Malignant Triton tumor in the retroperitoneal space associated with neurofibromatosis type 1: a case study

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Abstract

We report an extremely rare case of malignant Triton tumor developing in the retroperitoneal space in a patient with neurofibromatosis type 1. A 21-year-old man who had been diagnosed with neurofibromatosis type 1 was admitted to our hospital with the chief complaint of a palpable abdominal mass. Abdominal computed tomography revealed a huge heterogeneous tumor measuring approximately 17 cm in diameter occupying the left retroperitoneal space, and numerous metastatic lesions between the left psoas muscle and the left thigh with dissolution of the left hip joint. After the diagnosis of a retroperitoneal malignant neurogenic tumor associated with NF-1, a laparotomy was performed. A giant, firm, whitish, well-capsulated tumor was found adherent to the psoas muscle and to involve the abdominal aorta, there was no gross invasion of other adjacent organs. Resection of the main tumor and of the nodules in the retroperitoneal space that were suspected to be metastatic lesions, were found between the left psoas muscle and the left thigh with dissolution of the left hip joint (Figure 2). On magnetic resonance imaging, the tumor was visualized as a low intensity signal on T1-weighted images and as a high intensity signal on T2-weighted images.

After the diagnosis of retroperitoneal malignant neurogenic tumor associated with NF-1, a laparotomy was performed. A giant, firm, whitish, well-capsulated tumor was found occupying the left abdomen (Figure 3). Although the main tumor appeared to be tightly adherent to the psoas muscle and to involve the abdominal aorta, there was no gross invasion of other adjacent organs. Resection of the main tumor and of the nodules in the retroperitoneal space that were suspected to be metastatic lesions and reconstruction of the abdominal aorta were conducted.

Macroscopically, the extirpated main tumor measured 19x15x13 cm in size and was encapsulated. The cut surface of the tumor revealed solid and yellowish tissue in the peripheral portion and foci of hemorrhage and necrosis in the central portion of the tumor. Histopathological examination revealed spindle-shaped cells with wavy nuclei and slightly eosinophilic cytoplasm, showing dense proliferation and forming storiform structures, with a myxomatous stroma (Figure 4A). On immunostaining, the spindle-shaped tumor cells showed a positive response for S-100 protein and NSE, with sporadic distribution of rhabdomyoblastic cells that showed positive staining for myoglobin and desmin (Figure 4B). Immunohistochemical staining with S-100 and desmin indicated that the tumor cells originated from Schwann cells and showed rhabdomyosarcomatous differentiation. Based on these findings, a diagnosis of MTT associated with MPNST was made.

After surgical treatment, transarterial infusion chemotherapy was administered via the left common iliac artery for the remnant tumors in the left hip joint and the left thigh, and the patient was discharged five months after the surgery. However, eight months after being discharged, the patient was readmitted for dyspnea found to be caused by lung metastasis. Despite administration of transarterial chemotherapy, the tumors increased greatly in size, and the patient died 14 months after the surgery. An autopsy was not performed.

Figure 1. Abdominal computed tomography showing a huge tumor occupying the left retroperitoneal space and a metastatic mass in the left psoas muscle (arrows).
NF-1 is an autosomal dominant disorder characterized by café-au-lait spots, cutaneous neurofibromas, skeletal dysplasias, Lisch nodules, and sometimes malignant tumors, and it occurs with an incidence of approximately one in 3000 live births.1 MPNSTs are well known to arise from major and minor peripheral nerves or within pre-existing neurofibromas, and approximately 50 percent of patients with MPNSTs have NF-1.2,3 The World Health Organization coined the term MPNST in place of the previously used, often confusing, terminology such as “malignant schwannoma,” “malignant neurilemmoma,” “neurogenic sarcoma,” and “neurofibrosarcoma.”4 The precise cell of origin of MPNSTs has not yet been conclusively identified, although the Schwann cell is thought to be the major candidate cell for this. The overall five-year survival rate in patients with MPNSTs has been reported to be 34-44%, and the association of these tumors with NF-1 is predictive of a poor prognosis.2,3 The capacity of MPNSTs to undergo focal divergent differentiation is well known, and tumors showing rhabdomyosarcomatous differentiation, referred to as MTTs, are identified in approximately 12 percent of patients with MPNSTs.5

Woodruff et al. (1973) reported a series of 10 cases of malignant schwannoma with rhabdomyoblastic differentiation, which they were the first to refer to as MTTs.6 Immunohistochemical staining for skeletal muscle markers such as myoglobin or desmin is essential for making a correct diagnosis of MTT, because of the histological variations of MPNSTs. The average age of patients with MTTs is 31.7 years and these tumors occur with approximately equal frequency in males and females.6 The reported incidence of MTTs occurring in association with NF-1 is in the range of 44-69. Several authors have reported that MTTs are associated with a worse prognosis than MPNSTs, regardless of whether or not they are associated with NF-1,6,7 with a five-year survival rate of 12-26%.6,7

The reason for the more aggressive behavior of MTTs than classic MPNSTs still remains unclear. MTTs occur predominantly in the trunk, head and neck, and lower extremities.6 According to a review of 75 cases of MTTs by Yakulis et al. (1996), the tumor was located in the retroperitoneum in only one case (1.3%). Subsequent to the publication of this review, we could identify only one case of retroperitoneal MTT by a PubMed search of the English language literature using the terms “retroperitoneum” or “retroperitoneal” and “Triton.”7 All the three cases of MTTs developing in the retroperitoneal space reported so far, including the present case, presented with a huge abdominal mass,8 since these retroperitoneal tumors are often asymptomatic in the earlier stages.

Like most soft-tissue sarcomas, MTTs traditionally are insensitive to chemotherapy and radiotherapy. However, several authors have reported recently that repeated resection combined with chemotherapy and/or radiotherapy against recurrent MTTs might prolong the survival in patients with MTTs.10,11 Because MPNSTs and MTTs have a high malignant potential, patients with NF-1 should be followed by periodic check-ups, including computed tomography for early detection of such tumors as these, and multidisciplinary treatments including complete resection are required for patients with MTTs.

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