Epidermolytic Hyperkeratosis with Genuvalgum and Genu Recurvatum

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I. Introduction

• EPIDERMOLYTIC hyperkeratosis also known as bullous ichthyosiform erythroderma, inherited as autosomal dominant trait mostly.
• Mutation of k1/K10 is the cause.
• Prevalence is 1 in 2,00,000 to 3,00,000 population.
• HYPERKERATOSIS and vacuolar degeneration are main histological features.

Case report:

• A 5 years old girl presented with scaling of skin since 4 years, her mother reported that child was active and born by normal vaginal delivery in hospital. No blisters at birth, no history of consanguinity, at age of 9th months few blisters developed at elbows, knees, ankles, wrists and buttocks, later subsided, then thick dry scales appeared on the body. No familial inheritance, girl is only child.

Cutaneous examination:

• Generalised dryness with hyperkeratotic plaques, most prominent over flexures, knees, elbows, ankles, neck, buttocks, wrist sparing face.
• Palms and soles are involved with keratoderma.
• Plaques has hystrix/cobblestone pattern.
• Mucosa, teeth, hair normal.
• Genu valgum, Genu recurvatum present.

INVESTIGATIONS, TREATMENT:

• Routine investigations like blood, urine, stool are normal. Radiograph of knees showed valgum, recurvatum.
• Skin biopsy showed acanthosis, HYPERKERATOSIS of epidermis, vacuolar degeneration of upper dermis.
• On these findings baby treating with white soft paraffin, oral calcium 1000mg in two divided doses, and advised to regular exposure to sunlight. Now baby is on followup.
II. Discussion:

- Here no H/O of cosagunity or familial inheritance and it is sporadic form.
- Palms and soles involved without fissuring and contractures corresponds to PS TYPE 1 of EPIDERMOLYTIC HYPERKERATOSIS.
- Usually Lamellar ichyosiform associated with Rickets and Genu valgum.

It is a rare presentation of sporadic EPIDERMOLYTIC HYPERKERATOSIS PS TYPE 1, associated with GENU VALGUM.

References: