Clinical Case Report

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The value of 18F-FDG PET/CT in patient with neurofibromatosis type 1

A case report and literature review

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Abstract

Rationale: Neurofibromatosis type one (NF1) is characterized by cutaneous and nervous lesions, and the tendency to form plexiform neurofibromas (PNFs). PNFs may undergo malignant transformation into a malignant peripheral nerve sheath tumors (MPNSTs). MPNSTs often carry an significant morbidity and mortality.

Patient concerns: A 17-year-old man with gradually increased multiple subcutaneous soft lesions. He also presented with numerous lentigines and multiple café-au-lait macules on his body.

Diagnoses: These were collagen neurofibroma, which were definitively diagnosed by pathology. NF1 was eventually diagnosed.

Interventions: These lesions were abnormal uptake of radiotracer, when he underwent positron emission tomography (PET) with fluorine-18-fluorodeoxyglucose (FDG) scanning. Standard uptake value (SUV) and other parameters can help to distinguish benign and malignant lesions in patient with NF1. He was underwent serials ^{18}F-FDG PET/CT examinations to followed up, in order to monitor these lesions malignant transformation.

Outcomes: So far, these subcutaneous soft lesions were not malignant transformation.

Lessons: ^{18}F-FDG PET/CT is being increasingly used as an imaging modality to discover the systemic lesions and to discriminate between benign and malignant plexiform neurofibromas.

Abbreviations: BAT = brown adipose tissue, FDG = fluorine-18-fluorodeoxyglucose, MPNST = malignant peripheral nerve sheath tumours, NF1 = neurofibromatosis type 1, PET = positron emission tomography, PNF = plexiform neurofibroma, SUV= standardized uptake value.

Keywords: ^{18}F-FDG PET/CT, café-au-lait macules, neurofibromatosis

1. Introduction

Neurofibromatosis type one (NF-1) is a common inherited disorder with an incidence of 1:2500 to 1:3000, which characterized by cutaneous and nervous lesions, and the tendency to form plexiform neurofibromas (PNFs). And more worryingly, many PNFs may undergo malignant transformation into a malignant peripheral nerve sheath tumors (MPNSTs). However, MPNSTs carry a significant morbidity and mortality owing to delayed diagnosis, local recurrence, early metastasis, and poor therapeutic effect. Medical imaging can lead to earlier diagnosis and improve prognosis in patient with NF1. Magnetic resonance imaging (MRI) can detect MPNST more sensitively and specificity, which can provide a complete characterization of the genesis, growth dynamics, and the range of nerve sheath tumors. And whole-body MRI is more suited to serial follow-up in patients with PNFs, owing to its no radiation exposure.[1]^{18}F-FDG PET/CT usually provides a high sensitivity and specificity for monitoring malignant lesions, particularly for staging and restaging in patient with tumor, in order to change therapy planning in time and improve prognosis. Our aim is to discuss the role of ^{18}F-FDG PET/CT in NF1 patients.

2. Case report

A 17-year-old man presented with an increase in the size and number of cutaneous lesions (Fig. 1A) and subcutaneous soft lesions. ^{18}F-FDG PET/CT has been underwent which showed diffuse hypermetabolic foci of the neck, clavicular region, near iliac vessels of left, near piriformis, and gastrocnemius of left leg. The hypermetabolic tissue in pharynx was physiological (Fig. 1C). And hypermetabolic tissues in clavicular region was brown adipose tissue (BAT) (Fig. 1D), because of this young man was too shin and underwent ^{18}F-FDG PET/CT in winter.[2,3] The lesions under sternocleidomastoid (Fig. 1C), gastrocnemius (Fig. 1E) of left were neurogenic tumors, which were benign neurofibroma (Fig. 1B) by pathology.

The standardized clinical diagnostic criteria of NF1, which have existed since 1987.[4] Many numerous lentigines and multiple café-au-lait macules on his father were founded. So, NF1 of this young man was eventually diagnosed.

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The authors report no conflicts of interest.

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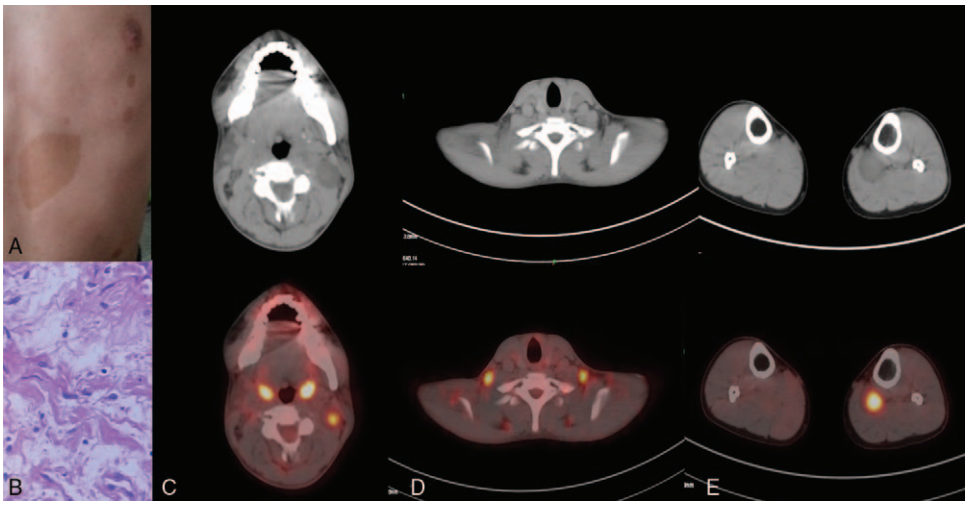

Figure 1. A–E. A 17-year-old man presented with several cafe-au-lait spots, the tissues of neck and left leg uptake of radiotracer were revealed by ^{18}F-FDG PET/CT scanning. Many café-au-lait macules were widely distributed in anterior and posterior chest wall of this patient, which is >15 mm of diameter (A). These (C, E) were confirmed as benign neurofibroma by pathology (B). The hypermetabolic lesions under sternocleidomastoid, near piriformis, and gastrocnemius of left were neurogenic tumors. The hypermetabolic soft tissues of pharynx were physiological (C). The symmetric hypermetabolic tissues, which located in clavicular region, were BAT (D). BAT = brown adipose tissue, FDG = fluorine-18-fluorodeoxyglucose, PET = positron emission tomography.

Ethical approval was given by the medical ethics committee of Shandong Cancer Hospital affiliated to Shandong University. The patient has consented for the publication of the present case report.

3. Discussion

The tumor suppressor gene mutation in chromosome 17, was considered as the reason of NF1.[5] The terrible results may be occurred, for instance the development of malignant tumors, because gene mutation. Many pathological types of tumor may be occurred in NF1 patient. Patients with NF1 have a cumulative lifetime risk of approximately 10% for developing an MPNST versus <0.1% for the general population.[6,7] MPNST is a life-threatening disease, which has poor prognosis. Therefore, a definite and early distinguish lesion was significant.

Standard uptake value (SUV) can help to distinguish benign and malignant lesions in patient with NF1. Some scholars found that the SUVmax of benign lesions (2.49) was significantly different from malignant lesions (7.63) in pediatric population, respectively. A cutoff SUVmax value of 4.0 had high sensitivity and specificity of 1.0 and 0.94 to distinguish between benign and malignant lesions. The SUVmax is 4.0 that can predict the optimal cutoff of malignant transformation.[8] For mean tumor SUV, the SUV was calculated for 20 NF1 patients and was obvious higher in 5 malignant tumors 5.4, than in 15 benign tumors 1.54. Some tumors were difficult to distinguish benign or malignant, when the SUV between 2.7 and 3.3.[9,10] The mean survival time was 13 months for patients with an SUV >3, compared with 52 months, for whom with an SUV <3. What's more, SUV has been had a significantly higher accuracy (94%), than tumor grade (69%), when they predicted long-term survival.[11]

SUV is a unitless semiquantitative measure of FDG. Several factors, for instance, ambient temperature, body weight, respiratory effort, blood glucose level, and the amount of time between injection of radionuclide and scanning, may affect the SUV measurement. These factors may interfere with diagnosis. Novel parameters such as metabolic tumor volume (MTV) and total lesion glycolysis (TLG) have been used which have shown promise but lack enough evidence to justify routine use.[1] Further research is needed to study for precise diagnosis.

It had the important significance of ^{18}F-FDG PET/CT in diagnosis and differential diagnosis the lesion nature in patient with NF1.[12] These can impact on treatment and prognosis of patient with NF1.

Author contributions

Conceptualization: jing zhou.
Data curation: zheng fu.
Investigation: zheng fu.
Resources: zheng fu.
Supervision: zheng fu.
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