GENETICS AND GENODERMATOSES

ATYPICAL PRESENTATION OF DARIER DISEASE

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Background: Darier disease, a rare autosomal dominant genodermatosis with variable phenotypic expression, is characterized by a persistent eruption of red-brown keratotic papules in a seborrheic distribution, pitting of palms and soles, nail abnormalities and mucosal changes. Typically, the lesions appear between the ages of 6 and 20 years. Oral mucosal involvement is found in approximately 50% of the cases. However, Darier disease confined to the oral cavity without cutaneous manifestations have been reported in one previous case only according to our knowledge.

Observation: A 34-year-old woman presented for whitish lesions on oral mucosa associated with fissured, crusted and enlarged lips since the age of 30. Total gingival hypertrophy was also observed. The remainder of the physical examination was unremarkable. She was treated by topical corticosteroids without satisfactory improvement. The family history was not contributory. A biopsy of the oral mucosa showed focal dyskeratosis with formation of basal corps round and superficial grains consistent with the diagnosis of Darier disease. Oral isotretinoin 20 mg per day was given for a total duration of 8 months with significant improvement. Patient was followed over a period of 6 months with no signs of recurrence.

Key message: We report a rare and atypical case presentation of intraoral Darier disease successfully treated with isotretinoin.