

GENETICS AND GENODERMATOSES

THE EXTRA-CUTANEOUS FINDINGS IN DARIER DISEASE- A COHORT STUDY OF 76 PATIENTS

A Yeshurun⁽¹⁾ - M Ziv⁽²⁾ - E Cohen-barak⁽²⁾ - S Vered⁽³⁾ - D Rozenman⁽²⁾ - M Sah⁽²⁾ - M Khayat⁽⁴⁾ - O Polyakov⁽⁵⁾ - E Adler⁽⁶⁾ - C Nadler⁽⁷⁾ - B Amichai⁽⁸⁾ - A Zlotogorsky⁽⁹⁾ - S Shalev⁽⁴⁾ - R Dodiuk-gad⁽²⁾

None, Bruce Rappaport Faculty Of Medicine, Technion, Haifa, Israel, Haifa, Israel⁽¹⁾ - None, Department Of Dermatology, Emek Medical Center, Afula, Israel, Afula, Israel⁽²⁾ - None, Department Of Statistics, Haifa University, Haifa, Israel, Haifa, Israel⁽³⁾ - None, Genetic Institute, Emek Medical Center, Afula, Israel, Afula, Israel⁽⁴⁾ - Home Front Command, Israel Defense Forces, None, Afula, Israel⁽⁵⁾ - None, Department Of Ophthalmology, Emek Medical Center, Afula, Israel, Afula, Israel⁽⁶⁾ - None, Oral Maxillofacial Imaging, Oral Medicine Department, Hebrew University, Hadassah School Of Dental Medicine, Jerusalem, Israel, Jerusalem, Israel⁽⁷⁾ - None, Dermatology Unit, Meir Medical Center, Tel Aviv, Israel⁽⁸⁾ - None, Department Of Dermatology, Hadassah – Hebrew University Medical Center, Jerusalem, Israel, Jerusalem, Israel⁽⁹⁾

Introduction: Extra-cutaneous morbidities including learning disabilities and neuropsychiatric disorders have been reported in Darier disease (DD), an orphan autosomal-dominant genetic disorder. Systemic findings have not been fully characterized.

Objective: To investigate systemic findings in a large group of patients with Darier disease.

Materials and Methods: Thirty-four families comprising 76 individuals affected with DD were evaluated by detailed questionnaires and physical examination. Medical information was also gathered by retrospective assessment of patient's medical records.

Results: Systemic findings included recurrent salivary gland obstruction in 9 patients (12%), thyroid disease in 9 patients (12%), and ophthalmic pathologies in 26 patients (34%). Significant variation in the prevalence and severity of these disorders was found within and among families.

Conclusions: Darier disease has important extra-cutaneous findings that vary within and between families. Hence, it is important to conduct proper systemic assessment in patients with Darier disease and to include this information in the genetic counseling of families.