GENETICS AND GENODERMATOSES

THE CUTANEOUS MANIFESTATIONS OF DARIER DISEASE PROPOSING A NEW DISEASE CLASSIFICATION- A COHORT STUDY OF 76 PATIENTS.

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Introduction: Darier disease is an orphan autosomal-dominant genetic disorder, only a few studies have assessed the clinical findings in the disease.

Objective: To investigate the clinical features of a large group of patients with Darier Disease.

Materials and Methods: Seventy-six individuals affected with Darier Disease from 34 families were evaluated utilizing a structured, questionnaire-based interview followed by a physical examination. Medical information was also gathered by retrospective assessment of patient's medical records.

Results: Most patients (88%) exhibited a combination of disease patterns (flexural, seborrheic and acral). The most frequent locations of lesions were the hands (99%) and fingernails (93%). Oral mucosa involvement was found in 43% of patients, equally distributed between men and women. External genital lesions were more common in women (F-24%; M-8%). Patients with severe disease were younger at onset than patients with mild disease. (13.7±11.4 years vs. 18.6±10.0 years, P= 0.01).

Conclusions: Documentation of disease on the hands and fingernails provides a highly sensitive means to aid in the diagnosis of Darier disease. Classification of Darier Disease, as previously proposed, based on lesion location was found to be impractical since most patients exhibited a combination of the suggested patterns. Hence, we suggest a new

classification system based on the morphologic characteristics of lesions: Classical vs Non-Classical (Variants of Darier disease: acral keratoderma, leukodermic macules, giant comedones, kelloid-like vegetations, and acral hemorrhagic blisters). This system was easily applied in our cohort. (67% of patients had Classical Darier disease and 33% had Non-classical Darier disease.)