Malignant peripheral nerve sheath tumor arising from plexiform neurofibroma of the mesentery in a patient with neurofibromatosis 1

An unusual case of a 34-year-old woman with a malignant peripheral nerve sheath tumor (MPNST) associated with neurofibromatosis type 1 (NF1), arising from plexiform neurofibroma of the mesentery is presented here. She presented with complaints of abdominal pain of 10 days duration. Imaging revealed the presence of multiple nodules along with a large mass in the mesentery of the small bowel. Small bowel resection along with the mesenteric mass was done and histopathologic examination showed MPNST arising from a plexiform neurofibroma of the mesentery. A review of the literature showed that till date only three cases of MPNST arising from neurofibroma of the mesentery in NF1 patients have been reported out of which one was from a plexiform neurofibroma.

Key words: Malignant peripheral nerve sheath tumor, mesentery, neurofibromatosis 1, plexiform neurofibroma

INTRODUCTION

Malignant peripheral nerve sheath tumor (MPNST) is an aggressive and uncommon neoplasm that develops within a peripheral nerve, most of which are associated with neurofibromatosis type 1 (NF1).[1] The paraspinal region of the abdomen, extremities, and head and neck region are the most common locations for MPNST in NF1 patients.[2] MPNST arising from the mesentery is very unusual and only three cases have been reported till date associated with NF1.

CASE REPORT

A 34-year-old woman presented with dragging abdominal pain of 10 days duration. The patient had clinical stigmata of NF1. Patient’s mother had numerous neurofibromas all over her body [Figure 1]. Genetic analysis had not been performed.

The patient was admitted and evaluated for her complaints. Her hemogram, renal, liver and bleeding parameters were normal. Computed tomography scan of abdomen showed a lobulated mixed density lesion in the mid and lower mesentery. Bilateral renal calculi were also found. The patient was posted for surgery and small bowel resection with anastomosis and appendectomy were done.

Histopathological examination showed a 40-cm long segment of small intestine with attached mesentery. The mesentery showed multiple nodular masses with the largest measuring 9 × 8 × 6 cm, 1 cm away from the bowel wall [Figure 2a]. The cut surface of the large mass showed a variegated appearance with solid grey white, myxoid and hemorrhagic areas [Figure 2b]. The smaller nodules were clustered together and the cut surface showed a whitish glistening appearance. The mucosal surface of the small intestine was normal with no nodules or ulceration. Resected margins of the intestine were free of tumor infiltration.

Sections from the largest nodule showed a spindle cell neoplasm with dense cellular and hypocellular areas with areas of myxoid change, necrosis and hemorrhage [Figure 3]. The tumor cells were...
arranged in cellular interlacing fascicles and had wavy buckled nuclei which in some areas were plump and spindle shaped. There was a proliferation of tumor cells around thin walled blood vessels. Mitotic figures were increased (4-5/hpf). Sections from the smaller nodules showed the features of a plexiform neurofibroma. Based on these features, a differential diagnosis of gastrointestinal stromal tumor/MPNST arising from a plexiform neurofibroma was made. Immunohistochemistry showed focal positivity for S-100, and negativity for smooth muscle actin, CD117 and DOG1. Ki 67 index was high (30%) [Figure 4]. The postoperative period was uneventful and the patient was discharged and is now under follow up.

**DISCUSSION**

Neurofibromatosis 1 is one of the most common genetic disorders affecting about 1 in 3000 population. Common abdominal neoplasms in NF1 patients are localized and plexiform neurofibromas which occur in the para spinal and sacral region. The majority of abdominal neurofibromas are asymptomatic. The neurofibromas which involve abdominal viscera and the mesentery in NF1 patients are of the plexiform type. Mesenteric neurofibromas usually present as multiple nodules involving the mesentery.  

MPNSTs are rare soft-tissue tumors accounting for 5-10% of all soft tissue tumors, and up to 50% of these tumors are found in patients with NF1. The extremities are the most common sites of involvement by these tumors. In these patients the cumulative life-time risk of developing MPNSTs is considered to be about 10% and may be as high as 30% in those with symptomatic plexiform neurofibromas.  

Most MPNSTs arise in relation to major nerve trunks, like the sciatic nerve, brachial plexus and sacral plexus with the most common sites of occurrence being the proximal portions of the upper and lower extremities and the trunk. In the setting of NF1, MPNST originates from a peripheral nerve sheath or plexiform neurofibroma. MPNST arising in an extremity most commonly manifests as a painful mass. In contrast, those tumors that arise in the abdomen and retroperitoneum are often clinically silent. MPNST arising from the mesentery is very rare with only six cases having been reported in the literature. Of these, three cases were associated with NF1 as mentioned in Table 1. Among the cases arising in the setting of NF1 only one study mentions association with plexiform neurofibroma.  

![Figure 1: Clinical manifestations of neurofibromatosis 1 in the patient (a) and her mother (b)](image1)

![Figure 2: (a) Segment of small intestine with attached mesentery showing a large nodule with adjacent smaller nodules. Cut surface of smaller nodule is whitish and glistening (yellow arrow). (b) Variegated appearance of larger nodule](image2)

![Figure 3: (a) Scanner view of the malignant tumor with an adjacent neurofibroma (H and E x40). (b) Spindle cells with high mitotic count (H and E x400)](image3)

![Figure 4: CD117 (a), DOG1 (b) and smooth muscle actin (c) showing negativity. Ki-67 (d) and S-100 (e) showing focal positivity. S100 positivity (f)](image4)
CONCLUSION
Malignant peripheral nerve sheath tumor (MPNST) is an aggressive neoplasm and when associated with NF1, it has poor prognosis. Paraspinal region of the abdomen, extremities and head and neck region are the common sites in patients with NF1. Mesenteric MPNST is very rare in occurrence. As MPNSTs of the mesentery region are the common sites in patients with NF1. Mesenteric paraspinal region of the abdomen, extremities and head and neck neoplasm and when associated with NF1, it has poor prognosis. Malignant peripheral nerve sheath tumor (MPNST) is an aggressive neoplasm and when associated with NF1, it has poor prognosis. Other poor prognostic factors include size ≥5 cm, incomplete resection and location in a nonextremity. MPNST is the main cause of death in NF1 and occurs in 8 to 13% of NF1 patients during their life span.

REFERENCES

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