CASE REPORT

A case of von Recklinghausen's disease with coincident malignant peripheral nerve sheath tumor and gastrointestinal stromal tumor

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Abstract :^{18}F-fluorodeoxyglucose positron emission tomography (^{18}F-FDG PET/CT) was performed to evaluate a left femoral subcutaneous mass in a patient with von Recklinghausen's disease (vRd) that gradually enlarged, causing pain and numbness. The left femoral mass showed intense FDG uptake with the maximum standardized uptake value (SUVmax) of 9.0. Other subcutaneous masses considered benign and neurogenic in nature also showed FDG uptake (SUVmax around 3 or less), but the degree of FDG uptake differed considerably from the left femoral mass. This observation suggested that the degree of FDG uptake may be a useful indicator of malignant transformation. Incidentally, PET/CT also showed an asymptomatic large abdominal mass with intense FDG uptake (SUVmax 8.8). The abdominal mass was resected and confirmed as gastrointestinal stromal tumor (GIST) of the small intestine. Three months later, the left femoral mass was operated on and pathologically diagnosed as a malignant peripheral nerve sheath tumor (MPNST). Various malignant diseases are known to occur with high frequency in vRd. Therefore, vRd patients need to have periodical examinations including PET/CT. We present a rare case of a patient with vRd with a MPNST of the left femur and coincidental GIST of the small intestine. J. Med. Invest. 56 : 76-79, February, 2009

Keywords : *^{18}F-FDG PET/CT, von Recklinghausen's disease, MPNST, GIST*

INTRODUCTION

von Recklinghausen's disease (vRd, Neurofibromatosis type 1, NF-1) is an autosomal dominant hereditary disease due to an alteration in the long arm of chromosome 17 and has a prevalence of approximately 1 in 3000 (1). It is characterized by cutaneous and plexiform neurofibromas and abnormal skin pigmentation called café au lait spots. vRd patients are known to have a high frequency of various malignant diseases (2-4). Malignant peripheral nerve sheath tumor (MPNST) is the malignant counterpart to benign soft tissue tumors, such as neurofibroma or schwannoma and occurs in about 2-5% of patients with vRd whilst gastrointestinal stromal tumor (GIST) occurs in about 7% of patients with vRd (2, 5).

We present a rare case of a 57-year-old male with vRd affected by MPNST of the left femur and coincidental GIST of the small intestine.

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CASE REPORT

A 57-year-old man suffering vRd since childhood noticed a subcutaneous mass on his left femur was gradually becoming enlarged. He complained of pain and numbness and had a focal fever around the left femoral mass. Physical examination revealed numbness on the lateral surface of the left femur in the region supplied by the lateral femoral cutaneous nerve. He also had other multiple, soft, painless, subcutaneous masses and café au lait spots on the trunk. He had a family history of vRd that his mother and son were also with vRd.

Laboratory findings showed mildly elevated leukocyte counts (12,300/μl) and elevated C-reactive protein (4.92 mg/dl). No increase was seen in tumor markers such as carcinoembryonic antigen (CEA) or carbohydrate antigen 19-9 (CA19-9).

^{18}F-fluorodeoxyglucose positron emission tomography (^{18}F-FDG PET/CT) was performed to evaluate the left femoral mass (Fig. 1). ^{18}F-FDG (3.7 MBq/kg of body weight : 215 MBq) was administered intravenously and the scan was obtained 60 minutes after the injection. The left femoral mass, measuring 5.0×5.5×10 cm, showed intense FDG uptake with the maximum standardized uptake value (SUVmax) of 9.0 on PET/CT (Fig. 2a). Moderate FDG uptake was also observed in other subcutaneous masses (Fig. 3b). Incidentally, PET/CT showed a large abdominal mass measuring 10×12 cm with intense FDG uptake (SUVmax 8.8 ; Fig. 4a). In spite of such a large abdominal mass, he had no abdominal pain or constipation.

The patient underwent further radiological studies. On magnetic resonance imaging (MRI), the left femoral mass showed hypointensity on T1-weighted images and hyperintensity on both T2-weighted and diffusion-weighted images (Fig. 2b, c), with strong enhancement on post contrast T1-weighted images (Fig. 2d). Other subcutaneous masses showed similar intensity to the left femoral mass (Fig. 3c, d).

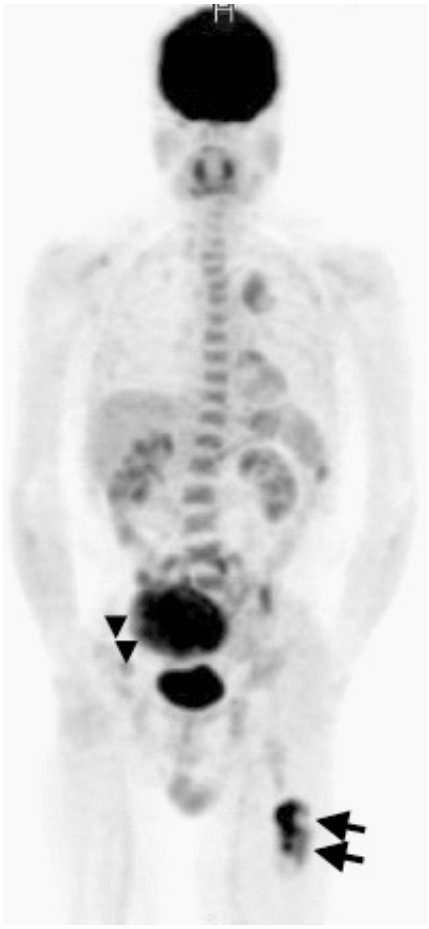

Fig. 1. FDG-PET image (maximum intensity projection image). High uptake of FDG is demonstrated in the left femur (arrows) and intense uptake is also demonstrated in lower abdomen (arrowheads).

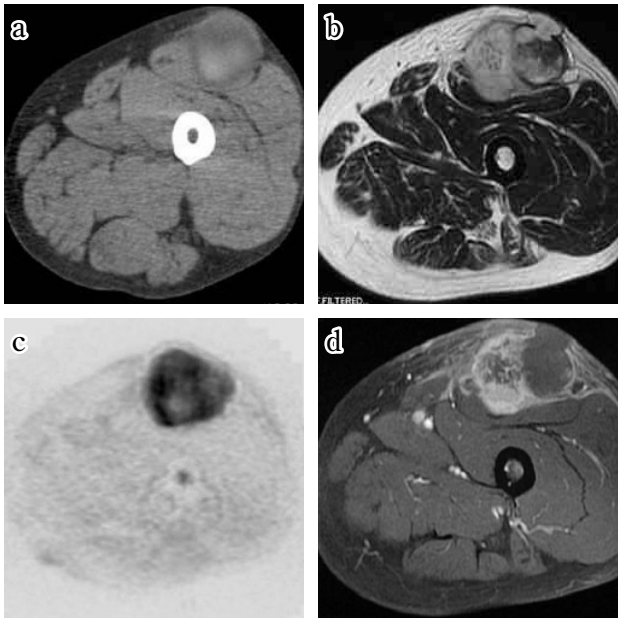

Fig. 2. The left femoral mass shows high FDG uptake (SUVmax 9.0) on a PET/CT image (a) and hyperintensity on both T2-weighted (b) and diffusion-weighted images (c), with strong enhancement on a post contrast T1-weighted image (d).

The large abdominal mass with central decreased uptake, suggestive of ulcer formation or necrosis, showed hypointensity on T1-weighted images, hyperintensity on both T2-weighted and diffusion-weighted images (Fig. 4b). Contrast-enhanced computed tomography showed strong enhancement of the large abdominal mass (Fig. 4c).

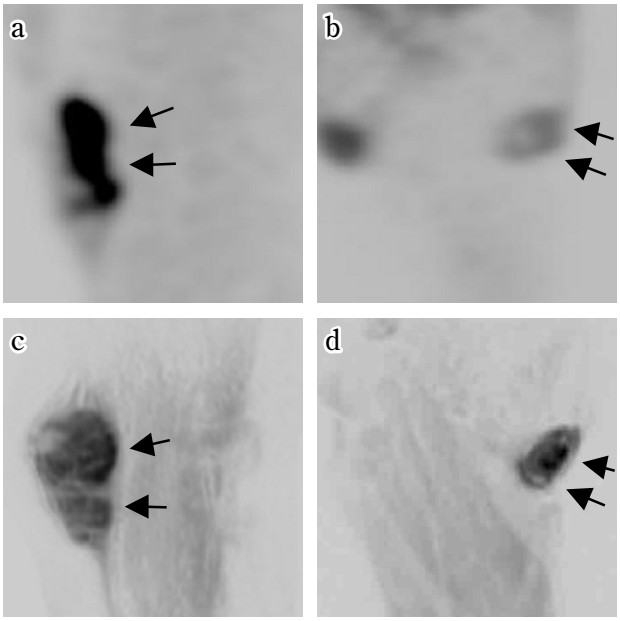

Fig. 3. High FDG uptake is observed in the left femoral mass on a sagittal PET image (a, arrows), whereas moderate FDG uptake is observed in the other subcutaneous mass considered benign and neurogenic in nature on a sagittal PET image (b, arrows). On diffusion-weighted image, similar hyperintensity is seen in both the left femoral mass (c, arrows) and the other subcutaneous mass (d, arrows).

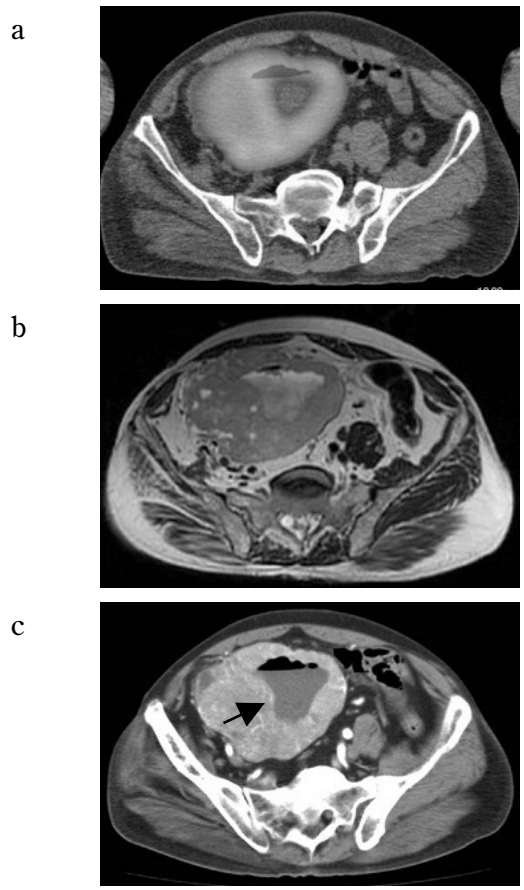

Fig. 4. A large abdominal mass, with central ulcer formation or necrosis (c, arrow), shows intense FDG uptake (SUVmax 8.8) on PET/CT image (a) and hyperintensity on a T2-weighted image (b), with strong enhancement on contrast-enhanced computed tomography (c).

Consequently, we preoperatively diagnosed the left femoral mass as a malignant peripheral nerve sheath tumor (MPNST) and the large abdominal mass as a tumor of the small intestine, such as MPNST or gastrointestinal stromal tumor (GIST), as patients with vRd have a high risk of these tumors which possess generally high-level FDG uptake.

The abdominal mass was resected and confirmed as GIST of the small intestine. Three months later, the left femoral mass was operated and pathologically diagnosed as MPNST.

DISCUSSION

MPNST with vRd is considered the result of the malignant transformation of a benign neurogenic mass such as neurofibroma or schwannoma (6). Growth tendency, subjective symptoms of pain or numbness, and local fever have been reported as good indicators of malignant transformation (6, 7). In our case, gradual enlargement of the left femoral mass with pain and numbness and local fever were suggestive of MPNST as well. The left femoral mass and other multiple subcutaneous masses showed increased FDG uptake, but the degree of FDG uptake differed considerably. The former showed high FDG uptake (SUVmax 9.0) (Fig. 3a), but the latter showed moderate FDG uptake (SUVmax around 3 or less) (Fig. 3b). The difference of FDG uptake between the left femoral mass and other subcutaneous masses in our case suggested that the degree of FDG uptake could be another useful indicator of malignant transformation. It is reported that there is a significant difference in the percent increase in neurofibroma volume in the following year for lesions that had an SUV > 2 compared with those with lower values (8).

On the other hand, not only the left femoral MPNST, but also multiple subcutaneous soft masses considered benign and neurogenic in nature, showed similar hyperintensity on diffusion-weighted images (Fig. 3c, 3d). In our case, it is difficult to distinguish MPNST from benign neurogenic tumor by means of diffusion-weighted images.

It is reported that GIST with vRd occurs often in the small intestine and show a tendency of occurring in multiple localities (9). In our case, GIST of the small intestine was noticed unexpectedly. It is known that various malignant diseases occur with high frequency in vRd patients (2-4). Therefore, vRd

patients need to have periodical examinations, including PET/CT.

Although both MPNST and GIST are not rare with vRd, to our knowledge, there has only been one case reported of vRd with coincident occurrence of these tumors (6). We report a case of vRd with MPNST and GIST. In this case, the asymptomatic large abdominal mass (GIST) was found incidentally on PET/CT performed for the evaluation of a left femoral mass (MPNST). As far as we know, there has only been one report of vRd with coincident MPNST and GIST. The degree of FDG uptake may be a useful indicator of the malignant transformation of a benign neurogenic mass turning into a malignant tumor (MPNST) in the follow up study.

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