Unusual histological variant of malignant peripheral nerve sheath tumor with rhabdomyoblastic differentiation

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Abstract

Malignant peripheral nerve sheath tumor (MPNST) with rhabdomyoblastic differentiation is called as malignant triton tumor (MTT). It is highly aggressive soft tissue tumor with higher local recurrence rate. MTT has poor prognosis than MPNST. MTT seems to be more aggressive in patients with neurofibromatosis (NF-1). We herein, reporting an interesting case of 55 years male with multiple neurofibromas all over the body since 30 years and multiple café-au-lait spots, diagnosed as NF-1. Since 6 years, he had an enlarged mass in left thigh. Wide excision of mass was done. On histopathological examination revealed the diagnosis of MTT and diagnosis of which was confirmed on immunohistochemistry.

Key words: Malignant triton tumor; Neurofibromatosis-1; Desmin; S-100 protein

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Core tip: Meticulous histopathological examination along with immunohistochemistry is the mainstay to arrive at such rare histological diagnosis. The surgical pathologist should keep in mind such rare entity while dealing with such kind of patients.

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INTRODUCTION

Malignant peripheral nerve sheath tumor (MPNST) is an unusual type of soft tissue sarcomas. It accounts for about 5%-10% of all soft tissue sarcomas[1]. Amongst them malignant triton tumor (MTT) constitutes about 5% of all MPNSTs[12]. MTT is a subtype of MPNST that...
Peripheral nerve malignant lesions are displaying a spectrum of histopathological features. These tumors typically arise from peripheral nerves and can mimic other soft tissue sarcomas. Immunohistochemical analysis is crucial for accurate diagnosis, as these tumors often show positivity for skeletal muscle markers such as desmin, myoglobin, or muscle actin. Malignant peripheral nerve sheath tumor (MPNST) is a rare sarcoma that arises from the peripheral nerves, and it has a poor prognosis compared to other soft tissue sarcomas.

### CASE REPORT

A 55-year-old male patient presented with multiple swellings all over the body ranging from 1-3 cm since 30 years (Figure 1). These nodules were histopathologically reported as neurofibromas. A subcutaneous nodule in the left thigh was enlarged to attain a present size of 8 cm × 4 cm × 3 cm. He had café-au-lait spots on the trunk ranging from 0.5-1.8 mm. On family history, the patient's mother, sister, and brother had similar complaints. So he was diagnosed as a case of NF-1. Ultrasonography was suggestive of neoplastic lesion mostly soft tissue sarcoma. Fine needle aspiration cytology was done and reported as malignant soft tissue tumor with possibilities of malignant fibrous histiocytoma and rhabdomyosarcoma (Figure 2A). Complete resection of the mass was done and the specimen was sent for histopathological evaluation.

On gross examination, a mass of 8 cm × 4 cm × 3 cm in size, covered with skin. Cut surface was soft, pale grey in colour with areas of necrosis. Histopathological examination showed dense areas of malignant spindle shaped cells with oval nuclei with prominent mitoses, marked nuclear pleomorphism along with foci of necrosis. Many nuclei were bizarre and hyperchromatic. Alternating with hypercellular Antoni A areas, there were hypocellular myxoid areas called as- Antoni B areas. Thick walled congested blood vessels were found. Interspersed within it are seen many Scattered round cells with abundant eosinophilic cytoplasm with atypical nuclei, which were recognized as rhabdomyoblasts (Figure 2B).

The surgical cut margins were free of tumor invasion. On Immunohistochemical evaluation, the spindle cells showed focal S-100 positivity (Figure 3A) and cells with deeply eosinophilic cytoplasm (rhabdomyoblasts) showed positivity for Desmin (Figure 3B).

After histopathological confirmation, the patient was under treatment with radiotherapy and doing well without any recurrence on follow-up since last 6 mo till date.

### DISCUSSION

Peripheral nerve malignant lesions are displaying a wide range of histological features. The high frequency of grade III histology in this sarcoma is significant, indicating an aggressive behavior. MPNST constitutes 5%-10% of all soft tissue sarcomas, and about one fourth to one half occur in the setting of neurofibromatosis type 1 (NF-1). Patients with NF-1 have a higher propensity to transform into sarcoma after a prolonged latent period (10-20 years). Our patient was diagnosed as having NF-1 and after long latency period of 30 years, he developed MPSNT with rhabdomyoblastic differentiation. This poor outcome is mainly attributed to the high frequency of grade III histology in this sarcoma.

MTT is a rare tumor arising from peripheral nerves. It is an autosomal dominant disorder. It has a strong association with neurofibromatosis (type 1). The common sites of occurrence are head, neck, extremities and trunk. The symptoms are mainly attributed to mass effect giving rise to neurological signs and symptoms. In 1973 Woodnelf et al proposed the classification of MTT by establishing three criteria for diagnosis: (1) Tumor with peripheral nerve involvement in a patient with NF-1; (2) Majority of the cells in the tumor are Schwann cells; and (3) Presence of Rhabdomyoblasts.

Our patient had NF-1 and histology showed all the above mentioned criteria. The pathognomonic feature of this tumor is the presence of rhabdomyoblasts. The number of rhabdomyoblasts varies from area to area in the same tumor. They are having abundant eosinophilic cytoplasm. Desmin is demonstrated in the rhabdomyoblasts.

The histogenesis of this unusual tumor is discussed by Masson. He postulated that both cell lines have similar origin, i.e., from less well differentiated neural crest cells. The strong relation between neural
tissues and rhabdomyoblastic differentiation has been reported as the development of skeletal muscle differentiation within other neural tumors such as Ocular Medulloblastoma[8], Ganglioneuroblastoma[9,10]. The five years survival rate for MTT is only 11% in contrast to 39% for MPNST[11]. MTT is significantly worse than the usual MPNST. The aggressiveness of MTT is attributed to high grade (grade Ⅲ) nuclear features with high proliferative capacity[4]. Radical excision followed by high dose radiotherapy is the conventional treatment for this unusual tumor[3].

MTT is an uncommon sarcoma which is having high propensity of local recurrence and distant metastases. Histopathologically, the diagnosis of MPNST with mesenchymal differentiation is difficult. So meticulous histopathological examination and immunohistochemical demonstration of neural markers and skeletal muscle markers help to hit the correct diagnosis. Early diagnosis, complete resection of the tumor followed by radiotherapy can help to increase survival of the patient.

**COMMENTS**

**Case characteristics**
A 55-year-old male patient presented with multiple swellings all over the body ranging from 1-3 cm since 30 years.

**Clinical diagnosis**
Multiple neurofibromas.

**Differential diagnosis**
Neurofibromas, fibrosarcoma, malignant peripheral nerve sheath tumor (MPNST).

**Laboratory diagnosis**
Fine needle aspiration cytology, histopathology with Immunohistochemistry showing Desmin positivity in rhabdomyoblasts.

**Imaging diagnosis**
Ultrasonography was suggestive of neoplastic lesion soft tissue sarcoma.

**Pathological diagnosis**
An unusual histological variant of MPNST with rhabdomyoblastic differentiation.

**Experiences and lessons**
Careful histological examination along with clinical work up is important to arrive at such unusual diagnosis.

**Peer-review**
Good overview of the diagnostic challenges in the correct interpretation of this rare tumor.

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