

## Muir-Torre Syndrome with Different Clinical Presentations

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**The presence of sebaceous gland neoplasm and visceral malignancy is a very rare condition.**

**We present two cases of Muir-Torre Syndrome (MTS) with different clinical presentations. The syndrome is associated with high incidence of multiple malignancies in various systems. One patient presented with a sebaceous cyst carcinoma and later with colonic neoplastic polyps while the other, after several colectomies for malignancy, developed a sebaceous cyst neoplasm. MTS is a hereditary disease; therefore, the family members may need to be counseled and screened, as early detection improves the overall prognosis and survival of the affected individual.**

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The presence of sebaceous gland neoplasm and visceral malignancy is a very rare condition<sup>1</sup>. This syndrome is associated with high incidence of multiple malignancies in various systems. Moreover, this is a hereditary disease, therefore, family members are advised to be screened as well. MTS deviations pathologically could be seen in the MSH2, MLH1, and MSH6 genes.

Cancer-associated genodermatoses, such as Muir-Torre syndrome are hereditary cancer syndromes associated with distinguishing cutaneous signs. Muir-Torre syndrome is an autosomal dominant syndrome caused by mutations in the mismatch repair genes and is thought to be a subtype of Lynch syndrome<sup>1,2</sup>. It is characterized by the presence of at least one sebaceous skin tumor (adenoma, epithelioma, carcinoma or keratoacanthoma) and a visceral malignancy, in the absence of other precipitating factors<sup>3,4</sup>.

MTS usually presents with one cutaneous neoplasm and at least one visceral malignancy. Cutaneous neoplasms are usually of sebaceous origin. Visceral malignancies could be of gastrointestinal or genitourinary origin.

The aim of this report is to present two rare cases of Muir-Torre Syndrome with different clinical presentations.

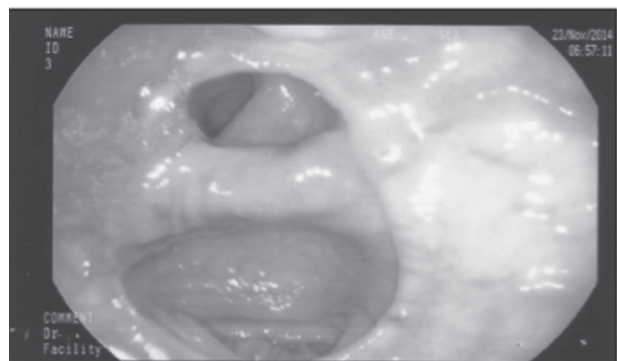
### THE CASE

#### Case 1

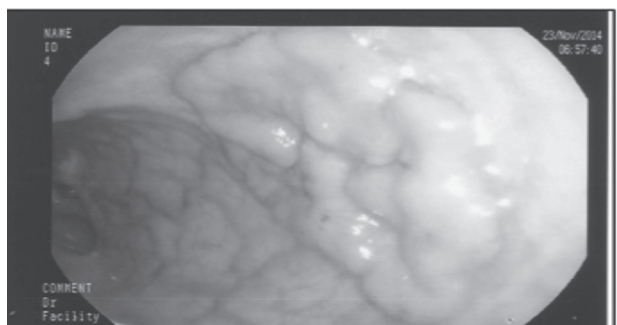
A sixty-three-year-old male who had multiple segmental colectomies for colonic adenocarcinoma (underwent pan proctocolectomy and ileorectal J-pouch anastomosis in 2006) presented in 2014 with a painful right axillary swelling which was progressively increasing in size. The swelling was initially noticed seven months prior to his presentation with purulent discharge. On clinical examination, there was a non-tender, 7 cm fungating mass with irregular borders at the right axilla, with ulcerations, oozing purulent foul smelling discharge. No palpable lymph nodes were felt in the axilla. On abdominal examination, a midline scar from previous surgery was seen.

On rectal examination, a mass of 2 cm from the anal verge was felt.

CT revealed a well-defined heterogeneous mass in the right lateral chest wall, abutting the pectoralis muscles with no evidence of invasion or intra-abdominal colonic cancer recurrence. Serum CEA was 0 µg/L. Colonoscopy revealed a sessile polyp 2 cm from the anal verge, see figure 1 (A and B). Excisional biopsy of the polyp revealed tubulovillous adenoma with focal high-grade dysplasia without invasion to the muscularis layer.



**Figure 1 (A): Colonoscopy Image of Ileoanal J-pouch Anastomosis**



**Figure 1 (B): Polyp 2 cm from the Anal Verge**

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Excisional biopsy of the chest wall mass was reported histopathologically as atypical sebaceous and basaloid cells with areas of necrosis, typical of sebaceous cell carcinoma, see figure 2 (A and B).

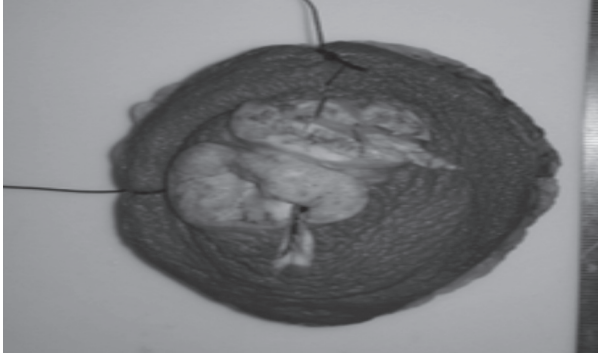


Figure 2 (A): Anterior View of the Axillary Mass Post-excision

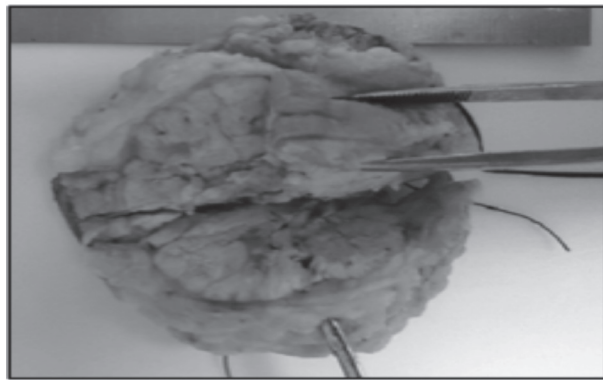


Figure 2 (B): Posterior View of the Axillary Mass Post-excision

The patient was referred to the genetics for assessment of genetic mutation and advised family screening.

### Case 2

A fifty-year-old male underwent excisional biopsies of multiple facial and scalp lesions since 2011. Histological findings were as follows: sebaceous cystadenoma, nodular sebaceous hyperplasia, sebaceous gland hyperplasia, sebaceous adenoma and sebaceous carcinoma. The findings raised the possibility of Muir-Torre syndrome. The patient had a colonoscopy in 2016, which revealed multiple benign colonic polyps, see figure 3. The patient was screened for other malignancies.

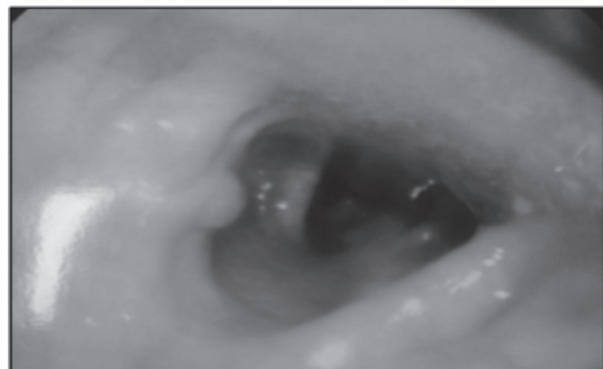


Figure 3: Colonoscopy Image of Colonic Polyp

### DISCUSSION

MTS was first described by Muir et al in 1967 and Torre in 1968, approximately 250 cases were reported since then<sup>5-8</sup>. The occurrence of multiple sebaceous adenomas with a primary ampulla of Vater carcinoma was first described by Torre, followed by a case report by Stewart et al in 1977 of a woman who had Bowen disease of the vulva, carcinoma of the rectum and a keratoacanthoma<sup>6</sup>.

The presence of one or more sebaceous gland neoplasms with visceral malignancies (including but not limited to colon, rectum and/or genitourinary tract) should raise the suspicion of MTS. Males are commonly affected with 3:2 ratio<sup>8,9</sup>. The mean age at presentation of sebaceous neoplasms is 53 years while that of visceral malignancy is 50 years<sup>7,10</sup>. Visceral malignancy is usually diagnosed after the appearance of sebaceous neoplasms in 22%, occur simultaneously in 6% and develop later in 56% of MTS<sup>7,10-12</sup>. Although the visceral malignancy is less aggressive, they are often more malignant and require close follow-up<sup>4,13</sup>.

MTS is inherited with a high degree of penetrance. A family history could be elicited in about 50% of affected patients<sup>14</sup>. Given the wide range of associated malignancy, screening for all possible cancer occurrences in an individual is difficult in MTS. Therefore, Cohen et al recommended a screening program that involves annual examination of the thyroid, breast, testicles and a digital rectal examination. CEA, chest radiography, urine cytology and mammography should be done yearly. Colonoscopy or barium enema along with endometrial biopsies should be done every 3 to 5 years<sup>15</sup>. Other authors have advocated annual colonoscopy from the age of 25 years, given the higher frequency of colonic cancer and its proximal predominance<sup>13</sup>.

Immunohistochemical staining for hMSH2 and hMLH1 could confirm the diagnosis after the clinical criteria have been met<sup>7,12</sup>. Patients diagnosed to have Muir-Torre syndrome should consider genetic counseling and targeted cancer surveillance.

### CONCLUSION

**MTS is a rare genetic disorder that affects more than one system with malignancy. Awareness of this disorder facilitates its early detection, thereby allowing for early treatment and better prognosis. Early screening for other family members is advised.**

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**Competing Interest:** None.

**Sponsorship:** None.

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