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IMAGE | COLON

# Muir-Torre Syndrome, a Rare Phenotype of Hereditary Nonpolyposis Colorectal Cancer With Cutaneous Manifestations

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## CASE REPORT

A 67-year-old woman presented to her dermatologist with 3 newly developed skin lesions on the face and upper back (Figure 1). Her medical history was significant for endometrial cancer treated with hysterectomy at age of 51 years, and her family history was positive for bladder cancer. The patient had no history of tobacco use or other significant risk factors. The lesions were removed and determined to be sebaceous adenomas by histopathology. The diagnosis of Muir-Torre syndrome (MTS) was suggested, and repeat colonoscopy was advised, despite having a normal routine colonoscopy 4 years prior. The colonoscopy showed a mass lesion in the proximal ascending colon, and biopsy of the specimen revealed high-grade, poorly differentiated, invasive adenocarcinoma (Figure 2). The patient underwent right hemicolectomy; at operation, no visceral metastases or lymph node involvement was identified. Immunohistochemical analysis of the lesions revealed normal expression of MLH1 and PMS2, with loss of expression of MSH2 and MSH6. Southern blot analysis of MSH6 revealed the presence of a 32-kb deletion involving exons 1–6.

MTS is a rare genodermatosis, defined by sebaceous neoplasms, keratoacanthomas, and visceral malignancies, most commonly genitourinary and colon cancer.[1] Sebaceous tumors are unusual neoplasms that generally appear on the face, scalp, and eyelids as yellow to brown papules or nodules, and histologically, as sebaceous lobules of various sizes that are incompletely differentiated, and contain basaloid cells at the periphery and mature sebaceous elements with cytoplasmic vacuoles in the center.[2]

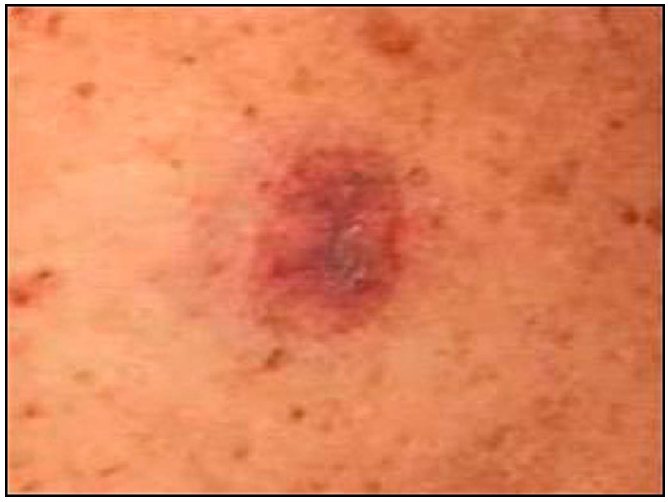

**Figure 1.** Sebaceous adenoma located on the upper back.

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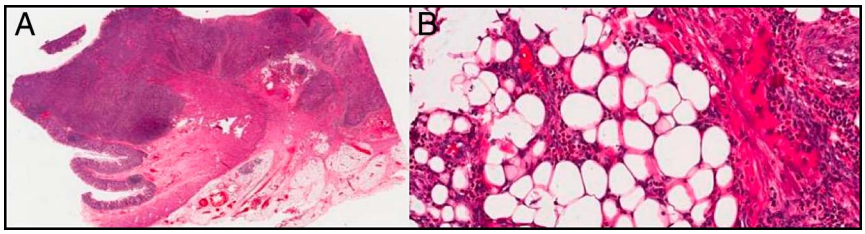


**Figure 2.** (A) Biopsy specimen showing high-grade, poorly differentiated, invasive adenocarcinoma. (B) The same specimen magnified.

Although necessary for the diagnosis of MTS, sebaceous neoplasms may occur decades before or after the diagnosis of a visceral malignancy, as displayed in our patient.[3]

Colorectal cancer is the most common visceral malignancy in patients with MTS. Lesions are typically located in the proximal colon and occur 10-20 years earlier than in the general population.[2] They tend to be of low grade and carry a relatively good prognosis, even in the presence of locoregional metastasis.[4] Tumors of the urogenital system (endometrium, ovary, bladder, kidney, and ureter) are diagnosed in about one-quarter of patients.[4] Malignancies of other sites, including the breast, parotid gland, upper gastrointestinal tract, brain, lung, and hematologic and neuroendocrine malignancies, have been reported.[2,3] In patients with MTS, it is suggested that screening colonoscopy start as early as age 18 years and be repeated every 2–3 years.[3] The finding of a high-grade neoplasm supports this 4 years after a normal colonoscopy in our patient.

## DISCLOSURES

Author contributions: D. Rubay, L. Ohanisian, and N. Kaur reviewed the literature and wrote and edited the manuscript. MP Bank wrote and edited the manuscript. T. Genuit and A. Ross revised the manuscript for intellectual content. D. Rubay is the article guarantor.

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