Case 7

A 34-year-old woman

Chief complaint: Pruritus on both ears for 4-5 years

Present illness: She presented with pruritus on both ears for 4-5 years. She has never noticed any skin lesion, but the doctor noted multiple pruritic papules on her ears, as well as on several areas on her body. There is no family member with the same skin lesion.

Past history : nil

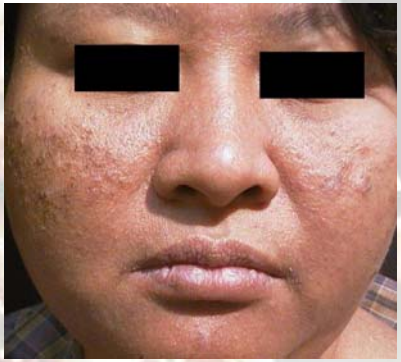
Skin examination:

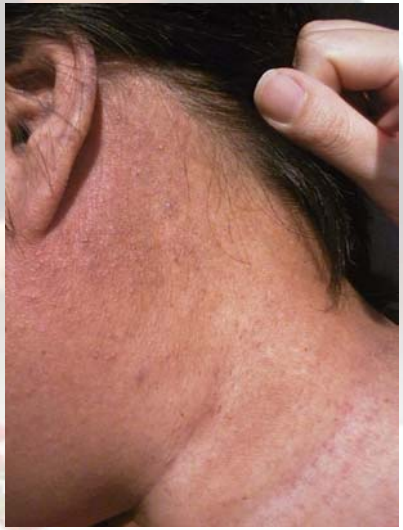
Skin: multiple discrete pruritic hyperkeratotic papules on face, neck, periauricular area, back, and upper chest.

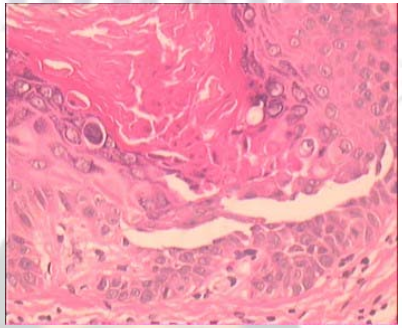
Palms: multiple pits.

Nails: multiple longitudinal red and white bands, V-shaped nicks.

Mucous membrane: normal







Histopathology: slide no. S00-17594

- focal suprabasalar clefts
- acantholytic, dyskeratotic cells in the spinous and granular zone, above clefts focal parakeratosis overlying the suprabasalar clefts

Diagnosis: Darier-White Disease

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Discussion

Darier-White Disease is an autosomal dominant disorder with abnormal keratinization of the epidermis, nails, and mucous membranes. However, many cases appear to occur as new mutations. Recently, ATP2A2, the defective gene which encodes the sarco/endoplasmic reticulum Ca(2+) ATPase isoform 2, was identified on the long arm of chromosome 12 within the region 12q23-24.1. The pathogenesis is unknown. The onset is usually in the first and second decades of life. Skin lesions appear as multiple, discrete, scaling, rough, crusted, pruritic papules, which are skin-colored or yellow-brown, on seborrheic areas. The palms and soles may show punctate or filiform keratoses or minute pits. Lesions of the mucous membranes are uncommon, but cobblestone papules on mucosa may be found. The nails are

usually abnormal. Nail changes include longitudinal ridges, red and white longitudinal bands ,and characteristic V-shaped scalloping and subungual thickening. Patients with Darier's disease appear to have an increased susceptibility to herpes simplex infections and an increased incidence of chronic pyogenic infection.

Reference

1. Sakuntabhai A, Dhitavat J, Burge S et al. Mosaicism for ATP2A2 mutations causes segmental Darier's disease. J Invest Dermatol 2000 Dec;115(6):1144-7
2. Powell J, Sakuntabhai A, James M et al. Grover's disease, despite histological similarity to Darier's disease, does not share an abnormality in the ATP2A2 gene. Br J Dermatol 2000 Sep;143(3):658.
3. Sakuntabhai A, Burge S, Monk S et al. Spectrum of novel ATP2A2 mutations in patients with Darier's disease. Hum Mol Genet 1999 Sep;8(9):1611-9.
4. 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.