Sebaceous Carcinoma of the Abdominal Wall: A Potential Indicator of Muir–Torre Syndrome

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Introduction: *Sebaceous carcinoma is a rare dermatologic tumor affecting the pilosebaceous apparatus of the skin. While the majority of sebaceous carcinomas arise from sebaceous glands in the ocular area, extraocular sebaceous carcinomas, arising from any region populated with sebaceous glands have also been reported. Sebaceous carcinoma can present as a single lesion or in association with secondary malignancies, most commonly with those found in Muir–Torre syndrome (MTS), an autosomal dominant condition associated with several types of sebaceous neoplasms as well as a variety of visceral malignancies. The most common form of MTS has been described as a variant of hereditary non-polyposis colorectal cancer (Lynch syndrome).*

Patient profile: *Here, we describe the case of a 55-year-old male, with a known history of colorectal cancer, presenting with a rapidly enlarging abdominal wall mass.*

Interventions and outcomes: *Surgical excision of the mass histologically demonstrated sebaceous carcinoma. This diagnosis, the incidental discovery of a papillary thyroid carcinoma and the patient's history of colorectal cancer, prompted referral for genetic counseling, the results of which are still pending.*

Discussion: *Sebaceous carcinoma is one of several diagnostic criteria of MTS and its presence should prompt a complete evaluation for underlying internal malignancies.*

Keywords: *sebaceous gland; sebaceous carcinoma; abdominal wall; Muir–Torre syndrome; colorectal cancer; HNPCC.*

INTRODUCTION

Sebaceous glands, found most abundantly in the skin of the head and neck, are exocrine glands arising from the epidermis and epidermal appendages. These oil-producing glands secrete sebum, a mixture of lipids and cellular debris, into the hair follicle to reduce evaporation from the epidermal surface. The ocular region has a high density of sebaceous glands, including the modified Zeis glands of the cilia and meibomian glands of the eyelid.[1] While the head and neck are the most populated areas, sebaceous glands are found on any hair-bearing regions of the body.

Sebaceous carcinoma is a rare tumor affecting the sebaceous glands. These tumors are classified as ocular or extraocular, depending on the involvement of the eyelid structures. Seventy-five percent of sebaceous carcinomas are ocular, most commonly arising from the meibomian glands, while 25% are extraocular, arising from any region populated with sebaceous glands.[1] While sebaceous carcinoma can present as a single lesion, it is frequently associated with secondary malignancies, most commonly those found in Muir–Torre syndrome (MTS). MTS is an autosomal dominant condition associated with sebaceous neoplasms, keratoacanthomas and a variety of visceral malignancies, including colorectal, endometrial and urological.[2] While MTS can arise in individuals without a family history, it has a prominent familial association; the most common type of MTS is considered a variant of hereditary non-polyposis colorectal cancer (HNPCC, Lynch syndrome), a genetic condition characterized by defects in DNA mismatch repair genes. While both the MLH1 and MSH2 genes are mutated in HNPCC, mutations in MSH2 are more frequently reported in cases of MTS.[3,4] Sebaceous carcinoma is often considered a potential diagnostic sign of MTS[4] (Table 1), and its presence should prompt a complete evaluation for gastrointestinal and genitourinary cancers. Due to its inheritance pattern, relatives of patients diagnosed with MTS should also be examined for sebaceous and visceral malignancies.[5]

PATIENT PROFILE

The patient was a 55-year-old Caucasian male presenting with a 5-year history of a non-painful abdominal wall mass that had been rapidly increasing in size over the past year. He denied any erythema or drainage from the lesion; however, he did note a 10–20

Table 1. Diagnostic criteria for Muir–Torre syndrome

Group A
1) Sebaceous adenoma
2) Sebaceous epithelioma
3) Sebaceous carcinoma
4) Keratoacanthoma with sebaceous differentiation
Group B
1) Visceral malignancy
Group C
1) Multiple keratoacanthomas
2) Multiple visceral malignancies
3) Family history of Muir–Torre syndrome

Diagnosis requires one criterion from Group A and Group B, or all three from Group C.
From Ref. (4).

pound weight loss over the past year. The patient's past medical history was significant for colorectal cancer (T4N0) diagnosed approximately 16 years prior to the date of presentation. This was treated with subtotal colectomy and partial cystectomy along with adjuvant chemotherapy. A basal cell carcinoma of the nose and keratoacanthoma had been locally excised prior to presentation. The patient's father had a history of gallbladder cancer; however, the rest of the family history and social history were unremarkable. On physical examination of the neck, a 1 cm mobile, non-tender nodule was palpated at the angle of the left mandible; no goiter was noted. An 8×8 cm, mobile, non-tender firm mass was located over the right mid-abdomen; the mass elevated the skin but did not demonstrate any drainage or erythema. The abdomen was found to be soft, non-tender and non-distended.

INTERVENTIONS AND OUTCOME

The patient underwent an extensive diagnostic work-up to determine the source of the abdominal wall lesion. A core needle biopsy of the abdominal wall mass was performed, with pathology of the specimen demonstrating carcinoma with extensive necrosis, suggestive of an urothelial primary source. Due to the patient's history of colorectal carcinoma, a carcinoembryonic antigen (CEA) blood level and flexible sigmoidoscopy were also completed at this time, both with normal results. A CT scan of the abdomen (Fig. 1) demonstrated a solitary, heterogeneous mass of the right abdominal wall with a distinct fat plane between the mass and anterior rectus fascia. Due to the unexpected urothelial tumor markers, a PET CT was performed to determine the site of the primary lesion. This demonstrated hypermetabolic regions in both the anterior abdominal

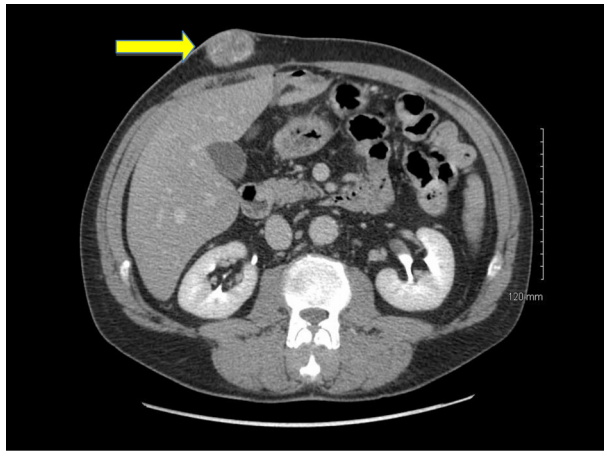

Figure 1. CT scan demonstrating a solitary, heterogeneous 6×4 cm mass of the right abdominal wall (arrow) with a distinct fat plane overlying the anterior rectus fascia. Diagnosis of sebaceous carcinoma was made upon surgical excision.

wall as well as a left thyroid nodule; there was no evidence of distant metastatic disease. Ultrasound guided biopsy of the left thyroid nodule was suggestive of papillary thyroid carcinoma, while the mandibular mass found on physical exam was benign. At that time, the patient was referred to urology due to the possibility of a primary urothelial source; however, cystoscopy with FISH analysis was negative for evidence of urothelial carcinoma.

Despite an extensive workup, the etiology of the mass was still unclear, so the patient was taken to the operating room for a wide local excision. A 6×4 cm mass with 1 cm margins was removed, and it demonstrated a focally hemorrhagic sebaceous carcinoma with clear margins. The patient returned for a total thyroidectomy where a $1.7 \times 1.4 \times 1.2$ cm solid nodule, consistent with papillary thyroid carcinoma, was removed. Due to his history of visceral malignancy and diagnostic workup uncovering both sebaceous and internal malignancies, the patient was referred for genetic counseling. At the time of this article's publication, the results of the patient's genetic tests are still pending.

DISCUSSION

Extraocular sebaceous carcinomas, although rare, are most likely to arise from the skin of the head and neck. Less commonly involved regions include the extremities and external genitalia.[1] Lesions are clinically described as painless, yellow to pink, slowly enlarging, subcutaneous nodules; ulceration and bleeding are

rare secondary changes.[6] Incidence of the lesion is generally slightly higher in male patients, with a median age of diagnosis of 73 years.[7] A review of the literature suggests that ocular and extraocular sebaceous carcinomas share a similar prognosis. A number of metastatic cases of both types have been reported and must be monitored for.[7]

In patients presenting with a sebaceous neoplasm, the diagnosis of MTS requires at least one associated visceral malignancy. While sebaceous carcinoma is not as specific a marker for MTS as a sebaceous adenoma, it has been reported in at least 29 patients with MTS and is considered a possible marker of the syndrome.[4] Thus, the presence of any sebaceous tumor warrants a search for internal malignancy, as well as MTS. According to a review by Cohen et al, the most commonly associated visceral neoplasms in MTS are colorectal (51%) and genitourinary (25%); cutaneous lesions may occur before or concurrently with the diagnosis of visceral malignancies.[5]

Although the results of genetic testing of the patient discussed in this report were still pending at the time of publication, his diagnosis of sebaceous carcinoma along with his history of multiple internal malignancies suggests a possible case of MTS. We recommended regular follow-up and routine monitoring of both the patient and his family members; this includes regular screening for colorectal cancer as well as annual dermatologic examinations. This case demonstrated the complex diagnostic workup that may be required in patients with multiple malignances suggestive of MTS. In the future, an earlier suspicion for MTS in similar patients may prompt a more efficient diagnostic process.

Sebaceous neoplasms, especially in extraocular regions, often mimic more benign cystic lesions leading to misdiagnosis. While the majority of patients will be discovered to have benign lesions, this case demonstrates the importance of obtaining an accurate diagnosis, as sebaceous carcinoma may be an important clue to underlying visceral malignancies associated with MTS.

LEARNING POINTS

1. Seventy-five percent of sebaceous carcinomas are ocular, while 25% are extraocular, arising from any hair-bearing region of the body.
2. Sebaceous carcinoma is commonly associated with MTS, an autosomal dominant condition considered a variant of HNPCC (Lynch) syndrome.
3. In patients presenting with a sebaceous neoplasm, the diagnosis of MTS requires at least one associated visceral malignancy, most commonly colorectal, genitourinary or endometrial.
4. Sebaceous carcinoma is considered a potential diagnostic sign of MTS and its presence should prompt a complete evaluation for internal malignancies, including colorectal cancer screening and annual dermatologic exams.
5. Due to its inheritance pattern, relatives of patients diagnosed with MTS should also be examined for sebaceous and visceral malignancies.

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