Gastrointestinal stromal tumour in neurofibromatosis 1: ^{18}F FDG PET/CT imaging

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DESCRIPTION

A middle-aged patient with known neurofibromatosis type 1 (NF1) presented with an increase in the size and number of cutaneous lesions. The lesions were further evaluated with positron emission tomography (PET)/CT that showed diffuse hypermetabolic neurofibromas of the neck, thorax, proximal lower extremities (figure 1) and a fluorodeoxyglucose (FDG)-avid lesion in the left lower abdominal quadrant. Biopsy of the FDG-avid lesions on the left shoulder and right thigh were histologically consistent with neurofibromas. A composite PET/CT fused image (figure 2) demonstrated a FDG-avid lesion in the small bowel, and the corresponding CT images showed a

3.5 cm contrast-enhancing mass arising from the jejunum (figure 3). Subsequent biopsy, surgery and pathology found a low-risk 3.6 cm gastrointestinal

Figure 1 Coronal positron emission tomography/CT image demonstrating diffused hypermetabolic lesions in the neck, thorax and proximal lower extremities consistent with neurofibromas. A further fluorodeoxyglucose-avid lesion (black arrow) is identified in the left lower abdominal quadrant.

Figure 2 An axial composite positron emission tomography/CT fused image demonstrating a suspicious fluorodeoxyglucose-avid lesion arising from the small bowel (white arrow).

Figure 3 Axial CT image showing a contrast-enhancing mass arising from the jejunum (white arrow).

Learning points

- ▶ There is a need for close tumour surveillance in the neurofibromatosis type 1 (NF1) population.
- ▶ Patients with NF1 are at increased risk of several malignancies, including gastrointestinal stromal tumour (GIST) and malignant peripheral nerve sheath tumours.
- ▶ The incidental finding of GIST demonstrating the clinical utility of positron emission tomography/CT as a diagnostic tool in clinical oncology.

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stromal tumour (GIST) with two tiny GISTs in close proximity beyond PET and CT resolution.

There is a known association between NF1 and GISTs, with GISTs developing in 5–30% of patients with NF1.¹ The role of PET/CT in GISTs is not yet well established, but is useful in assessing tumour response to tyrosine kinase inhibitors therapy.² The most frequent tumours seen in NF1 are neurofibromas. Of concern, these may transform from benign plexiform neurofibroma (PNF) into malignant peripheral nerve sheath tumours (MPNST). FDG-PET and PET/CT have demonstrated their clinical utility in discriminating between benign PNF and MPNST based on maximal standard uptake values.³

Competing interests None declared.

Patient consent Obtained.

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