Malignant peripheral nerve sheath tumor in a patient with neurofibromatosis 1

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Introduction

Malignant peripheral nerve sheath tumor (MPNST) is the malignant counterpart to benign soft tissue tumors, such as neurofibroma or schwannoma. It occurs in about 2.5% of patients with neurofibromatosis 1 compared with an incidence of 0.001% in the general population8).

We report a rare case of MPNST in a young female patient with neurofibromatosis 1. The tumors were found in the retroperitoneum and extradural space of the sacrum. She complained of right severe sciatica, and was treated by surgical resection and adjuvant chemotherapy. She died of liver and lung metastases 19 months after the first surgery.

Case report

A twenty-year-old female complained of severe radiating pain from the right buttock area to the right posterior thigh and calf. The pain had been worsening and was disturbing her sleep. Physical examination showed elastic hard masses in the right lower abdomen and right chest wall. She also had several café au lait spots. There was no neurological deficit of the lower extremities. Straight leg raising test for the right side was positive. There was no sign of either urinary or fecal dysfunction. Magnetic resonance images of the sacrum showed an extradural mass of tumor extending to the presacral space (Fig. 1), and those of the pelvis showed an enormous mass of another tumor infiltrating the right psoas muscle (Fig. 2). Computed tomogra-
Figure 1  T2-weighted magnetic resonance image of the sacrum showing an extradural mass extending to the presacral space.

Figure 2  T2-weighted magnetic resonance image of the pelvis showing an enormous mass infiltrating the right psoas muscle.

Figure 3  Computed tomography of the chest wall showing right intercostal mass compressing the liver.

Figure 4  Sacral tumor in the extradural space that was not continuous with the cauda equina.

and was not continuous with the cauda equina (Fig. 4). The tumor in the right lower abdomen extended as a retroperitoneal mass and continued with the femoral nerve. The tumor invaded the right ureter, which was intralesionally resected. On histological examination, these tumors were composed of spindle-shaped cells with hypercellularity and mitoses (Fig. 5). Hemorrhage, necrosis, and invasion of white blood cells were found. Immunohistochemical staining for S-100 and vimentin were positive. Cytokeratin was negative. These tumors were diagnosed
as MPNST. The tumor in the right chest wall was marginally resected. This tumor was a typical neurofibroma. Systemic adjuvant chemotherapy was administered with a combination of doxorubicin (80mg) and ifosfamide (9g). After two courses of chemotherapy, the size of the tumor in the sacral area decreased. A further four courses of the same chemotherapy were given with the addition of cyclophosphamide (100mg). However, she developed a local relapse of the retroperitoneal tumor. Repeated resection of the tumor in the retroperitoneal space was performed twice to lessen the severe pain. The patient’s condition gradually deteriorated, and she died of liver and lung metastases 19 months after the first surgery. Her family was informed that data from the case would be submitted for publication, and they gave consent.

Discussion

MPNST is a rare tumor, usually arising in somatic soft tissues or peripheral nerves. The single most common nerve of origin is the sciatic nerve, and then in order of decreasing frequency by the brachial plexus, spinal nerve roots, vagus, and femoral nerves. In our case, the retroperitoneal tumor seemed to be the primary tumor arising from the femoral nerve and the extradural tumor of sacral area was considered metastatic, because the extradural tumor had no recognizable continuity with the nerves.

Extradural metastases of MPNST is rare. A review of the available literature found six other cases of MPNST metastatic to the spine. However, these cases mainly involved metastasis to the vertebra and lamina. Only one case showed extradural metastasis.

Figure 5  A. Photomicrograph of the extradural tumor. The tumor was composed of spindle shaped cells with pleomorphic nuclei. Mitoses were abundant. (H.E.×100).
B. Photomicrograph of the retroperitoneal tumor. The tumor was composed of spindle shaped cells with hypercellularity. The nuclei showed atypia and mitoses. (H.E.×100).
Although the aim of surgery is complete removal of the lesion with a tumor-free margin, resection of intra-abdominal or sacral tumors was challenging to confirm the histological diagnosis and relieve the pain. The effects of adjuvant chemotherapy are controversial. There have been a few case reports indicating that chemotherapy was effective for MPNST. Masui et al. reported a case of MPNST of the sacrum that decreased after administration of ifosfamide, vincristine and cyclophosphamide. Kinebuchi et al. reported lung metastases of MPNST that decreased in response to carboplatin and etoposide. In our case, because the patient showed a transient response to adjuvant chemotherapy, we continued chemotherapy extensively. However, the effect of chemotherapy was not persistent.

References