A 45-year-old lady without pathological antecedents for NF1 was admitted with pain in the right lower abdomen and multiple episodes of bilious vomiting for three months. Past history was unremarkable except for presence of controlled hypertension and Type 2 Diabetes Mellitus. On clinical examination, she had tender right iliac fossa with no palpable lump in the abdomen. Preoperatively, intussusception was diagnosed in the small bowel with USG and CECT abdomen showing characteristic “Target sign” suggestive of intussusception. On laparotomy, ileo-ileo intussusception (proximal ileum telescoping into distal ileum) was found 2 feet proximal to ileo-caecal junction with surrounding inflamed mesentery and presence of intraluminal tumour as lead point. Resection of involved segment of ileum along with its mesentery was done followed by ileo-ileo anastomosis. Histopathology was suggestive of high grade MPNST. The follow-up period has been uneventful. Written informed consent was obtained from the patient for publication of this case report and accompanying images.

**DISCUSSION**

Malignant peripheral nerve sheath tumour (MPNST) is defined as tumour arising from a peripheral nerve or exhibiting neural differentiation on histology. Proximal portions of the upper and lower extremities and the trunk are the most common sites of occurrence. Around 50% are associated with Neurofibromatosis Type 1 (NF1) with incidence of two to five per cent in patients with NF1. The estimated incidence in general population without NF1 is 0.0001% of which gastrointestinal MPNST are extremely rare. A 45-year-old lady without pathological antecedent for NF1 was admitted with pain in right lower abdomen and multiple episodes of vomiting for 3 months. Preoperatively intussusception was diagnosed in the small bowel with USG and CECT abdomen showing characteristic “Target sign.”

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**Keywords:** Gastrointestinal MPNST, ileum, Neurofibromatosis
intussusception [7]. Imaging techniques used for evaluation includes ultrasonography, CT scan, MRI and FDG-PET contributing to easy detection, staging, therapy planning and follow up [10]. MPNST usually tends to be heterogeneous in CT attenuation and MRI signal intensity because of frequent necrosis within the mass [11]. When presented with Intussusception, characteristic “Target sign” is seen in ultrasonography and CT abdomen. It is quite impossible to predict the nature of tumour viz. benign or malignant on imaging, although in some studies Gallium citrate (67Ga) scintigraphy has been found to be helpful with tumour with malignant potential having increased Gallium citrate uptake but no uptake doesn’t rule out malignancy [12,13]. On histopathological analysis, MPNST usually show spindle cells with high mitotic index. Confirming diagnostic features include a) alternating hypocellular and hypercellular region, b) appearance of thin, wavy, comma or bullet shaped nuclei usually in hypocellular mass, c) presence of nuclear palisading, which may also be seen in leiomyosarcoma, d) presence of nerve like whorls or tactoid bodies resembling wagner-meissner corpuscles, e) prominent thick walled vasculature and presence of heterologous element. Immunohistochemistry markers for MPNST includes S-100 (in 50-90% of patients) characteristic of neural derived neoplasm, Leu 7 and Myelin base protein (in 50% and 40% of patients respectively) [14,15]. Desmin and α-SMA positivity exclude smooth muscle tumours whereas CD-34 and CD-117 (cKIT) positivity excludes GIST [16]. High levels of PS3 and Ki67 may also be related to malignancy in a peripheral nerve sheath tumour according to a study [17]. Current protocols for treatment of MPNST arising from small bowel are still undefined. Current recommendation may be radical surgical treatment with wide excision as followed elsewhere. Poor prognostic factors which may entail patient to local recurrence and distant metastasis are Size 5cm or more, incomplete resection and association with NF-1. Most case series of such tumours with poor prognosis suggest limited benefit and high morbidity of using adjuvant radiotherapy and chemotherapy. Although No Phase I & III trials are available, Doxorubicine-ifosfamide based Chemotherapy has been found to be having better outcome than placebo in some series which warrants further consideration [18].

CONCLUSION

We report a rare case MPNST presenting as ileo-ileal intussusception. High index of suspicion along with multidisciplinary approach is essential for diagnosis and treatment of these tumours. There is no definitive guideline available for treatment of small bowel MPNST, although radical surgical treatment with wide excision is universally done. Role of adjuvant radiotherapy and chemotherapy is still under debate reserved only for advanced and recurrent tumours.

REFERENCES


PARTICULARS OF CONTRIBUTORS:

1. Post Graduate Student, Department of Surgery, Lady Hardinge Medical College, New Delhi, India.
2. Senior Resident, Department of Surgery, Lady Hardinge Medical College, New Delhi, India.
3. Associate Professor, Department of Surgery, Lady Hardinge Medical College, New Delhi, India.
4. Associate Professor, Department of Surgery, Lady Hardinge Medical College, New Delhi, India.
5. Associate Professor, Department of Surgery, Lady Hardinge Medical College, New Delhi, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Ankur Verma,
134D Pocket F, GTB Enclave, New Delhi-110093, India.
E-mail: geniusav23@gmail.com

FINANCIAL OR OTHER COMPETING INTERESTS: None.