# Sebaceous neoplasia leading to the diagnosis of Muir-Torre syndrome in an African American man

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## INTRODUCTION

Most cases of Muir-Torre syndrome (MTS) have been reported in Caucasians from developed countries, with little data from other populations.[1] MTS, a variant of hereditary nonpolyposis colorectal cancer (HNPCC) or Lynch syndrome, is characterized by tumors seen in HNPCC, in addition to sebaceous tumors and keratoacanthomas. We describe the case of an African American patient who presented with a pruritic hypopigmented papule on his back. Further history and workup resulted in a diagnosis of MTS.

## CASE REPORT

A 71-year-old African American man presented with a pruritic growth on his right lateral back for 3 months. The growth was a 4-mm hypopigmented papule (Fig 1). A shave biopsy was performed, and histopathologic examination demonstrated a sebaceous adenoma (Fig 2). Further history obtained from the patient revealed that he underwent a left colon resection for numerous hyperplastic polyps and colon adenocarcinoma at age 60. The family history was significant for a brother who developed colon cancer in his fifties. Based on suspicion for MTS, additional immunohistochemical staining was obtained. MLH1 and PMS2 revealed no nuclear staining in lesion cells (abnormal), whereas MSH2 and MSH6 revealed intact nuclear staining. Tumor MLH1 promoter analysis revealed a hypomethylation, and testing for MLH1 confirmed a germline mutation, confirming the diagnosis of MTS.

*Abbreviations used:*

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| HNPCC: | hereditary nonpolyposis colorectal cancer |
| MMR: | mismatch repair |
| MTS: | Muir-Torre syndrome |

## DISCUSSION

MTS is a distinct variant of HNPCC or Lynch syndrome associated with keratoacanthomas and sebaceous neoplasms (adenoma, carcinoma, or epithelioma), with more than 200 cases reported worldwide.[2] Most cases have been reported in Caucasians from developed countries, with little data from other populations. A review of the literature revealed two cases of MTS in African Americans. Although many articles do not mention ethnicity, the rarity of cases in African Americans illustrates the importance of reporting ethnicity to determine the epidemiologic patterns of MTS.[3,4]

MTS is inherited in an autosomal dominant manner, is seen in 9.2% of patients with HNPCC, and has a male-to-female ratio of 3:2 (with an estimated incidence of HNPCC of 1:440).[2] The median age of onset of malignancy is 53 years, with the earliest reported case at 23 years and the oldest at 89 years. MTS is caused by a germline mutation in mismatch repair (MMR) genes leading to DNA replication errors leading to repetitive DNA sequences known as microsatellite instabilities, which drive tumorigenesis. MTS includes at least one internal malignancy (colorectal adenocarcinoma being

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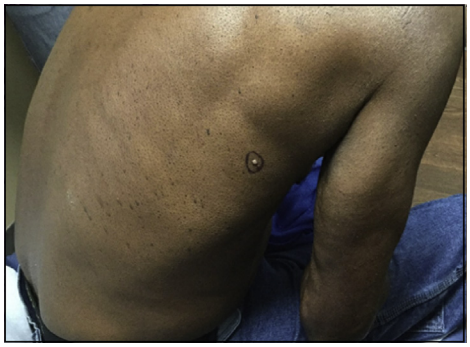

**Fig 1.** A 4-mm hypopigmented papule (circled).

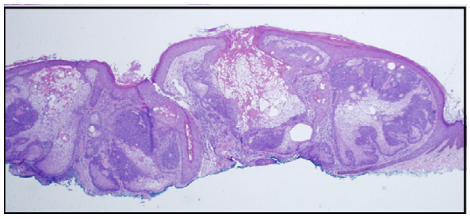

**Fig 2.** Well-demarcated proliferations displaying numerous lobules composed of sebaceous cells surrounded by basaloid cells.

the most common, but genitourinary, small bowel, breast, lung, brain, and hepatobiliary cancer having representation), plus one sebaceous tumor or keratoacanthoma.

Although sebaceous hyperplasia is a common finding in our aging population, sebaceous neoplasms, such as adenoma, epithelioma, and carcinoma, are much less common and are typically observed on the face.[5] Benign sebaceous neoplasms typically present as slow-growing, painless growths with or without a central ulceration. They are often associated with MTS, especially when occurring on the trunk and extremities. Our patient's sebaceous adenoma was hypopigmented (Fig 1). The diagnosis of a sebaceous neoplasm or multiple keratoacanthomas should elicit a thorough family and personal history of malignancy. If there is concern for MTS, the tumor should be subjected to immunohistochemistry staining for MMR gene products after patient consent is obtained, and the tumor should be resected with free margins.[5] Negative immunohistochemistry staining, indicating absence of MMR gene products, should prompt gene analysis for microsatellite instabilities, genetic counseling, and annual surveillance for internal and cutaneous malignancy.[5]

Our report demonstrates the importance of maintaining a broad differential diagnosis. Although MTS has been rarely reported in African American patients, it is difficult to ascertain whether this is because of a true low incidence or a reporting bias. Future work should focus on epidemiologic patterns of MTS and their effect on disease presentation and outcomes.

**Conflicts of interest**

None disclosed.

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