



GENETICS AND GENODERMATOSES

DARIER DISEASE WITH ESOPHAGEAL INVOLVEMENT: A RARE LOCALIZATION WITH NEOPLASTIC POTENTIAL

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Background: Darier disease is an autosomal dominant genodermatosis. It is characterized by the presence of keratotic papules arranged in the seborrheic areas of the body, nail dystrophy and palmar pits. Mucous membranes are uncommonly involved with white centrally depressed papules and often with a cobblestone appearance. Darier disease has been rarely observed with esophageal localization.

Observation: a 61-year-old patient affected by Darier disease referred to our clinic for a persistent symptom of pyrosis involving the esophageal tract. An esophagogastroduodenoscopy was performed, disclosing multiple papular lesions with a cobblestone appearance of the esophageal mucosa. Different biopsies were performed and the histopathological features were compatible with the diagnosis of Darier disease.

Key message: The mucosal lesions of Darier disease are uncommon, with an incidence varying from 15% to 50%. Only few cases with esophageal involvement have been described in the literature. Moreover it has been reported a case with Darier disease of the esophagus developing a squamous cell carcinoma. The common molecular alterations in squamous cell carcinoma and Darier disease may be related for a shared pathogenetic mechanism

