GENETICS AND GENODERMATOSES

CLINICAL PRESENTATIONS OF DARIER DISEASE

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Background: Darier disease (DD) or dyskeratosis follicularis is an autosomal dominant skin disorder, which occasionally manifests in a segmental form. This genodermatosis is characterized by generalized brownish and pruritic papules, which are especially dense in the seborrheic areas of the body, nail abnormalities and mucous membrane changes. The disease typically begins in the first two decades of life and is often exacerbated by sun exposure and perspiration.
Two types of distributions have been described in segmental DD. Type 1 lesions follow Blaschko's lines unilaterally on a background of normal skin while type 2 lesions represent areas of increased severity superimposed on a background of generalized DD. A distinction between types is necessary for genetic counseling.

Observation: We report three cases of biopsy confirmed DD, each belonging to one of the three types of DD (Classical DD, Type 1 mosaic DD and type 2 mosaic DD)
Patient 1 Classical DD
A 42-year-old woman, with a family history of DD presented with generalized brownish pruritic papules mainly in seborrheic areas and nail abnormalities. She was diagnosed of non-segmental DD.
Patient 2 Type 1 Mosaic DD
A 33-year-old woman presented with recurrent brownish, unctuous papules following a linear C shape distribution over the right intermammary and inframammary region following Blaschko´s lines, no other lesions were seen elsewhere. Lesions got worst during the summer months but improved with isotretinoin during those periods.
Patient 3 Type 2 mosaic DD
A 47-year-old man with family history of DD and a long-time history of brownish, unctuous papules following lines of Blaschko over the left trunk, developed a generalized eruption with oral manifestations.

Key message: DD is a prototypic disease to show two different types of cutaneous mosaicism.