VIA MEDICA

Nuclear Medicine Review 2024, 27, 13–16
DOI: 10.5603/nmr.99686

ISSN 1506–9680, e-ISSN 1644–4345

Clinical vignette

A case of Muir–Torre syndrome on [^{18}F]FDG PET/CT

Thomas Fredericks, Khaled Mohamed, Minh X Nguyen, Sindhu Kumar, Savas Ozdemir
Department of Radiology, University of Florida Health, Jacksonville, USA

[Received 6 III 2024; Accepted 9 V 2024]

Abstract

Muir–Torre syndrome (MTS) is a rare genetic disorder, considered a subtype of Lynch syndrome, that causes sebaceous cutaneous tumors and increases the risk of internal visceral tumors. We present a case of a 63-year-old male with a history of MTS with sebaceous tumors, colorectal, and urothelial cancers who underwent fluorine-18-deoxyglucose positron emission tomography/computed tomography [^{18}F]FDG PET/CT to follow-up on multiple [^{18}F]FDG avid skin lesions and right pelvic lymph nodes. Although few reports are available detailing the utility of [^{18}F]FDG PET/CT in this rare disease, this modality appears useful, and superior, to computed tomography in the diagnosis and follow-up of MTS.

KEYwords: Muir–Torre syndrome; [^{18}F]FDG PET/CT; Lynch syndrome
Nucl Med Rev 2024; 27: 13–16

Muir–Torre syndrome (MTS) is a rare autosomal dominant phenotype of hereditary non-polyposis colorectal cancer (a variant of Lynch syndrome) that predisposes individuals to sebaceous tumors of the skin and visceral malignancies, particularly colorectal cancer, endometrial cancer, and urinary tract cancers [1]. The genetic basis of MTS, like Lynch syndrome, involves mutations in DNA mismatch repair that disrupt cell's ability to correct errors in DNA replication, such as *MLH1*, *MSH2*, *PMS2*, and *MSH6* [2]. Thus, diagnosis is made with biopsy histopathologic specimens showing loss of function of these genes [3].

We present a case of a 63-year-old male with a history of MTS with surgically removed sebaceous carcinomas, colorectal cancer, and invasive right urothelial carcinoma s/p nephrectomy who underwent fluorine-18-deoxyglucose positron emission tomography/computed tomography ([^{18}F]FDG PET/CT) to follow-up on multiple [^{18}F]FDG avid skin lesions and right pelvic lymph nodes. The examination performed showed multiple [^{18}F]FDG avid skin lesions within the anterior right chest wall, left medial thigh, posterior right thoracic back (Fig. 1), lateral left knee (Fig. 2), as well as avid pelvic lymph nodes (Fig. 3). The patient subsequently underwent excisional biopsies of all [^{18}F]FDG avid skin lesions which demonstrated pathologic findings of sebaceous carcinoma, with immunohistochemical stains showing absence of *MSH2* and *MSH6*, diagnostic for MTS (Fig. 4). Tissue sampling of the avid pelvic nodes was not performed after patient determination to forego further invasive sampling/treatment was made, however likely

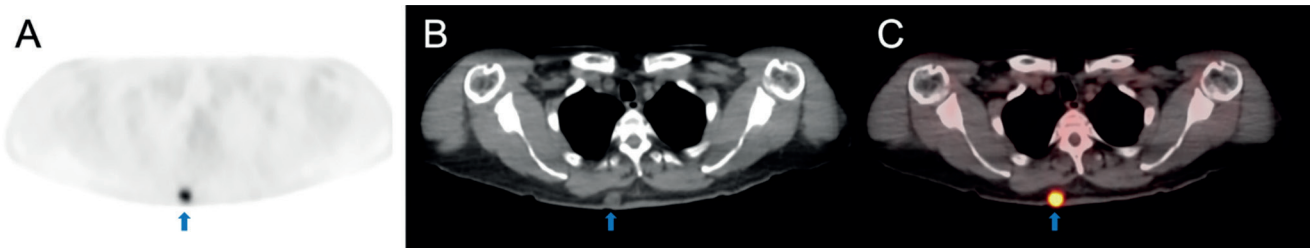

Figure 1. Axial attenuation corrected [^{18}F]FDG PET (**A**), axial CT (**B**), and axial fused [^{18}F]FDG PET/CT (**C**) images demonstrate a posterior right back subcutaneous nodule with significantly increased [^{18}F]FDG avidity (blue arrows)

Correspondence to: Thomas Fredericks, Department of Radiology, University of Florida Health, 655 West 8th St., 32209 Jacksonville, USA, e-mail: thomas.fredericks@jax.ufl.edu

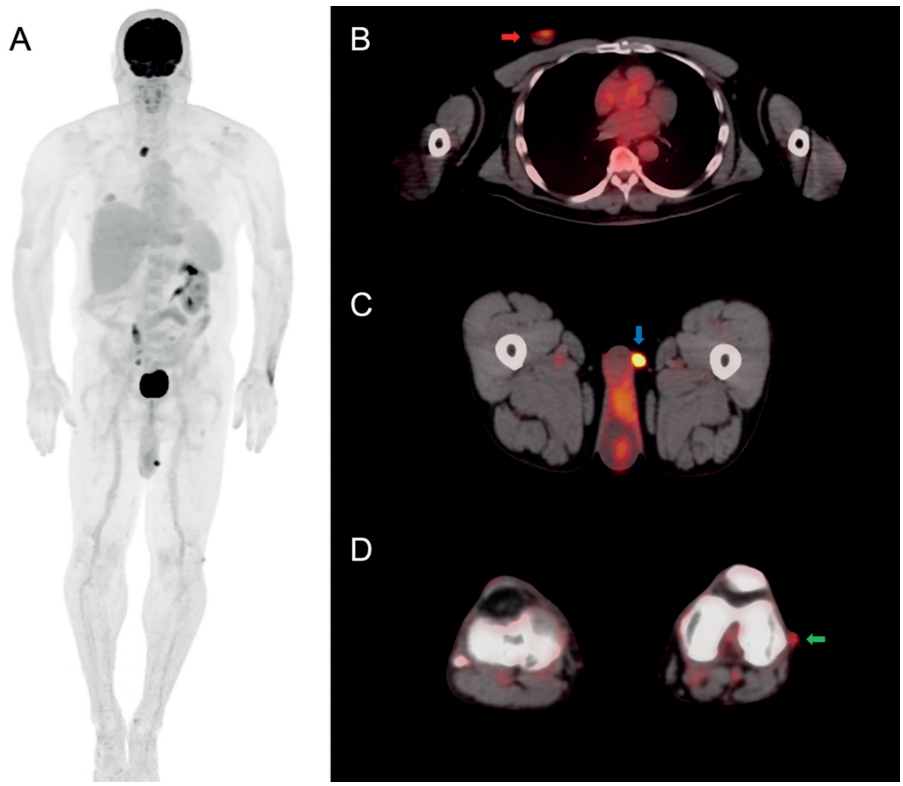

Figure 2. [^{18}F]FDG maximum intensity projection (MIP) (**A**) shows increased [^{18}F]FDG avidity within the anterior right chest wall, posterior right thoracic back (demonstrated on Figure 2), left medial thigh, lateral left knee, and right iliac lymph node chains (demonstrated on Figure 3). Note the absence of physiologic uptake within the right kidney status post right nephrectomy. Fused axial [^{18}F]FDG PET/CT images (**B–D**) demonstrate increased [^{18}F]FDG avidity with CT correlate subcutaneous nodules within the anterior right chest (**B**), left medial thigh (**C**), and lateral left knee (**D**)

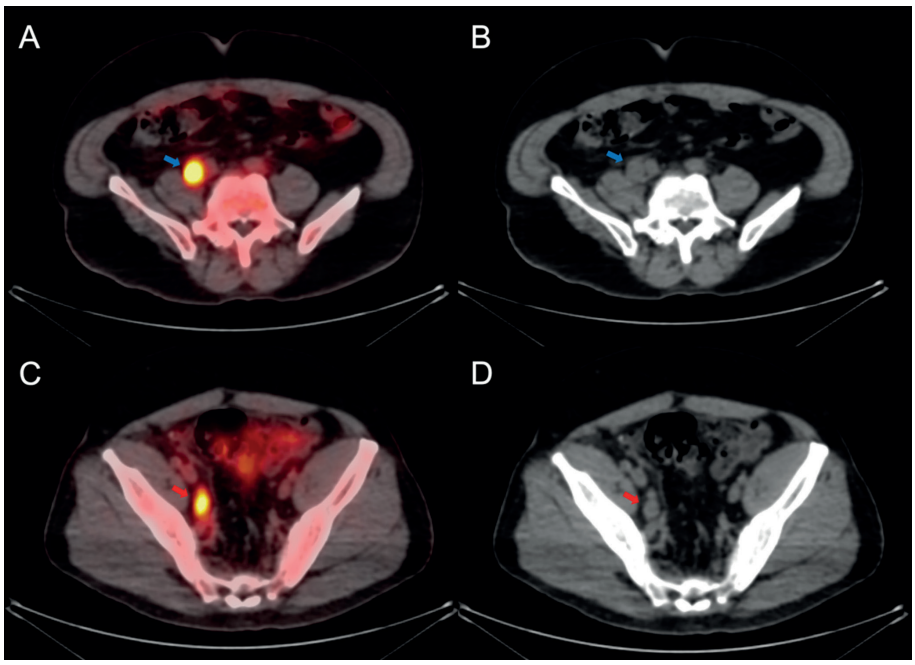

Figure 3. Axial fused [^{18}F]FDG PET/CT (**A, C**) and axial CT (**B, D**) images show a [^{18}F]FDG avid right common iliac node (**A** and **B**) (blue arrows), as well as a [^{18}F]FDG avid right internal iliac lymph node (**C** and **D**) (red arrows), suspicious for metastatic disease given patient history

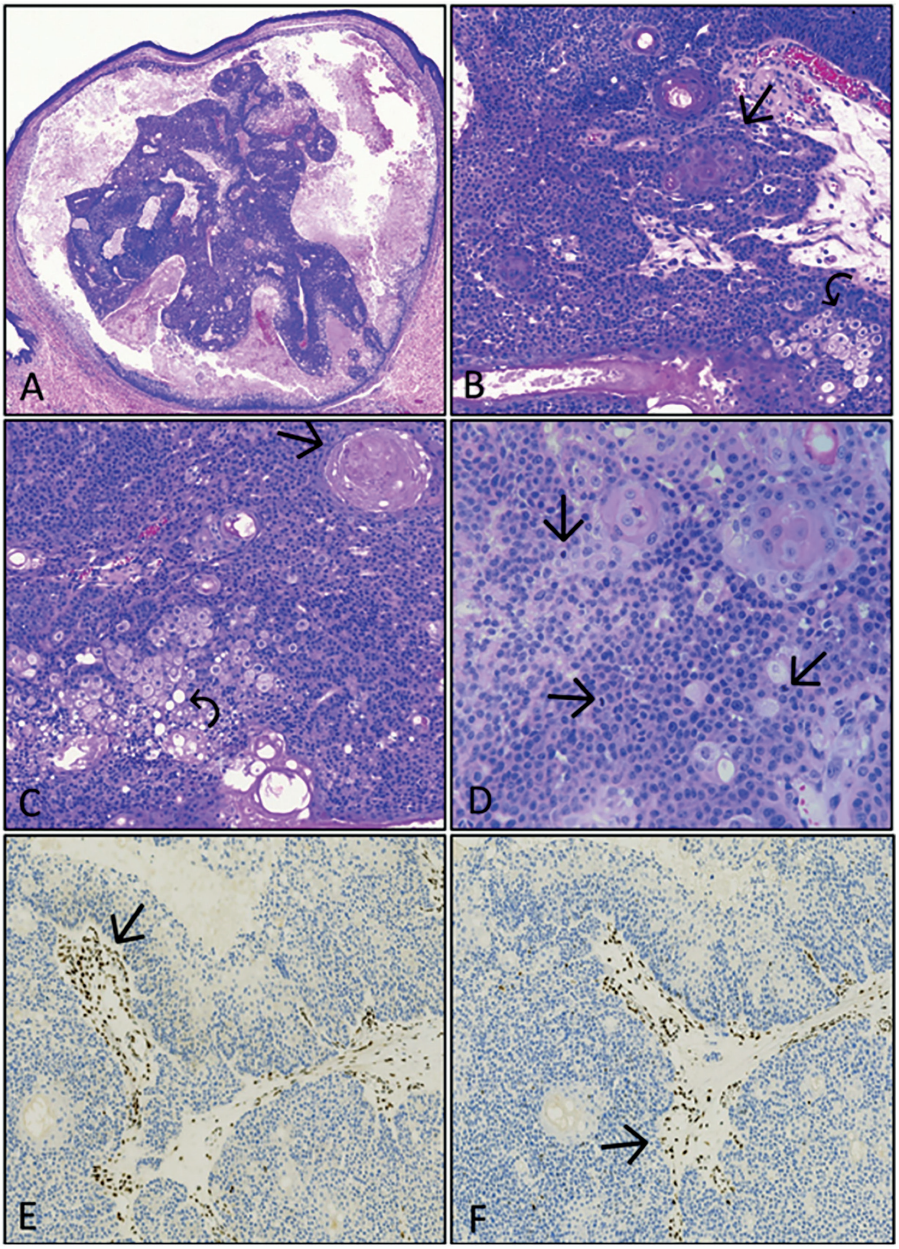

Figure 4. Histopathologic specimen of the [18F]FDG PET/CT avid lesion in the upper right back shows a dermal-based infiltrative nodular tumor (**A**); (**B**) reveals a moderately differentiated tumor consisting of variably differentiated epithelioid cells; predominantly basaloid cells with areas of squamoid (black straight arrow) and clear (black curved arrow) cells; (**C**) shows areas of comedonecrosis (black straight arrow) and focal clear spaces consistent with sebaceous ducts (black curved arrow); (**D**) displays enlarged epithelioid cells with abundant clear to eosinophilic cytoplasm with hyperchromatic pleomorphic nuclei, prominent nucleoli, and multiple mitotic figures including atypical forms (black straight arrow). Immunohistochemistry at 10× power for MSH2 (**E**) and MSH6 (**F**) show loss of nuclear staining in tumor cells and intact staining in the stromal cells that serve as internal controls (black straight arrows)

was related to neoplastic involvement from prior treated colorectal or urothelial carcinoma.

As illustrated in our case, cutaneous lesions can be diffusely found and recurrent, while visceral tumors can be numerous, arise from multiple organ systems (gastrointestinal, urinary, reproductive), and can recur with poor outcomes. Whole body [18F] FDG PET/CT appears useful and superior to computed tomography in the diagnosis and follow-up of MTS, particularly in identifying less conspicuous cutaneous tumors and MTS-associated internal malignancies [4–6].

References

1. Bhaijee F, Brown AS. Muir-Torre syndrome. Arch Pathol Lab Med. 2014; 138(12): 1685–1689, doi: 10.5858/arpa.2013-0301-RS, indexed in Pubmed: 25427047.
2. Gamret AC, Klingbeil KD, Fertig RM, et al. Muir-Torre syndrome. Skinmed. 2020; 18(6): 382–384, indexed in Pubmed: 33397571.
3. John AM, Schwartz RA. Muir-Torre syndrome (MTS): an update and approach to diagnosis and management. J Am Acad Dermatol. 2016; 74(3): 558–566, doi: 10.1016/j.jaad.2015.09.074, indexed in Pubmed: 26892655.
4. Ishiguro Y, Homma S, Yoshida T, et al. Usefulness of PET/CT for early detection of internal malignancies in patients with Muir-Torre syndrome: report of two cases. Surg Case Rep. 2017; 3(1): 71, doi: 10.1186/s40792-017-0346-7, indexed in Pubmed: 28537014.
5. Bertagna F, Biaotto G, Bosio G, et al. F18-FDG-PET/CT in a patient affected by Lynch syndrome. Nucl Med Rev Cent East Eur. 2010; 13(2): 87–89, indexed in Pubmed: 21598234.
6. Kviatkovsky B, Landau E, Siddique M, et al. FDG-PET-positive lower-extremity sebaceous-gland carcinoma in a patient with Muir-Torre syndrome. Radiol Case Rep. 2016; 10(2): 1115, doi: 10.2484/rcr.v10i2.1115, indexed in Pubmed: 27398125.