GENETICS AND GENODERMATOSES

A CASE OF MUIR-TORRE SYNDROME: THE EMPHASIS ON THE ROLE OF IMMUNOHISTOCHEMICAL STUDY

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Background: Muir-Torre syndrome (MTS) is a rare autosomal dominant genodermatosis associated with mutations in the DNA mismatch repair (MMR) genes. It is characterized by the presence of sebaceous neoplasm with or without keratoacanthoma (KA), and at least one internal malignancy mainly affecting the colon, rectum and urogenital tract. We report a case of MTS with sebaceous adenoma in a patient who had history of KA and colon cancer.

Observation: A 70-year-old male presented with a solitary, skin-colored nodule with central umbilication on left chin for 3 months. Histopathologic examination showed irregular sebaceous lobules composed of basaloid cells at the periphery and mature sebocytes in central, consistent with sebaceous adenoma. Immunohistochemical (IHC) staining reveals positive for MSH-2, MSH-6 and negative for MLH-1. The patient had a history of both hemicolectomy for colon cancer in 1992, and squamous cell carcinoma on right perioral area and KA on scalp have widely excised in 2002 and 2011, respectively. With the presence of sebaceous adenoma with lack of expression of MLH-1, history of KA and visceral malignancy of colon, we diagnosed with MTS.

Key message: We herein report an educational case of MTS to consider screening for visceral malignancy in case of diagnosis with sebaceous neoplasm. Furthermore, IHC staining is a useful and easily accessible method for diagnosis of MTS by confirming the absence of MMR gene expressions in patients with sebaceous neoplasm.