On-line Case Report

Muir–Torre syndrome: a case report and screening recommendations

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Muir–Torre syndrome is a rare genodermatosis (hereditary cancer syndrome associated with distinguishing cutaneous signs) characterised by occurrence of sebaceous neoplasia and visceral malignancy. Colorectal cancer is the commonest visceral malignancy associated with Muir–Torre syndrome. We discuss a case report along with recommendations for diagnosis and screening.

Keywords: Muir–Torre syndrome – Sebaceous neoplasia – Colorectal cancer – Visceral malignancy

Muir–Torre syndrome is a rare, autosomal-dominant, genetic condition. It is defined by the occurrence of sebaceous tumours (sebaceous adenoma, epithelioma or carcinoma and/or kerato-acanthoma) and visceral malignancies. The most common visceral malignancy associated with Muir–Torre syndrome is colorectal cancer. The internal malignancies may occur many years before or after the skin lesions. Timely diagnosis can help in screening and surveillance of patients and their families. We present here a case report and recommendations for screening and diagnosis of this rare condition.

Case report

A 57-year-old Caucasian woman presented in April 2004 to the surgical out-patient clinic with a 4-cm diameter lump near the left buttock, which was excised. The histopathologist reported this as a sebaceous adenoma and also raised the possibility of the association of these lesions with visceral neoplasms. The patient had undergone hysterectomy for endometrial cancer at the age of 29 years. She also had an actinic keratosis and a basal

cell carcinoma previously. It was found that her sister had died of bowel cancer in her mid-fifties. In view of these facts, a colonoscopy was performed which revealed a malignant lesion at the ileocaecal region. A staging computerised tomography (CT) scan did not show any distant metastases and the carcino-embryonic antigen level was 42 µg/l. She underwent a laparoscopic right hemicolectomy, the histology of which revealed a poorly differentiated 'signet ring' adenocarcinoma with lymph node involvement (Dukes' C1). She was given an adjuvant chemotherapy regimen and was on our follow-up and surveillance programme. Seven months later, a further cutaneous lesion was excised from her back which was proven to be a sebaceous carcinoma. She has been referred to the genetics department for assessment of genetic mutation and advice regarding screening of the family.

Discussion

Muir (1967) and Torre (1968) separately described this condition where there is association of sebaceous neoplasms and at least one visceral malignancy.^{1,2} The

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Table 1 Screening recommendations for patients with Muir–Torre syndrome and their first-degree relatives⁵

Procedure	Guidelines
Physical examination	Yearly, including breast in women, testicular and prostate in men
Laboratory tests	FBC, CA-125, CEA, FOB, urinalysis
Colonoscopy	Every 1–2 years, from age 25 years or 5 years before the youngest age of diagnosis of colorectal cancer in the family and annually from age 40 years
Pelvic examination	Annually in women with transvaginal ultrasonography and endometrial biopsy in patients with a gene mutation from age 25 years
Consider gastroscopy	Every 1–2 years in families with history of gastric cancer
Consider renal ultrasonography	Every 1–2 years in families with history of renal tract cancer
Consider prophylactic colectomy	In patients with gene mutation

CA-125: carbohydrate antigen 125; CEA: carcino-embryonic antigen; FBC: full blood count; FOB: faecal occult blood.

skin lesions found in association with Muir–Torre syndrome are sebaceous adenoma, epithelioma, sebaceous carcinoma and/or kerato-acanthoma. As these are rare skin lesions, detection of these lesions should raise suspicion of Muir–Torre syndrome and should lead to screening for visceral malignancy. Of the skin lesions in Muir–Torre syndrome, 56% occur after diagnosis of visceral malignancy, 6% occur synchronously and 22% of skin tumours occur as the first lesion of Muir–Torre syndrome.³ The cutaneous lesions may occur 25 years before or 37 years after the visceral malignancy.⁴ Multiple visceral carcinomas may occur metachronously. Colorectal cancer is the commonest visceral cancer accounting for approximately 50% of cases and the lesions may be multiple. It is estimated that genito-urinary cancer occurs in about 25%. Other associated cancers include lung, endometrial, ovarian, parotid, breast, gastric, small intestinal and haematological.⁵

Muir–Torre syndrome has been recognised as a subset of Lynch syndrome type II (hereditary non-polyposis colon cancer [HNPCC]). Genetic mutations are common

in Muir–Torre syndrome and the majority occur in the MSH2 gene. These patients should be treated by the standard surgical and medical treatment and followed up carefully.

Muir–Torre syndrome is a rare condition, but diagnosis of a sebaceous tumour should lead to screening (Table 1) for visceral malignancy in the patient and the family members.

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