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 inframammary area
- Multiple discrete skin-colored hyperkeratotic papules on dorsum of fingernails
- Multiple red and white longitudinal streak on fingernails.
- 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 polydactylyous longitudinal erythronychia with V-shaped nick.1 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 keratinization2
Onset of Darier’s disease is generally around in the puberty. However, its prevalence differs from place to place3, 4. 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. 5-7
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. 8
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. 9
Oral retinoids are effective in severe cases. Many successful cases treated by ablative or non-ablative laser were reported10-12. Gene therapy is promisingly being developed. 13

References