



MUIR-TORRE SYNDROME

<http://www.aocd.org>

Muir-Torre syndrome (MTS) is a rare inherited disorder that involves at least one sebaceous tumor and/or **keratoacanthoma** in addition to one visceral malignancy.

Sebaceous adenomas are the most common cutaneous tumors in MTS. They appear as multiple yellow papules or bumps on areas such as the trunk, face, and scalp. In some cases, only a solitary tumor may be present. Other sebaceous tumors include sebaceous epithelioma or sebaceous carcinoma which are advanced forms of sebaceous adenomas. Sebaceous carcinomas are especially concerning on the eyelid as they can invade the orbit and metastasize to nearby structures. Keratoacanthomas (KAs), on the other hand, are less commonly associated with MTS. They can occur as single or multiple dome-shaped nodules lined with blood vessels and a central keratin plug. KAs often appear on the face but can appear anywhere on the body. The presence of any such lesions should alert your dermatologist to the possibility of a visceral malignancy.

Muir-Torre syndrome is an autosomal dominant disorder due to mutations in the DNA repair genes hMSH2 and hMLH1. Although the mutation is passed from one generation to the next, the severity of MTS expression varies among family members. If suspicious lesions are present and/or a strong family history of colorectal cancer exists, the dermatologist will gather tissue samples for immunohistochemical staining. Loss of such genes on immunohistochemical analysis may prompt additional genetic testing for confirmation of MTS.

Colorectal cancer accounts for more than half of all visceral cancers occurring in MTS followed by genitourinary cancer at 25%. Other malignancies associated with MTS include breast, lung, gastric, small intestine, and hematological cancers. In a majority of cases, the sebaceous tumors appear after the onset of internal malignancy but can occur before or concurrently with the internal tumors. In spite of the risk of multi-factorial disease, the malignancies are low-grade and typically pursue a non-aggressive course.

Treatment for MTS consists of excisional removal of sebaceous tumors and KAs. Oral **isotretinoin** has been shown to prevent the development of further cutaneous tumors in MTS. Overall, treatment for MTS is multidisciplinary and genetic counseling and additional screening tests such as a colonoscopy, urinary test, and pelvic examination will be recommended for follow-up with the proper specialists.

This information has been provided to you compliments of the American Osteopathic College of Dermatology and your physician.

The medical information provided in this article is for educational purposes only and is the property of the American Osteopathic College of Dermatology. It is not intended nor implied to be a substitute for professional medical advice and shall not create a physician - patient relationship. If you have a specific question or concern about a skin lesion or disease, please consult a dermatologist. Any use, re-creation, dissemination, forwarding or copying of this information is strictly prohibited unless expressed written permission is given by the American Osteopathic College of Dermatology. For detailed information including links to related topics on this and many other skin conditions with photos, visit: <https://www.aocd.org/page/DiseaseDatabaseHome>