Malignant Peripheral Nerve Sheath Tumour of Small Intestine, an Unusual Tumour: A Case Report and Review of the Literature
Metta Parvathi1, Konkena Janardhana Rao2, Gunta Santa Rao3, Kollu Srinubabu4 and Prasad Neelam5*

ABSTRACT
Neurofibromatosis type-1 (NF1), also known as von Recklinghausen disease, is a common autosomal dominant condition. It is a multisystem disorder. Abdominal involvement in patients with NF1 has been described in the form of neurofibromas within the liver, mesentery, retroperitoneum and gastrointestinal (GI) tract. Involvement of bowel is rarely seen. Malignant peripheral nerve sheath tumour of small intestine is a rare entity with very few cases reported in literature. Surgery remains the only curative option. Chemotherapy may be used as an adjuvant for advanced diseases. We report a case of malignant peripheral nerve sheath tumour of small intestine which presented as acute intestinal obstruction.

KEYWORDS: Neurofibromatosis type-1(NF1), Malignant peripheral nerve sheath tumour, Acute intestinal obstruction

INTRODUCTION
Neurofibromatosis presents usually with classical triad of cutaneous pigmentary lesions, tumours of the skin and multiple tumours of peripheral nerves. The incidence of malignant peripheral nerve sheath tumours is 1/100,000 corresponding to 3–12% of soft tissue sarcomas[1]. They are commonly seen arising on the trunk, extremities, head and neck, and paravertebral region[2]. Benign neurofibromas undergo malignant transformation in 5–15%, especially in patients above the age of 40 years[3]. Involvement of small bowel is very rare. In the literature, there are reports of 14 cases of neurofibrosarcoma of small bowel, of which three arose in the duodenum and the remainder in the jejunum or ileum[4]. Herewith, we report a case of malignant peripheral nerve sheath tumour arising from the ileum.

CASE REPORT
A 56-year-old female patient presented with pain abdomen, abdominal distension and multiple episodes of foul smelling and bilious vomiting of 2 days duration. On examination, there were neurofibromas present all over the body associated with café-au-lait spots [Figure 1] which were present since childhood. Abdomen was distended with diffuse tenderness. Bowel sounds were exaggerated.

Hematological and biochemical investigations were within normal limits. X ray erect abdomen showed
dilated small bowel loops with multiple air fluid levels. Ultrasonography of abdomen revealed a mass lesion in the lower abdomen adherent to the small bowel with dilated small bowel loops. Contrast enhanced computerized tomography (CECT) of the abdomen was suggestive of a mass lesion in the pelvis adherent to small bowel causing dilatation of jejunal loops.

On exploration of abdomen, there was a tumour of size 12 × 10 × 8 cm present at the ant mesenteric border of the mid ileum causing intestinal obstruction. Proximal ileum and jejunum were dilated and distal ileum was collapsed. There were multiple nodules over the serosal surface of the small intestine. Resection of the involved segment of the intestine with 10 cm margin on either side was done. End to end ileo-ileal anastomosis was done. Post-operative period was uneventful and patient was discharged in good general condition.

On gross examination, it was a well circumscribed, firm, grey white mass with multiple hemorrhagic, friable and necrotic areas with cystic spaces.

Histological examination of the resected mass with ileum showed multiple spindle shaped cells with elongated and wavy nuclei. Nuclei are enlarged, hyperchromatic and they show atypia. There is moderate increase in cellularity with minimal mitotic activity. There is extensive hemorrhage with necrosis. These features are suggestive of low grade malignant peripheral nerve sheath tumour arising from ileum. Resected margins were free from tumour. Two lymphnodes showed reactive hyperplasia. Sections from separate nodules showed features suggestive of neurofibroma.

DISCUSSION

Malignant peripheral nerve sheath tumour is a rare variety of soft tissue sarcoma of ectomesenchymal origin. Malignant peripheral nerve sheath tumour may arise spontaneously in adult patients in the third to sixth decade of life. The main features of NF1 disease are cutaneous and subcutaneous neurofibromas; similar multiple tumours are also found along the deeper nerves and in the mucous and serous membranes and viscera. The clinical symptoms of these tumours of the gastrointestinal tract are nonspecific, including abdominal pain (63%), vomiting (43%), weight loss (44%), and gastrointestinal bleeding (23%). Majority of these malignant tumours, develop in the preexisting neurofibromatous nodules.

Histological features are palisading arrangement, bizarre giant cells, nuclear atypia, mitotic figures, and necrosis. These tumours have morphological heterogeneity, and staining analysis reveals highly cellular spindle cell tumour in fascicles. S-100 protein is highly characteristic of neural-derived neoplasms. However, S100 protein immunoreactivity is detected in only 50–60% of malignant peripheral nerve sheath tumour and is also expressed in a range of other tissues and tumour types. Different markers are used to exclude other spindle cell tumours. Desmin and α-SMA are used to exclude smooth muscle tumours, and CD34 and CD117 (c-kit) are used to exclude GIST.
is known for this tumour in other locations of the body[3]. The current recommendations say, radical surgical treatment with wide excision for malignant peripheral nerve sheath tumour is the treatment of choice. Adjuvant radiotherapy or chemotherapy should be reserved for cases of positive margins, recurrent disease, or when wide local excision is unfeasible[2]. The strongest independent predictors of survival were primary versus recurrent disease, tumour size, tumour site, and margin status[10].
CONCLUSION

Neurofibromatosis type 1 is a common autosomal dominant disorder. Gastrointestinal involvement is rare. Malignant transformation of gastrointestinal neurofibromas is uncommon. Malignant peripheral nerve sheath tumour of the ileum is a rare entity with fewer than 14 cases reported in literature. Because of its rarity, a high index of suspicion is required for the diagnosis of these tumours. Imageology not only helps in the diagnosis and location of these tumours but also helps in the follow up of these patients. Wide excision of the tumour is the treatment of choice. Chemotherapy and radiotherapy has only an adjuvant role.

REFERENCES


