Neurofibroma in the retroperitoneum associated with neurofibromatosis type 1

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DESCRIPTION

A 71-year-old woman presented with epigastric pain after having meals visited our hospital. On examination, she was diagnosed with gallstone cholecystitis, complicated with common bile duct stones.

She was treated appropriately, and her conditions improved. It was fortuitously noted that she had a retroperitoneal mass with a maximum diameter of >10 cm surrounding the abdominal aorta (figure 1A).

However, she had no associated symptoms.

Considering this finding, we repeated the physical examination and medical interview. Several neurofibromas and café-au-lait spots were noted on the body, and a detailed interview revealed that she had been diagnosed with neurofibromatosis type 1 (NF1) previously (figure 1B). Contrast-enhanced CT showed a mild contrast around the mass. Positron emission tomography/CT (PET/CT) showed mild accumulation of the mass: the maximum standardised uptake value (SUV_{max}) was 3.7. Therefore, we assumed that the mass was a neurofibroma associated with NF1; however, we could not exclude the possibility of a malignant peripheral nerve sheath tumour (MPNST) based on imaging studies alone. Therefore, a tumour biopsy was performed simultaneously with laparoscopic cholecystectomy for a definitive diagnosis. The pathological diagnosis was confirmed as neurofibroma rather than MPNST because no positive findings for malignancy were observed (figure 2).

Until 4 years after the biopsy, the patient has been followed-up by imaging, and the neurofibroma in the retroperitoneum has been stable in size.

MPNSTs can arise from any neurofibroma, regardless of location, and patients with NF1 may have MPNSTs, especially in the retroperitoneum.[1] The prognosis of MPNSTs may be poor because of their rapid growth and the possibility of distant metastasis.[2] Furthermore, retroperitoneal tumours rarely present with specific symptoms, and delayed diagnosis is one of the factors for poor prognosis.[2 3] The lifetime risk of developing MPNST in patients with NF1 patients is estimated to be 8%–13%.[4] Although it is difficult to predict the occurrence of MPNST, it should be noted that MPNST is more common in adolescents, and symptoms such as pain may occur. Furthermore, imaging studies may show a rapidly enlarging mass.[5 6] International guidelines, such as the European Society for Medical Oncology, recommend performing a core-needle CT scan-guided biopsy for diagnosis, and surgical resection of the tumour is recommended for MPNSTs even if they are asymptomatic.[2 7]

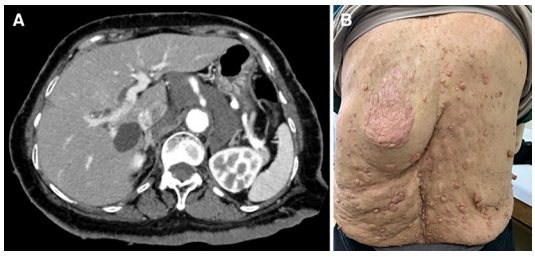

Figure 1 (A) Contrast-enhanced CT scan indicating a retroperitoneal mass around the abdominal aorta. (B) Numerous neurofibromas appearing on the back.

A report involving imaging studies suggested the possibility of MPNST in the presence of two or more of the following four findings: intratumoural cystic lesion, largest dimension >10 cm, peripheral enhancement pattern and perilesional oedema-like zone.[8] Our patient presented with two features: largest dimension >10 cm and peripheral enhancement pattern, which led to the suspicion of MPNST. In contrast, another report suggested that malignancy should be considered when SUV_{max} exceeds 4.8 on PET/CT.[9] In our patient, the SUV_{max} was 3.7; therefore, we did not suspect malignancy.

Since the results of MRI and PET/CT were difficult to diagnose, we performed a biopsy to produce an early diagnosis. Some reports indicate that diagnosis based on imaging studies alone is generally difficult, and careful pathological examination is necessary.[4 10]

If a neoplastic lesion is observed in a patient with NF1 and is strongly suspected to be malignant on imaging studies such as PET/CT, pathological examination should be performed at an early stage and surgical resection should be considered, despite the risk of invasiveness.

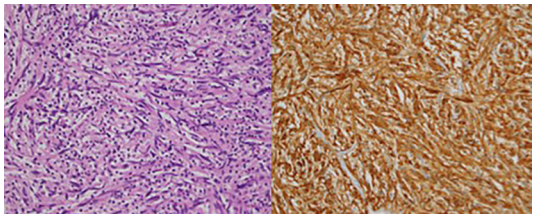

Figure 2 Histological samples showing spindle-shaped cells, although no increase in cell density or mitosis was observed, and immunohistochemical staining showing positivity for S-100 protein, leading to the diagnosis of neurofibroma.

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Learning points

- Neurofibromatosis type 1 may be associated with retroperitoneal tumours, among which malignant peripheral nerve sheath tumour (MPNST) is highly malignant, with rapid progression and the possibility of metastasis.
- Retroperitoneal tumours are less likely to cause symptoms and may be detected incidentally on imaging examinations.
- A pathological diagnosis should be considered necessary, and the patient should be closely followed because there is a possibility of malignant retroperitoneal tumours, such as MPNST.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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