Anaemia presenting as the sole symptom in a young woman: A case of malignant peripheral nerve sheath tumour of the small intestine

Sohail Mushtaq, FRCS[1,2], Sim Sze Kiat, MS[1], Nik Azim Nik Abdullah, MS[2]

[1]Faculty of Medicine and Health Sciences, Universiti Malaysia Sarawak, Kota Samarahan, Sarawak, Malaysia, [2]Department of Surgery, Sarawak General Hospital, Kuching, Sarawak, Malaysia

SUMMARY

Malignant peripheral nerve sheath tumours (MPNSTs) with gastrointestinal involvement are uncommon. We report the case of a 22-year-old woman presenting with symptomatic anaemia requiring recurrent blood transfusions. Initial haematological evaluations revealed microcytic hypochromic anaemia. Both upper and lower endoscopies were normal, except for a positive rapid urease test result. However, computed tomography identified a tumour originating from the ileum and adherent to the urinary bladder, along with a mixed-density lesion in the right ovary. Following multidisciplinary team discussion, en-bloc resection of the ileal tumour and enucleation of the ovarian lesion, were performed. Histopathological analysis confirmed the ileal mass as an MPNST and the ovarian lesion as a benign cystic teratoma. This case highlights the rarity of ileal MPNSTs presenting with anaemia and emphasises the importance of complete surgical excision and vigilant follow-up owing to the potential for recurrence and metastasis.

INTRODUCTION

Malignant peripheral nerve sheath tumours (MPNSTs) are defined as neoplasms arising from peripheral nerves or exhibiting nerve sheath differentiation.[1] While most MPNSTs originate from major nerve trunks and are commonly located in the trunk, extremities, head, neck, or paravertebral regions, those arising from the nerves of the small intestine are exceedingly rare.[2] In this report, we present a case of ileal MPNSTs that manifested with anaemia as the primary symptom.

CASE PRESENTATION

A 22-year-old woman presented with a two-year history of progressive weakness, lethargy, anorexia, and weight loss. She also reported occasional episodes of epigastric discomfort and bloating but denied any haematemesis, melena, rectal bleeding, or other lower gastrointestinal symptoms. Her menstrual cycles were regular, and there was no family history of malignancy. The remainder of her systemic review was unremarkable.

On examination, the patient appeared alert but notably pale. She was afebrile, with a pulse rate of 80 beats per minute, blood pressure of 100/70 mmHg, and respiratory rate of 16 breaths per minute. Systemic examination revealed no palpable abdominal masses or other significant abnormalities.

Biochemical tests revealed a markedly low haemoglobin level of 4.4 g/dL, mild leucocytosis with a white blood cell count of 11.8 × 10^9/L, and an elevated platelet count of 870 × 10^9/L. Peripheral blood film and blood indices confirmed hypochromic microcytic anaemia. Results of renal profile, liver function tests, blood glucose levels, and coagulation profiles were all within normal limits. Chest radiography and electrocardiography findings were unremarkable. Oesophagogastroduodenoscopy (OGDS) showed no abnormalities, but the rapid urease test for Helicobacter pylori was positive. Similarly, colonoscopy findings were unremarkable.

She was admitted to medical ward for further assessment and blood transfusion because of the severity of her anaemia and transportation difficulties, as she lived far from the hospital. During her hospital stay, she developed a fever (38.8°C) and reported lower abdominal pain, although physical examination findings remained unremarkable. A computed tomography (CT) of the thorax, abdomen, and pelvis (Figure 1) revealed a left pelvic mass measuring 6.4 × 7.5 × 7.1 cm, originating from the small bowel and adherent to the dome of the urinary bladder. Additionally, the right ovary was found to be enlarged, with a complex mixed-density lesion measuring 4.4 × 3.3 × 2.1 cm. She was then referred to surgical team for management.

The case was reviewed in a multidisciplinary team (MDT) meeting comprising a general surgeon, gynaecologist, and radiologist. Given the primary origin of the tumour in the small bowel, the team recommended an initial laparoscopic assessment of resectability, followed by a lower midline laparotomy for definitive surgical management.

During the laparoscopic assessment, minimal ascites was noted in the pelvis, with no evidence of peritoneal seedlings. The surface of the liver appeared smooth. The tumour (Figure 2) was identified as originating from the small bowel,

This article was accepted: 19 August 2025
Corresponding Author: Sohail Mushtaq
Email: msohail@unimas.my

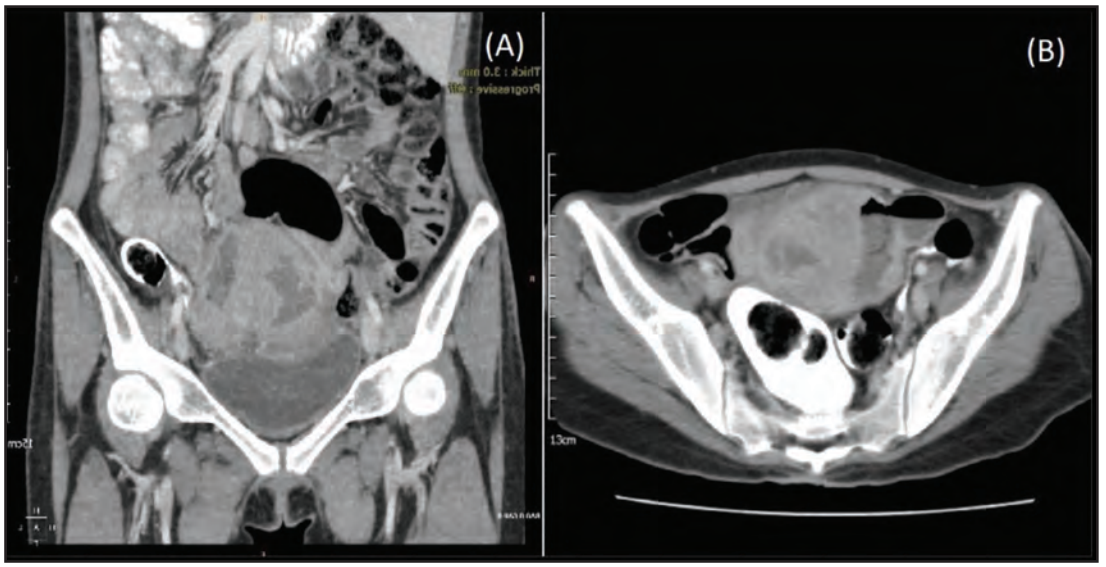

Fig. 1: CT scan of the abdomen and pelvis (A: coronal view; B: axial view) showing a pelvic enhancing tumour (blue arrow) arising from the small bowel and adherent to the dome of the urinary bladder

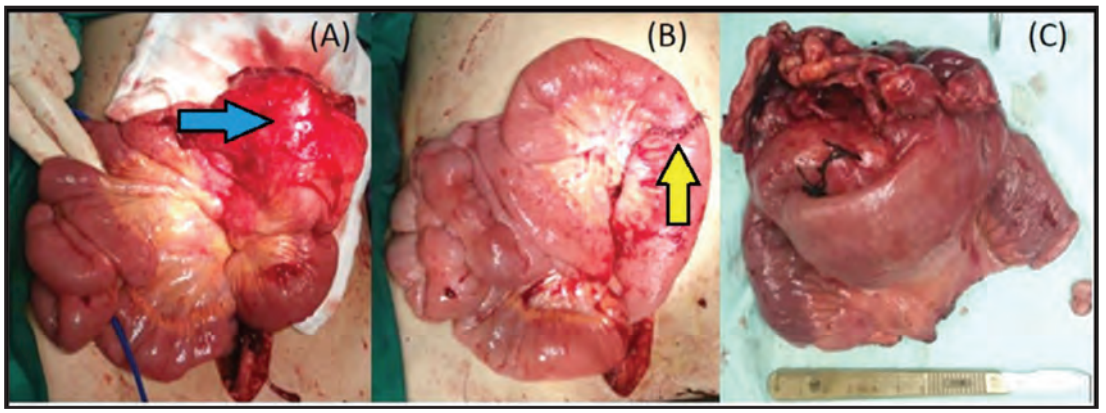

Fig. 2: A: Small bowel tumour (blue arrow) with multiple enlarged mesenteric lymph nodes; B: Primary anastomosis (yellow arrow) after en-bloc resection of the tumour; C: Resected specimen

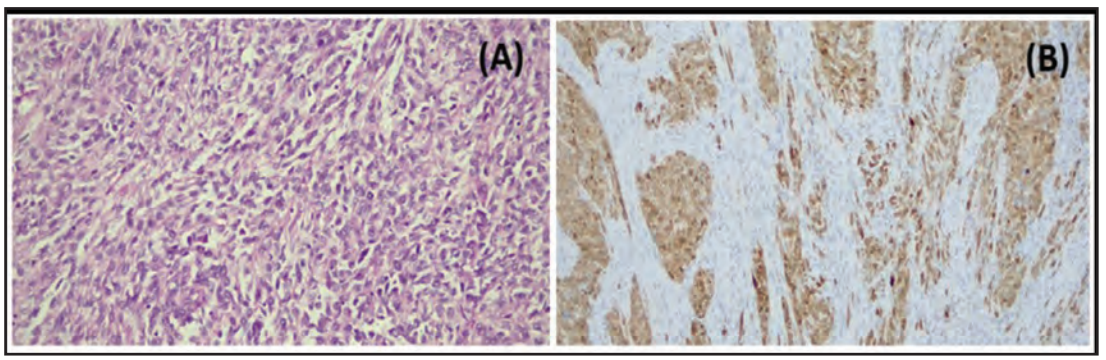

Fig. 3: A: Histopathological findings showed sheet and clumps of epithelioid tumour cells (Haematoxylin & Eosin stain); B: Positive S-100 in Immunohistochemical stain.

approximately 140 cm from the ileocecal junction, and was adherent to the dome of the urinary bladder. The greater omentum was also adherent to the tumour, and several enlarged mesenteric lymph nodes were observed adjacent to the mass. Because the tumour was operable conversion to laparotomy was performed and an en-bloc resection of the tumour was carried out, followed by primary end-to-end anastomosis of the bowel. Concurrently, the right ovarian lesion was successfully enucleated by the gynaecologist. There was no invasion of the ovary by the bowel tumour. The patient's postoperative recovery was uneventful. She passed stool and flatus by postoperative day 3 and was discharged on day 5 after tolerating oral intake well. She was doing well at her 3- and 6-month follow-up visits, with no bowel related symptoms.

Histopathological examination of the resected ileal tumour revealed a 23 cm segment of the ileum containing an annular, grey-white, fleshy mass measuring 4 cm in length and 2 cm in thickness. The tumour had narrowed the intestinal lumen to a diameter barely sufficient to admit a finger. Microscopically, the tumour consisted of sheets and clusters of epithelioid cells with clear cytoplasm, vesicular nuclei, and occasional spindle-shaped cells (Figure 3). Scattered osteoclast-like giant cells and areas of necrosis were present. The tumour infiltrated the full thickness of the ileal wall; however, mitotic figures were rare. All surgical margins were clear of malignancy, although five of the 14 resected lymph nodes showed metastatic involvement. The omentum was tumour-free. Immunohistochemistry showed that the epithelioid cells were strongly positive for S-100, with some spindle cells also staining positively, whereas the multinucleated giant cells were non-reactive. The tumour cells were negative for markers of lymphoma, gastrointestinal stromal tumour, leiomyosarcoma, germ cell tumour, and melanoma. The final diagnosis was a low-grade malignant peripheral nerve sheath tumour of the ileum (pT4N1). A histopathological examination of the right ovarian cyst confirmed that it was a benign cystic teratoma.

DISCUSSION

MPNSTs are exceedingly rare and rank as the sixth most common type of soft tissue sarcoma. Approximately 50% of the cases arise sporadically, whereas the remainder occur in individuals with neurofibromatosis type 1 (NF1).[1,2] In NF1-associated MPNST, tumours often develop through the malignant transformation of a plexiform neurofibroma or as a consequence of prior radiation exposure.[3,4] These tumours primarily affect adults aged 20 to 50 years, with a median age of 35. In this case, the patient was within the typical age range for MPNST but lacked the clinical features of NF1, suggesting a sporadic occurrence.

These tumours are most commonly found along the major nerve trunks, including those in the trunk, extremities, head, neck, and paravertebral regions. However, recent case reports have also documented MPNST in atypical locations, such as the liver, thyroid gland, skin, eighth cranial nerve, greater omentum, and small and large bowels.[3,4] MPNSTs originating from the small intestine are exceedingly rare. Most patients present with nonspecific symptoms, such as fatigue, weight loss, vomiting, abdominal pain, intestinal obstruction, or gastrointestinal bleeding.[4,5] In contrast, our patient presented atypically with anaemia, without overt signs of gastrointestinal disease. The anaemia was most likely due to slow, chronic bleeding from the tumour, which may explain the absence of obvious symptoms such as melena or haematochezia. The presence of hypochromic microcytic anaemia on the peripheral blood film further supports a chronic source of blood loss.

Diagnostic Role of Imaging and Histology

CT and magnetic resonance imaging (MRI) play important roles in the initial diagnosis and characterisation of tumours.[2,3,5] Key radiological features suggestive of MPNSTs include larger tumour size (>5 cm), infiltrative ill-defined margins, peritumoural oedema, intratumoural lobulation, bone destruction, and peripheral enhancement with a non-cystic or heterogeneous appearance on MRI. Morphologically, MPNSTs exhibit heterogeneity and often present as highly cellular spindle cell tumours arranged in fascicles. Immunohistochemical staining for S-100 protein is a key diagnostic marker for nerve sheath differentiation. The expression of additional markers, such as CD34 and Ki-67, can further assist in diagnosis. The presence of S-100 protein, along with elevated levels of p53 and Ki-67, provides valuable information for confirming the final diagnosis.[2,3,4]

Prognosis and Metastatic Potential

MPNSTs are associated with a high risk of local recurrence, with rates ranging from 40% to 60% within the first postoperative year. The lungs are the most common site of distant metastasis, while other metastatic locations include the liver, brain, bones, and adrenal glands. Factors associated with an increased risk of recurrence include the tumour's anatomic site, size (≥10 cm), and the adequacy of surgical margins.[6,7] The prognosis for MPNSTs of the small bowel tends to be poorer compared to other soft tissue sarcomas, although our understanding of this rare condition remains limited.[8,9]

Treatment Approach for MPNSTs of the Small Intestine (pT4N1)

The management of MPNSTs requires a multidisciplinary team (MDT) approach that integrates the expertise of surgeons, radiologists, and oncologists. Given the rarity of small intestinal MPNSTs, we applied oncological principles similar to those used for soft tissue sarcomas and gastrointestinal malignancies.

The primary treatment for MPNST with clinical pT4N1 status is surgical resection with wide negative margins (R0 resection) and regional lymphadenectomy. Complete surgical excision remains the cornerstone of treatment and the most significant predictor of survival.8,9 However, due to the high recurrence rate of MPNSTs, adjuvant therapy is often necessary even after R0 resection.

Adjuvant radiotherapy (RT) is often recommended to reduce local recurrence, particularly in cases of positive surgical margins or high-grade tumours.[10] Standard postoperative RT doses range from 50-66 Gy. However, its role in intra-abdominal MPNST remains uncertain due to potential toxicity to surrounding organs.[2] Given the patient's young

age, the achievement of clear surgical margins, and the lack of robust evidence supporting RT in this context, RT was not recommended.

While MPNSTs generally show a modest response to chemotherapy, adjuvant treatment may delay recurrence and improve survival, particularly in advanced or metastatic cases.[10] Because of the N1 status, which indicates a higher risk of systemic dissemination, doxorubicin, and ifosfamide combination chemotherapy was recommended. However, the patient declined adjuvant therapy.

Given the aggressive nature of MPNST, close posttreatment surveillance is essential. Given the limited data and lack of established guidelines for small-intestinal MPNSTs, our MDT recommended clinical assessments every 3–4 months, with abdominal and pelvic CT imaging every 6 months for 2 years, followed by annual surveillance for up to 5 years. Ideally, patients should be screened for NF1, because MPNSTs frequently arise in this genetic context, influencing prognosis and therapeutic decisions.

CONCLUSION

Our understanding of MPNSTs originating in the small bowel remains limited. Here, we present a rare case of MPNST arising from the ileum with an atypical presentation of anaemia. Accurate diagnosis and effective treatment require a multidisciplinary approach. Currently, no definitive treatment guidelines exist for small bowel MPNSTs. We recommend wide surgical excision as the primary treatment followed by vigilant monitoring for recurrence and metastasis. The efficacy of adjuvant RT and chemotherapy remains uncertain and warrants further investigation through clinical trials.

ACKNOWLEDGEMENT

We extend our gratitude to Universiti Malaysia Sarawak for continuous support in research and publication. We would like to thank Premier integrated labs for providing the HPE slide images.

DECLARATION

The authors declare no conflict of interest.

REFERENCES

1. Khan N, Hashmi I, Atallah L, Shaaban H, Guron G, Fedida A. A rare case report of malignant peripheral nerve sheath tumor involving both the small bowel and large bowel. Ann Afr Med 2021; 20(3): 228-31.
2. Zhu LB, Li PF, Xiao WH, Zhang PB, Li JQ, Sun MF. A distal ileum malignant peripheral nerve sheath tumor causing intussusception in a patient in China: a case report. World J Surg Oncol 2017; 15(1): 29.
3. Abraham AP, Franklyn J, Chandramohan J, Gaikwad P, Muthusami JC. Malignant peripheral nerve sheath tumour of the small bowel presenting with intussusception and perforation: a double jeopardy? Indian J Surg Oncol 2017; 8(2): 206-9.
4. Mohtaram A, Mesmoudi S, M'rabti H, Rami A, Latib R, Bernoussi Z, et al. Malignant peripheral nerve sheath tumour of the small bowel: an unusual presentation with fatal outcome. Case Rep Oncol Med 2013; 2013: 423867.
5. Rawal G, Zaheer S, Ahluwalia C, Dhawan I. Malignant peripheral nerve sheath tumour of the transverse colon with peritoneal metastasis: a case report. J Med Case Rep 2019; 13(1): 15.
6. Lee YJ, Moon H, Park ST, Ha WS, Choi SG, Hong SC, et al. Malignant peripheral nerve sheath tumour arising from the colon in a newborn: report of a case and review of the literature. J Pediatr Surg 2006; 41(2): e19-22.
7. Telem DA, Pertsemlidis D. Malignant peripheral nerve sheath tumour: an unusual cause of intussusception. J Gastrointest Surg. 2008; 12(9): 1609-11.
8. Le Guellec S, Decouvelaere AV, Filleron T, Valo I, Charon-Barra C, Robin YM, et al. Malignant peripheral nerve sheath tumour is a challenging diagnosis: a systematic pathology review, immunohistochemistry, and molecular analysis in 160 patients from the French Sarcoma Group database. Am J Surg Pathol. 2016; 40(7): 896-908.
9. Yao C, Zhou H, Dong Y, Alhaskawi A, Ezzi SHA, Wang Z, et al. Malignant peripheral nerve sheath tumours: latest concepts in disease pathogenesis and clinical management. Cancers (Basel). 2023; 15(4): 1077.
10. Wang BC, Kuang BH, Xiao BY, Lin GH. Doxorubicin/Adriamycin Monotherapy or Plus Ifosfamide in First-Line Treatment for Advanced Soft Tissue Sarcoma: A Pooled Analysis of Randomized Trials. Front Oncol 2021; 11: 762288.