

Malignant peripheral nerve sheath tumor of retroperitoneum involving small and large bowel: A rare case report

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ABSTRACT

Malignant peripheral nerve sheath tumors (MPNSTs) are rare and aggressive, soft-tissue sarcomas having a high rate of recurrence which largely occur in the extremities and the head-and-neck region. Gastrointestinal (GI) MPNSTs are rare. To date, only a few cases have been reported. We report the case of a 50-year-old man who presented with pain and a lump in his abdomen for 1 year. Clinically, the case was thought to be retroperitoneal neurofibroma. A computed tomography scan revealed an intra-abdominal large well-defined lobulated mass predominantly on the left side. Exploratory laparotomy and resection of the tumor were done as large and small bowel mesentery masses. Histopathological examination showed a spindle cell neoplasm composed of interlacing fascicles of slender to plump spindle cells with wavy buckled nuclei. Cells showed moderate to marked anisonucleosis with hyperchromatic and vesicular nuclei with prominent nucleoli along with many pleomorphic cells with multiple nuclei and many mitosis. The tumor seemed to be infiltrating the wall of the small bowel and colon. On immunohistochemistry, tumor cells were positive for vimentin, focally positive for S100, and negative for smooth muscle actin. Thus, the diagnosis of MPNST was confirmed. This case highlights that, although rare, the possibility of MPNST of retroperitoneum should be considered as a differential diagnosis in an unexplored retroperitoneal mass.

Key words: Colon, Malignant peripheral nerve sheath tumor, Retroperitoneum, Sarcoma

Malignant peripheral nerve sheath tumors (MPNSTs) are malignant neoplasms arising from the peripheral nerve. These tumors arise from or differentiate from peripheral nerve sheath cells [1]. The incidence of these tumors is 1/100,000, which corresponds to 3–12% of soft-tissue sarcomas [2]. GI MPNSTs are rare. Only 1% of MPNSTs occur in the retroperitoneal region. The age at presentation of MPNSTs is in the 3rd–6th decades [3]. The World Health Organization coined the term “MPNST” and replaced the previous terminology on tumors of neurogenic origin with similar biological behavior such as malignant schwannoma, malignant neurilemmoma, and neurofibrosarcoma in 1993 [1]. It can arise as a sporadic form with an incidence of 0.0001% in general population or can be associated with neurofibromatosis-1 (NF-1) with incidence of 2–5% [2]. They commonly arise on the trunk, extremities, head-and-neck, and paravertebral region [2]. Only very few cases of MPNSTs of the GI tract have been reported in the literature [4].

CASE REPORT

A 50-year-old man presented with a large abdominal mass that had been progressively increasing in size for 1 year. He complained of significant weight loss of approximately 6 kg in 3 months. There was no history of similar complaints or any intervention in the past. The remaining medical history, family history, and psychosocial history were unremarkable.

The vitals of the patient were within normal range. On per abdominal examination, a 20×15 cm lobulated mass was palpated extending from the left hypochondrium to the infraumbilical region laterally up to both flanks. The overlying skin was unremarkable.

Ultrasonography of the abdomen showed a hypoechoic mass 15 cm in diameter in the left hypochondrium to the infraumbilical region. Computed tomography scan revealed an intra-abdominal large well-defined lobulated mass of size 13.7×14.9×24.5 cm predominantly on the left side with abutments and mass effect in the adjacent organs (Fig. 1). A tru-cut biopsy from the abdominal mass was performed and diagnosed as neurofibroma.

A wide local excision was performed, and the specimen was sent for histopathological examination as a large and small bowel

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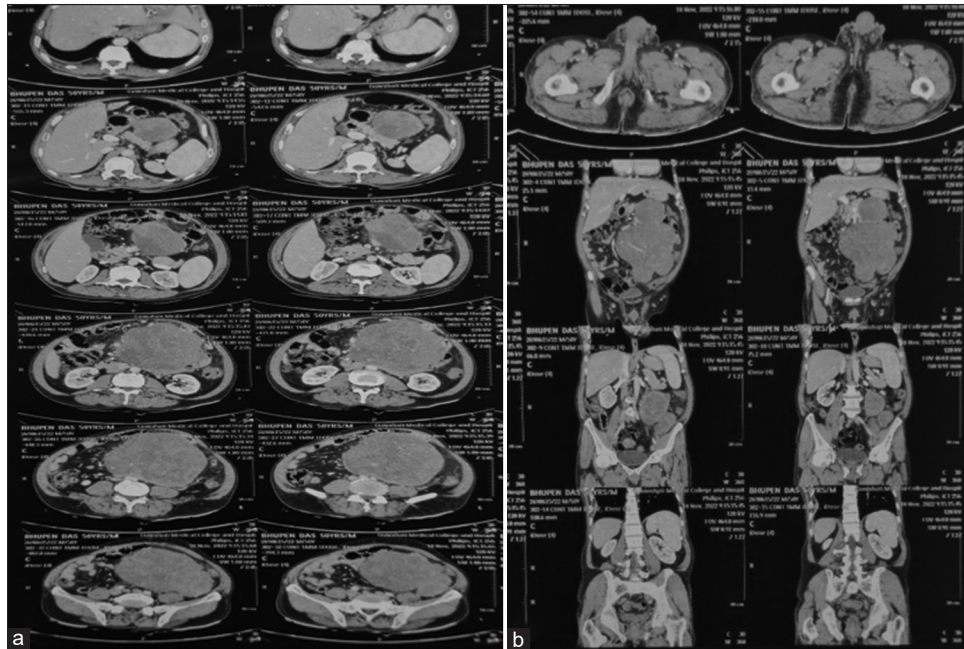


Figure 1: (a and b) Computed tomography scan showing a large, well-circumscribed, heterogeneous mass with abutments and mass effect in the adjacent organs

mesentery mass. On gross examination, the tumor was large and globular, measuring 22×18×13 cm. The cut section was solid, whitish with a few yellowish myxoid areas. The tumor seems to infiltrate the serosal layer of the colon and small intestine (Fig. 2).

Multiple sections taken from both tumors show a spindle cell neoplasm composed of interlacing fascicles of slender to plump spindle cells with wavy buckled nuclei in many places. Areas of myxoid change and necrosis were seen in many places. There are hypo and hypercellular areas within the tumor. Cells in the hypercellular zone show moderate to marked anisonucleosis with hyperchromatic and vesicular nuclei with prominent nucleoli along with many pleomorphic cells with multiple nuclei. The mitosis was 12/10 hpf (Fig. 3). All margins including proximal and distal resection margins of the large and small bowel were free of tumor. The tumor cells showed positivity for vimentin and focally positive for S100. Ki67 was found to be 30% (Fig. 4).

Hence, a final diagnosis of MPNST of the retroperitoneum involving the small and large bowel was given. The resected margins, as well as, resected lymph nodes were free of the tumor. Postoperatively, he was discharged and advised to review with medical and radiation oncologists. The patient did not receive any adjuvant treatment. He is presently doing well and has been under follow-up.

DISCUSSION

Primary retroperitoneal neoplasms are rare lesions accounting for 0.1–0.2% of all malignancies. Only 1% of MPNSTs occur in the retroperitoneal region [5]. MPNST is a highly aggressive soft-tissue sarcoma. It occurs more in association with NF1 (8–13%) and sporadic cases are rare (0.001% cases). MPNSTs may arise spontaneously in adult patients in the 3rd–6th decade of life, although 5–42% of these tumors have an association with type-I

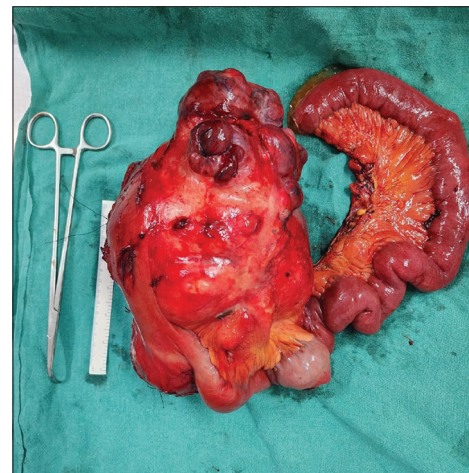


Figure 2: Gross specimen showing a large, globular tumor. Cut section is solid, whitish with few yellowish myxoid areas. The tumor seems to infiltrate the serosal layer of colon

multiple NF [6,7]. In our study, the patient did not have a history of NF1 and developed the condition at 50 years of age. The tumor typically originates from the nerve sheaths of the major nerves in the extremities (40%), trunk/retroperitoneal (38%), and head-and-neck region (21%) [8]. In our case, the tumor originated in the retroperitoneum.

The clinical symptoms of these tumors of the GI tract are non-specific, including abdominal pain (63%), emesis (43%), weight loss (44%), and GI bleeding (23%) [7]. Patients with MPNST of the GI tract clinically present with weight loss, fatigue, vomiting, or palpable mass, and as a result, can be diagnosed late in its course [1]. In our case also, the patient presented with weight loss.

Morphologically, MPNST is usually composed of monomorphic elongated cells in a hypocellular stroma with necrosis and frequent mitotic figures. Rhabdomyoblastic, metaplastic cartilage, or bone features may be present. Spindle cell is the most commonly

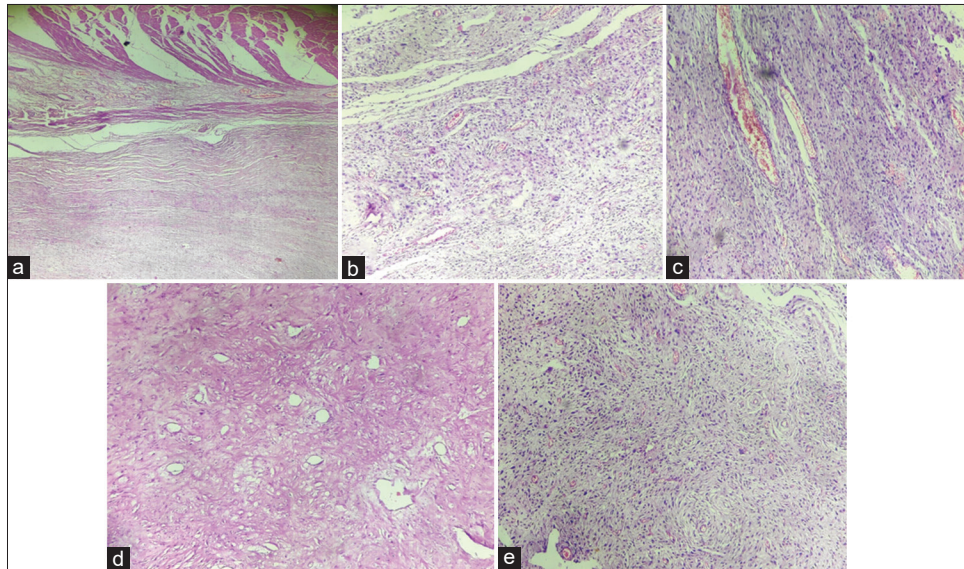


Figure 3: Photomicrograph showing (a) a tumor attached to the serosal layer of the colon whereas the muscular layer of the colon is free; (b and c) Hematoxylin and eosin sections showing sheets and fascicles of slender and pump spindle-shaped tumor cells. Tumor cells having elongated hyperchromatic nuclei and mild to moderate amount of cytoplasm, showing moderate degree of pleomorphism; (d and e) Photomicrograph showing necrosis and atypical mitotic figures

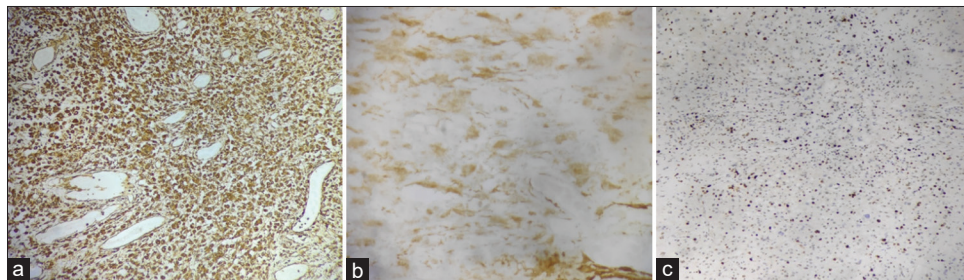


Figure 4: Immunohistochemistry showing (a) diffuse positivity for Vimentin; (b) focally positive for S100; (c) Ki67 30%

encountered histologic subtype (71.8%), followed by Triton (17.7%) followed by epithelioid MPNSTs (<5%). MPNSTs express the neural marker S-100 in 50–60% of cases. Other neural markers such as CD99, neuron-specific enolase, FLI-1, CD56, myelin basic protein, Leu-7, and protein gene product 9.5 may be positive. Among mesenchymal markers, vimentin is usually positive. Although MPNSTs generally do not express actin and desmin, the rare Triton tumor with rhabdomyoblastic elements may express desmin. Epithelial markers such as cytokeratins and CEA are negative except in the rare epithelioid variant which may express epithelial membrane antigen. A weak S-100 staining in MPNST is also associated with undifferentiated tumors and a five-fold higher risk of distant metastasis [1].

Wide resection of the primary tumor with negative margins is the treatment of choice in localized disease with adjuvant radiation therapy reserved for cases with positive margins or recurrent disease or bulky disease (>5 cm). Neoadjuvant chemotherapy or radiation may be considered in locally advanced or unresectable MPNST to make the disease more amenable to local treatment.

MPNST is an aggressive tumor showing a poor prognosis along with death occurring in 63%, usually within 2 years of diagnosis [9]. The prognosis for abdominal MPNST remains unclear. The presence of NF1 syndrome, female sex, deep location, positive margins, lack of adjuvant chemotherapy, lack of adjuvant

radiation therapy, and high-grade tumors were all associated with adverse disease-free survival in a recent review [10].

CONCLUSION

The prognosis of patients who develop MPNST of the retroperitoneum has been considered grave, and thus, accurate diagnosis has substantial significance. The optimal treatment of MPNST is not well established owing to its rare occurrence. The current standard of care for MPNST is complete surgical resection with wide negative margins, with the role of chemotherapy and radiotherapy still under evaluation. Proper management of such tumors requires a multidisciplinary approach.

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