequently by Heiko Traupe in 1986[2], is characterised by the following features:

1. Autosomal Dominant inheritance
2. Onset at birth
3. Easy bruisability and bullae formation after minor trauma
4. Hyperkeratosis, lichenification over knees, elbows, and around umbilicus. The hyperkeratosis is characteristically rumpled and is referred to as moulting or mauserung phenomenon.
5. Sparing of palms and soles
6. Seasonal variation, with improvement in winter
7. Spontaneous improvement in adolescence
8. Histologically, intracorneal and subcorneal localization of blister with mild changes of epidermolytic hyperkeratosis.

Though it's nosology was previously considered debatable, it is now recognised as a variant of epidermolytic hyperkeratosis and cases of both conditions have been described in the same family[4]. However, it differs from classic epidermolytic Hyperkeratosis of Brocq in the following clinical features:

1. Absence of significant erythema and milder clinical course.
2. Presence of superficial blisters similar to epidermolysis bullosa simplex
3. Distribution over buttocks, arms, legs, flexures
4. Mauserung phenomenon
5. Milder histological features, with intracorneal localization of blister and changes of EHK limited to upper stratum malphighi.
6. Improvement in adolescence

Electron microscopy shows the characteristic clumping of tonofilaments in stratum malphighi. Steijellen reported occurrence of subcorneal pusules in a case of IBS, suggesting the genetic hetorogenity of EHK[3]. Classic EHK has now been recognised to be due to mutation of gene leading to abnormality of keratins K5-16. In IBS, mutations in the keratin 2e gene has been reported[5].

Treatment of the condition is unsatisfactory. Etretinate has been tried with some benefit[6]. However, in our patient, administration of etretinate did not bring about any change in the condition.

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