

### Case 10

A 18-year-old Thai woman from Saraburi

**Chief complaint:** had itchy rash on scalp and face for 10 years.

**Present illness:**

The patient had dry itchy rash on her scalp and face for 10 years. Then the rash gradually developed at her flexural area.

**Personal history :** normal

**Family history:** none

**Skin examination**

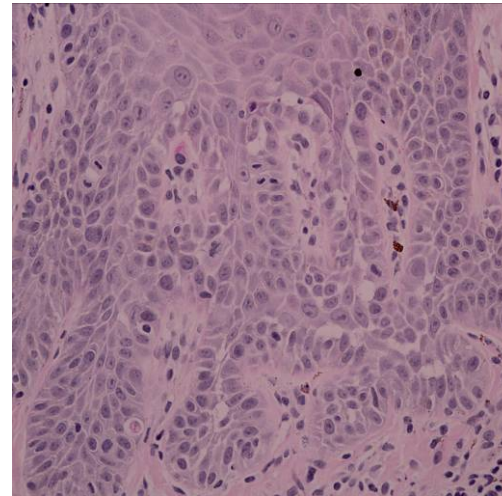
-Multiple discrete well defined erythematous scaly plaque on forehead, eyebrows, nose, infraorbital area, neck and inframamillary area

-multiple discrete skin-colored hyperkeratotic papules on dorsum of fingernails

-Multiple red and white longitudinal streak on finger nails.

-Normal oral mucosa ,no punctuated keratosis of palm and sole

**Histopathology** (S10-5291) hyperkeratosis and papillate epidermal hyperplasia with focal acantholytic dyskeratosis



**Investigation:**

CBC: WBC 6900 N=73% L=21%, Hb 7.8, Hct 25, MCV 65

**Diagnosis:** Darier's disease

**Treatment:** Emollient, topical steroid, Acitretin 10 mg daily

**Presenter:** Ornkes Panyanetinad

**Consultant:** Sutthinun Wichyanrat

**Discussion:**

Darier's disease is an autosomal dominant acantholytic disorders of the skin. It is classically characterized by skin-colored to brown greasy keratotic papules on seborrheic area, punctuated keratosis of palms and soles, leukoplakia of mucosa, and polydactylous longitudinal erythronychia with V-shaped nick.<sup>1</sup>

The pathogenesis is the mutation of ATP2A2 which encodes the sarco/endoplasmic reticulum Ca(2+)-ATPase isoform 2 (SERCA2). SERCA2 is a calcium pump of the endoplasmic reticulum (ER) transporting Ca(2+) from the cytosol to the lumen of ER. This leads to abnormal cell adhesion and keratinization<sup>2</sup>

Onset of Darier's disease is generally around in the puberty. However, its prevalence differs from place to place<sup>3, 4</sup>. Besides classical presentation, various forms of diseases were also reported. They included erosive form, bullous form, vegetating form, papulovesicular form, comedone-liked form, and hemorrhagic macule on palms and soles. Associated manifestations include neuropsychiatric abnormalities, abnormal kidney, corneal opacity, and multiple café-au-lait macules.<sup>5-7</sup>

Histopathology of Darier's disease composes of dyskeratosis resulting in corps ronds and grains, suprabasal acantholysis causing suprabasal clefts and also papillomatosis, acanthosis, and hyperkeratosis.<sup>8</sup>

Basic care of Darier's disease patients includes sun protection, heat avoidance and appropriate emollients. Topical vitamin A derivatives can be used to reduce hyperkeratosis. Both topical tacrolimus and pimecrolimus were reported being used<sup>9</sup>.

Oral retinoids are effective in severe cases. Many successful cases treated by ablative or non-ablative laser were reported<sup>10-12</sup>. Gene therapy is promisingly being developed<sup>13</sup>.

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