CASE 12

A 12 -year-old Thai girl from Phetchabun

Chief complaint:

Asymptomatic multiple small papules on the face, neck and both axillae

Present History:

The patient presented with a 4 year history of multiple non-pruritic brownish hyperkeratotic papules at the face, neck and both axillae. She felt that the rashes worsened and became smelly with heat and sweating. She came to Ramathibodi hospital 3 years ago with this problem. The biopsy was done and showed only superficial infiltrate of melanophages. She was diagnosed and treated as atopic eczema with slight improvement.

Past history:

She was previously healthy.

Family history:

She has one sister. No one in her family has similar skin lesions.

Physical examination:

Skin: multiple hyperkeratotic follicular brownish papules at face, neck and both axillae.

Fingernails: alternate longitudinal red and white ridges V-shaped nicked at nail edge

Oral mucosa: no lesion

Histopathology: (S06-08244)

Basket weave orthokeratosis and papulomatosis

Foci of suprabasilar clefts with acantholytic, dyskeratotic cells in the overlying epidermis including spinous, granular and cornified layer

Diagnosis : Darier's disease

Presenter : Pranee Wongkitisopon

Consultant : Natta Rajatanavin

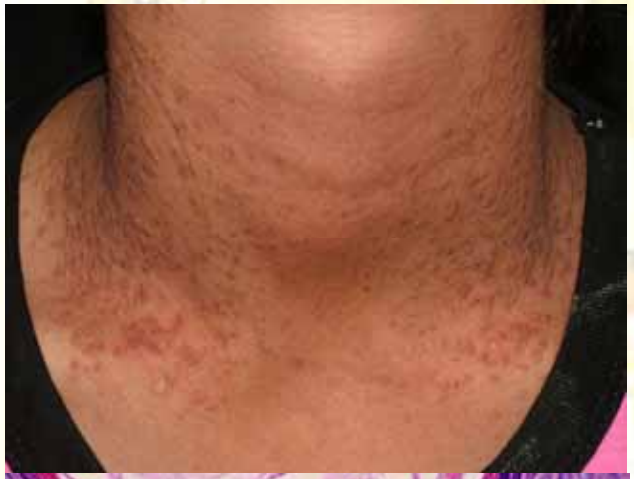

Fig. 12.1

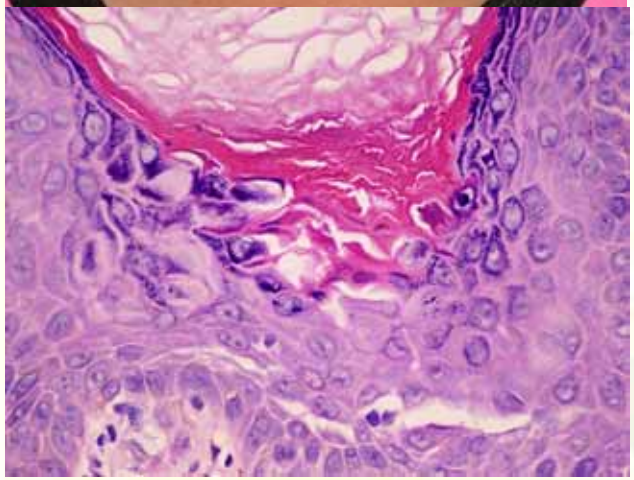

Fig. 12.2

Treatment: Advice the patient and her parents about the natural history and prognosis of the disease. The patient was given an antibacterial soap to reduce the odor and topical isotretinoin to control the disease.

Disscusion

Darier's disease is an autosomal dominant genodermatosis in which mutations in the ATP2A2 gene resulting in sarcoplasmic/endoplasmic reticulum Ca2+-ATP isoform 2 protein (SERCA2)[1,2] defect which leads to disrupted calcium homeostasis within cells. The most recent study found that the absence of coexpressed SERCA3[3] may explain the localization of the disease, which leads to loss of epidermal cell-to-cell adhesion and an abnormal keratinization. Histopathology revealed hyperkeratosis, acantholysis and dyskeratosis. The disease is characterized by hyperkeratotic, follicular papules distributed on the seborrheic areas and in the flexures, palmoplantar pits and variable nail dystrophies. There were 2 reports describing the esophageal involvement[4] in a patient with Darier's disease, however the complication includes secondary infection, serious HSV infection must be suspected when there is a sudden onset of painful vesicular lesion. Malignant transformation is rare. Recently retrospective review showed sun-exposure exacerbated the disease in half of patients[5] and there was a report of localized form restricted to sun-exposed areas[6] which may be due to inflammation induced by solar damage. Sunscreen and sun avoidance is essential. Now there are several modalities of treatment, the recent study in Asians showed topical steroids provided relief of itchyness, topical retinoids are more effective but irritation occurred. A report revealed successful treatment of Adapalene gel with minimal relapsed.[7,8] Systemic retinoids showed significant clinical improvement[9], however the use is limited by their side effects and costs. Surgical therapy includes dermabrasion, electrosurgery, and Mohs micrographic surgery. Carbon dioxide laser ablation of recalcitrant plaques has been reported in 2 patients. The Er:YAG laser, which in contrast to the carbon dioxide laser has decreased risk of thermal injury, was used in 2 patients. In those cases, remission of the treated lesions and pruritus lasted at least 11 and 20 monthS.[10] The family should be counseled about the risk of having affected offspring.

References

1. Burkhart CG, Bu Dhitavat J, Fairclough RJ, Hovnanian A, Burge SM: Calcium pumps and keratinocytes: lessons from Darier's disease and Hailey-Hailey disease. Br J Dermatol 2004 May; 150(5): 821-8.
2. Sakuntabhai A, Ruiz-Perez V, Carter S, et al: Mutations in ATP2A2, encoding a Ca2+ pump, cause Darier disease. Nat Genet 1999 Mar; 21(3): 271-7.
3. Tavadia S, Authi KS, Hodgins MB, Munro CS: Expression of the sarco/endoplasmic reticulum calcium ATPase type 2 and 3 isoforms in normal skin and Darier's disease. Br J Dermatol 2004 March; 151: 440-5.
4. Ahmed AR, Issam RH, Sultan K: Extensive Darier's disease with esophageal involvement. Int J Dermatol 2004 Nov; 43(11):835-9.
5. Otley CC, Momtaz K: Induction of Darier-White disease with UVB radiation in a clinically photo-insensitive patient. J Am Acad Dermatol 1996 May; 34(5 Pt 2): 931-4.
6. Kimoto M, Akiyama M, Matsuo I: Darier's disease restricted to sun-exposed areas. Clinical Dermaqtology 2004;29: 37-9.
7. English JC, Browne J, Halbach DP. Effective treatment of localized Darier's disease with Adapalene 0.1% gel. Cutis 1998; 63: 227-30.
8. Cianchini G, Colonna L, Camaioni D, Annessi G, Puddu P. Acral Darier's disease successfully treated with adapalene. Acta Derm Venereol 2001; 81:57-8.
9. Dicken CH, Bauer EA, Hazen PG, et al: Isotretinoin treatment of Darier's disease. J Am Acad Dermatol 1982 Apr; 6(4 Pt 2 Suppl): 721-6.
10. Beier C, Kaufmann R: Efficacy of erbium:YAG laser ablation in Darier's disease and Hailey-Hailey disease. Arch Dermatol 1999; 135(4):423-7.