INTRODUCTION

Muir-Torre syndrome (MTS) is a rare genetic disorder that causes internal malignancy as well as cutaneous manifestations. The criteria for diagnosis are having one sebaceous neoplasm and one internal malignancy. The most common associated malignancy is colorectal carcinoma. An associated internal cancer may appear before, after, and at the same time as the sebaceous tumor. Treatment involves excision of the skin lesion, resection of internal cancer, and/or screening for future malignancies. We present a case of microsatellite instability noted in a sebaceous adenoma with implications of possible MTS, in the background of immunosuppression and no current known malignancy.

CASE REPORT

A 64 year old Caucasian male presented to our clinic for a complete skin examination. His past cutaneous medical history included two squamous cell carcinomas in situ and one squamous cell carcinoma. His past medical history was significant for hypertension, hyperlipidemia, gout, and immunosuppression status post right kidney transplant in 1994 for IgA nephropathy. Medications that he takes daily were cyclosporine, azathioprine, prednisone, metoprolol, and atorvastatin. No significant family history reported.

The patient presented with a pale pink to yellow, umbilicated papule on the right nasal sidewall (Figure 1). The differential diagnosis included sebaceous, sebaceous hyperplasia, basal cell carcinoma, and molluscum contagiosum. A shave biopsy was performed on the papule after obtaining verbal and written consent, along with local anesthesia with 1% lidocaine with epinephrine. Histopathological examination revealed this papule to have basaloid cells and mature sebocytes, consistent with a sebaceous adenoma and had noted atypia; margins were still involved (Figure 2). This diagnosis led the dermatopathologist to perform further studies which included staining for microsatellite instability involved in Muir-Torre syndrome. The studies showed loss of nuclear staining for MSH2 (Figure 3) and MSH6, suggestive of MTS.

Our patient underwent Mohs micrographic surgery to ensure clearance and for future malignancies. We present a case of microsatellite instability noted in a sebaceous adenoma with implications of possible MTS, in the background of immunosuppression and no current known malignancy.

DISCUSSION

Muir-Torre syndrome (MTS) was first described in 1967 by Muir, and in 1968 by Torre.1 It is a rare germline disorder that predisposes the individual to sebaceous neoplasms and internal malignancies, most notably colorectal adenocarcinoma. This syndrome is described as a variant of hereditary nonpolyposis colorectal cancer (HNPPC) or Lynch syndrome.2,3 It is known to have a higher incidence in males than in females, stating a ratio of 3:2.4 The mean age for cutaneous manifestations is 53 years, and the internal malignancy may appear prior or after the discovery the sebaceous neoplasm.5 Our patient was 64 years old when his first sebaceous neoplasm was discovered, and at this time, does not have a known visceral malignancy. MTS is inherited in an autosomal pattern and caused by germline mutations in mismatch repair genes. The mutations cause microsatellite instability, specifically at MSH2, MLH1, MSH6, and PMS2. The most commonly found gene associated is MSH2,6 and only a small subset shows mutations in MSH6.7,7 However, a newly described subtype has been described, MTS II, that does not have microsatellite instability mutations associated with it but a base excision repair gene mutation in MYH, and shown to be autosomal recessive.8,9 Although MTS is autosomal dominant, immunosuppression has been reported to increase the development of tumors. Two cases of patients’ status post organ transplants that had eruptive sebaceous neoplasms while on tacrolimus.9,10 Our patient was on cyclosporine, azathioprine, and prednisone for immunosuppression and had a solitary sebaceous adenoma, but one may reason that these medications increased the development of the neoplasm. The patient did reveal loss of nuclear staining for MSH2 and MSH6.

Clinical findings associated with MTS are a variety of sebaceous neoplasms, including sebaceous adenomas, carcinomas, and epitheliomas, and keratoacanthomas. The individual must also have an internal malignancy.4 Gastrointestinal malignancy, specifically colorectal adenocarcinoma, is most commonly associated.

Our patient will continue to cancer screenings and made aware of his increased risk for colon cancer.

CONCLUSION

Our patient shows an unusual presentation for this already rare syndrome as he had a nephrectomy and status post kidney transplant due to IgA nephropathy years prior. He currently has no known visceral malignancy. His presentation of sebaceous adenoma with atypia led to the staining for microsatellite instability. The patient has had cancer screenings and will continue to be followed, allowing for early detection. This interesting case urges practitioners to remember MTS when pathology of sebaceous neoplasias arise in an atypical setting and promote further awareness of this genodermatosis.

REFERENCES