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**11** | **Fordyce granules and hyperplastic mucosal sebaceous glands as distinctive stigmata in Muir-Torre syndrome patients: characterization with reflectance confocal microscopy.**

**ABSTRACT**

**BACKGROUND:** The Muir-Torre syndrome (MTS), a variant of Lynch syndrome (LS), is characterized by the presence of sebaceous skin adenomas and/or carcinomas and keratoacanthomas associated with visceral malignancies. Fordyce granules (FGs) are oral mucosal lesions previously found in association with LS. The aim of this study was to analyze the specific frequency of FGs in sporadic individuals and gene carriers patients with MTS of known mismatch repair genes mutations. The secondary aim was to characterize FGs by means of reflectance confocal microscopy (RCM).  **METHODS:** A total of 13 patients belonging to nine different genetically unrelated MTS kindreds (MLH1 gene mutation n = 2; MSH2 gene mutation n = 11) and 140 genetically unrelated healthy controls were examined. Depending on the clinical examination of the oral mucosa surface, subjects were categorized as either FGs positive or FGs negative.  **RESULTS:** FGs were diagnosed in 13 of 13 (100%) of MMR gene carriers patients with MTS vs. 9 of 140 (6.4%) controls. The most common site for FGs in MTS was the vestibular oral mucosa, compared with the gingival mandibular and retromandibular pad in controls. RCM examination found multiple sebaceous acinar cells that appear as round or oval hyper-refractive globules and that create a lobular aspects of the sebaceous glands defined as 'moruliform' or 'berry-like' structures.

**CONCLUSIONS:** Clinical and RCM evidences of our study suggest that an activation of the sebaceous glands system occurs in patients with MTS. Fordyce granules and intra-oral sebaceous hyperplasia may constitute an additional clinical parameter, which may be adopted to distinguish individuals with highest likelihood of being affected from MTS.