CASE 1

A 14- year-old Thai boy from Petchaboon

Chief complaint:

Thickening of palms and soles for 10 years

Present illness:

The child has had a history of erythematous patches at flexure area since 4 months old. Ten years ago, his palms and soles became thicken. There were scaly plaques on flexure area of extremities, neck, and abdomen.

Then 2 years ago, the skin lesions extended to his entire chest wall and later the side of trunk.

Past history:

Unremarkable

Family history:

His mother also has thickening of palms and soles.

Physical examination:

There are symmetrical large well-demarcated erythematous scaly/hyperkeratotic plaques at the wrists, ankles, knees, elbows, antecubital fossae, popliteal fossae, anterior chest wall, abdomen, neck, and lateral aspects of trunk. There are dirty keratoderma of palms and soles as well as digital contracture.

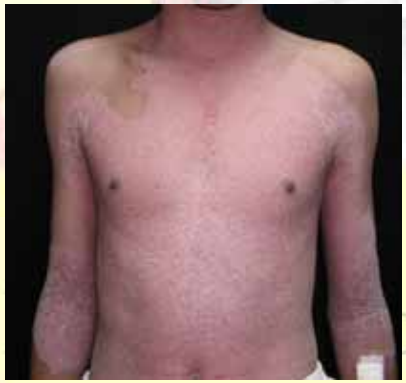

Fig. 1.1

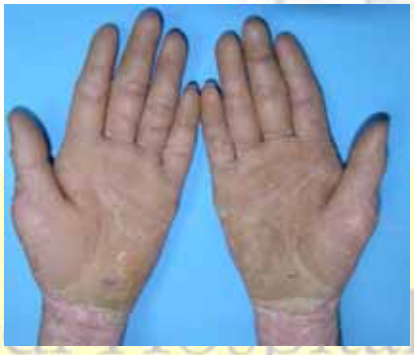

Fig. 1.2

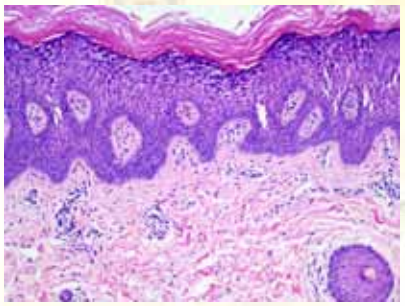

Fig. 1.3

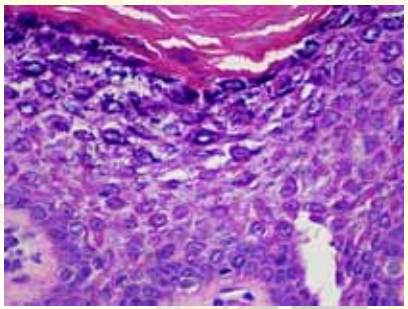

Fig. 1.4

Histopathology: (S05-17806)

Orthokeratotic hyperkeratosis and papillated epidermal hyperplasia

Hypergranulosis with coarse and fragmented keratohyaline granules

Epidermolysis with perinuclear vacuolization and indistinct cell boundary of granular and upper spinous layer

Diagnosis: Epidermolytic hyperkeratosis of Broq (Bullous ichthyosiform erythroderma)

Treatment: acitretin 10mg/day
20% urea and 5% lactic cream

Presesenter: Oraparn Techaritpitak

Consultant: Amornsri Chunharas
Suthep Jerasutus

Discussion:

Epidermolytic hyperkeratosis (EHK) is a rare disorder of keratinization, approximately 1 in every 100,000 – 300,000 live births will be affected.[1] The disorder has an autosomal dominant mode of inheritance due to mutations in highly conserved regions of either the keratin 1 or 10 gene.[2, 3] Keratins are expressed as intermediate filament protein pairs in a tissue-specific and differentiation specific fashion.[4] Errors in their formation may lead to clumped keratin filaments, keratinocyte fragility, and cytolysis.[5]

EHK manifests at birth with peeling, erosions and erythema, especially involving the flexural regions initially. Areas of denuded skin, as well as fissures in the intertrigenous areas may be present.[6] In adulthood, the disease is characterized by hyperkeratosis, predominantly over large flexural joint areas and on palms and soles. Overtime, skin fragility decreases while severe hyperkeratosis prevails. The clinical presentation may vary tremendously between patients and families. Variation of EHK has been described with at least 6 phenotypes. The most distinctive feature was presence (PS 1-3) or absence (NPS 1-3) of severe palmoplantar keratosis. Other distinguishing features included the presence or absence of erythroderma, quality of scale, extent of involvement, presence of digital contractures, and posture/gait abnormality.[1] The variety of amino acid changes which cause epidermolytic hyperkeratosis result in unique distortions of structural keratin proteins and therefore different clinical severity.[7]

The histology and ultrastructural findings are distinct as orthokeratosis hyperkeratosis, marked epidermal acanthosis, hypergranulosis with prominent vacuolar degeneration and dense clumped of keratohyaline granules.

The most common complication encountered is a foul smelling odor, produced by bacteria, when the macerated scales become infected.[8] soft tissue contractures of the hands, due to hypertrophied plantar fascia, have been described.[9]

As there is no cure for EHK, management involves symptom reduction. Therapy is aimed at reducing hyperkeratosis, removing scale and softening the skin. Keratolytic creams and emollients containing urea, salicylic acid, and alpha hydroxyl acids are effective. Topical tretinoin and vitamin D preparations are effective but may cause skin irritation. For severe cases, systemic retinoids may be used.[3]

References:

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