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]

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CASE 2

A 57-year-old female from Supanburi

Chief complaint:

Intermittent rash at intertriginous area since she was 37 years old.

Present illness:

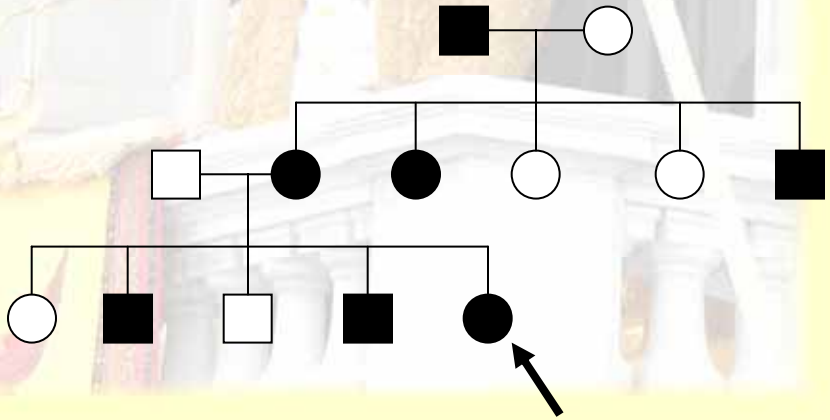
The patient had recurrent vesicles and erosions at neck, both axillae, both inframammary folds for 20 years. She experienced burning, itching and malodorous skin occasionally. The skin lesions get worse on summer months, wearing tight garments or friction. The lesions improved by topical corticosteroid application.

Past history:

Adrenal insufficiency confirmed by ACTH stimulation test due to prolonged use of topical corticosteroid.

Family history:

Many members in her family have similar skin lesions.



Physical examination:

Confluent well defined eroded erythematous plaques with fissured surface at inframammary area and extending to lateral side of abdominal skin bilaterally. Few flaccid vesicles are found at the periphery of the eroded plaques. Thin, easily bruising skin with visible vasculature is noted at chest.

Others -unremarkable

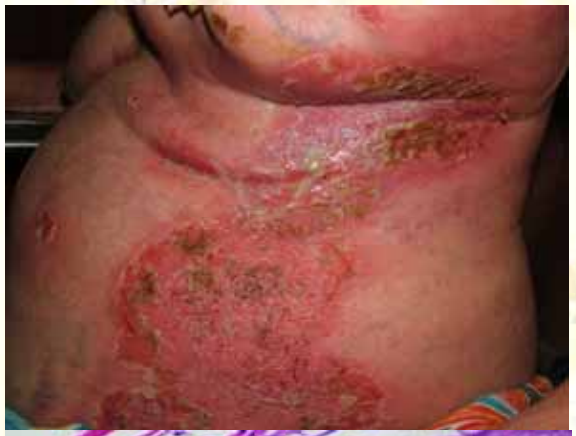


Fig. 1.1

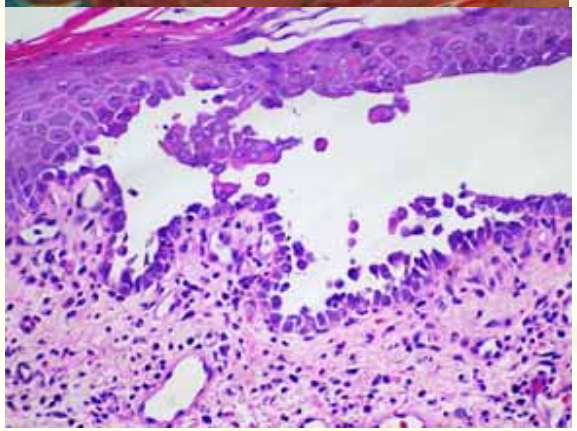


Fig. 1.2

Histopathology (S06-7614)

Suprabasal acantholytic blister in hyperplastic epidermis. Many acantholytic cells and acantholytic dyskeratotic cells in the blister.

Diagnosis: Hailey-Hailey disease
Presenter: Vasanop Vachiramon
Consultant: Siripen Puavilai
Discussion: