University Journal of Surgery and Surgical Specialities

ISSN 2455-2860 2020, Vol. 6(8)

Malignant peripheral nerve sheath tumour of the small intestine a case report

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Abstract : Malignant peripheral nerve sheath tumours of the intestine are a rare clinical entity. These are soft tissue tumours that are usually found uncommonly in the extremities or the head and neck region. There are only a few cases reported worldwide of patients presenting with malignant peripheral nerve sheath tumours (MPNST) of the intestine. We report the case of a 21-year old male who was referred with abdominal mass and features of intermittent intestinal obstruction for two months. Pre-operative imaging was suggestive of large bowel intestinal obstruction due to ileo-colic intussusception. He underwent exploratory laparotomy and excision of the involved segment. The histopathological report revealed malignant peripheral nerve sheath tumour arising from the intestine.

Keyword : Malignant peripheral nerve sheath tumours, intussusception Malignant peripheral nerve sheath tumour of the intestine - a case report

Introduction:

Malignant peripheral nerve sheath tumours (MPNSTs) arise from a peripheral nerve or are tumours that exhibit nerve sheath differentiation. Epidemiologically, MPNSTs affect approximately 1/100,000 which corresponds to around 5-10% of soft tissue sarcomas(1). The extremities, head and neck, and trunk are the common sites where this tumour is found (1). The MPNSTs that arise from the intestine are very rare. Literature regarding the same is also only confined to case reports(2). Most cases present non-specifically with no clinical or radiological signs that may suggest the disease prior to histopathological evaluation. The optimum treatment modality remains unclear and the role of adjuvant therapy is anecdotal at best. We report a case of malignant peripheral nerve sheath tumour of the gut which presented as ileocolic intussusception.

Case Report

A 21-year-old male patient with no co-morbid illnesses presented with abdominal pain for two months associated with occasional vomiting. He also complained of loss of appetite and significant weight loss. On examination, he was emaciated, abdominal examination revealed generalised abdominal distension. A 15x10 cm mass was palpable in the lower abdomen. Computed tomography of the abdomen revealed a large ileo-colic intussusception. (Figure 1)

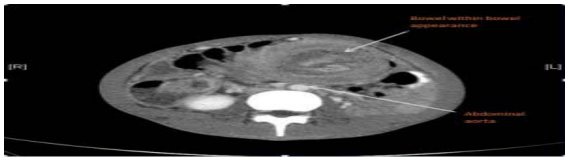

Figure 1

He was scheduled for exploratory laparotomy, which revealed ileo- colic intussusception up to the sigmoid colon. The lead point of the intussusception was a large, hard mass arising from the small bowel which had a whorled appearance. There was a linear perforation of the sigmoid colon with localised purulent material present in the pelvis. Subtotal colectomy, end ileostomy and colostomy (mucus fistula) were performed. (Figure 2) Histopathology report of the surgical specimen showed wall of ileum with the submucosa infiltrated by a tumour composed of short and long fascicles of spindle shaped cells with moderately pleomorphic, vesicular nuclei with coarse chromatin, inconspicuous nucleoli and moderate amounts of eosinophilic cytoplasm. Mitotic activity was ~20/10 hpf. There was surrounding desmoplasia with extensive areas of necrosis, acute and chronic inflammation. On immunohistochemistry the tumour cells were S100 positive. CD56, CD117, DOG-1, SMA and H-caldesmone were negative. The patient was discharged after recovering from the operation. He was discussed in the multidisciplinary tumour board meeting with an advice of adjuvant therapy.

Figure 2

Discussion

The term malignant peripheral nerve sheath tumour (MPNST) was used by the World Health Organisation (WHO) to replace the previously known ambiguous terms such as neurofibrosarcoma and neurilemomma(3). As this is a rare entity the pathogenesis is incompletely studied. They may arise *de novo* or as a component of a syndrome like neurofibromatosis type 1(4) . The clinical symptoms of the MPNSTs of the gastrointestinal tract are nonspecific; they include abdominal pain, vomiting, features of intestinal obstruction or GI bleed(5). Histological diagnosis and confirmation of peripheral malignant nerve sheath tumours is aided by immuno-histochemistry (6). The presence of atypical nuclei, giant cells and palisading arrangement are usually characteristic. The S-100 protein is usually present in the neural-derived neoplasms and can help diagnose MPNSTs. However, S-100 protein has a sensitivity of only around 50-60 % with a low specificity (7) . The presence of P53 and Ki67 can also help in diagnosing malignant peripheral nerve sheath tumour [11]. The presence of desmin and -SMA and CD34 and CD117 (c-kit) respectively can help in differentiating soft tissue sarcoma and gastrointestinal stromal tumour (GIST) from MPNSTs [10].

The management and prognosis of MPNSTs of the intestine have been extrapolated from MPNSTs of other locations. The treatment of choice is radical excision with wide margins. Routine nodal clearance is not indicated(8). The role of adjuvant therapy such as chemotherapy (doxorubicinifosfamide) or radiotherapy is confined to the recurrent disease and when there has been a positive margin after excision(9). The prognosis for these tumours is hard to assess objectively. The MPNSTs have the highest recurrence rates among sarcomas(10). The MPNSTs in other locations in the body have been studied and the prognosis of these tumours from other locations in the body is usually extrapolated to the MPNSTs of the gut(11). The factors which predict survival are the tumour size, completeness of surgical excision, metastasis and recurrent/primary tumour. Neurofibromatosis type 1 has also been associated with poor prognosis in a few reports(12)(13). The long term survival of these patients are poor(14). The 10-year survival is in the region of 35% for primary tumours, that reduces to 25% for ecurrent tumours and less than 20% for metastatic tumours (15). The present case was diagnosed to have ileo-colic intussusception with a soft tissue tumour forming the lead point. The diagnosis of MPNST was clinched on histopathological examination of the surgical specimen along with immune-histo chemistry. The present case is reported to highlight the importance of high index of suspicion in patients with unusual presentations of abdominal pain, the role of imaging in diagnosing intussusception and finally, immuno-histo chemistry in diagnosing MPNST of the gut.

4. Conclusion

Malignant peripheral nerve sheath tumours of the intestine is a rare finding, The disease is an enigma as there are very few case reports in the literature available. The diagnosis of this condition is arrived at after histopathological and imuuno-histochemical analysis. Surgical excision is essential, the role of adjuvant therapy such as chemotherapy is indicated in select cases, however there is no robust data to standardise treatment.

1. Hruban RH, Shiu MH, Senie RT, Woodruff JM. Malignant peripheral nerve sheath tumors of the buttock and lower extremity. A study of 43 cases. Cancer. 1990;66(6):1253–65.
2. Telem DA, Pertsemlidis D. Malignant peripheral nerve sheath tumor: an unusual cause of intussusception. J Gastrointest Surg Off J Soc Surg Aliment Tract. 2008 Sep;12(9):1609–11.
3. Darko Tomica DD. Aggressive intestinal schwannoma malignum mimicking gynaecological pathology - A case report. Coll Antropol. 2011;35 Suppl 1:339–43.
4. Doorn PF, Molenaar WM, Buter J, Hoekstra HJ. Malignant peripheral nerve sheath tumors in patients with and without neurofibromatosis. Eur J Surg Oncol. 1995 Feb 1;21(1):78–82.
5. Young P, Sung, Woo. Malignant Peripheral Nerve Sheath Tumor in Descending Colon - A Case Report. Korean J Pathol 2002 36 179-83. 2002;
6. Klijanienko J, Caillaud J-M, Lagacé R, Vielh P. Cytohistologic correlations of 24 malignant peripheral nerve sheath tumor (MPNST) in 17 patients: The Institut Curie experience. Diagn Cytopathol. 2002;27(2):103–8.
7. Gonzalez-Martinez T, Perez-Piñera P, Díaz-Esnal B, Vega JA. S-100 proteins in the human peripheral nervous system. Microsc Res Tech. 2003 Apr 15;60(6):633–8.
8. Kar M, Deo SS, Shukla NK, Malik A, DattaGupta S, Mohanti BK, et al. Malignant peripheral nerve sheath tumors (MPNST) – Clinicopathological study and treatment outcome of twenty-four cases. World J Surg Oncol. 2006 Aug 22;4(1):55.
9. Kroep JR, Ouali M, Gelderblom H, Le Cesne A, Dekker TJA, Van Glabbeke M, et al. Firstline chemotherapy for malignant peripheral nerve sheath tumor (MPNST) versus other histological soft tissue sarcoma subtypes and as a prognostic factor for MPNST: an EORTC soft tissue and bone sarcoma group study. Ann Oncol Off J Eur Soc Med Oncol ESMO. 2011 Jan;22(1):207–14.
10. Collin C, Godbold J, Hajdu S, Brennan M. Localized extremity soft tissue sarcoma: an analysis of factors affecting survival. J Clin Oncol. 1987 Apr 1;5(4):601–12.
11. Ducatman BS, Scheithauer BW, Piepgras DG, Reiman HM, Ilstrup DM. Malignant peripheral nerve sheath tumors. A clinicopathologic study of 120 cases. Cancer. 1986;57 (10):2006–21.
12. Wong WW, Hirose T, Scheithauer BW, Schild SE, Gunderson LL. Malignant peripheral nerve sheath tumor: analysis of treatment outcome. Int J Radiat Oncol. 1998 Sep 1;42(2):351–60.
13. Ramanathan RC, Meirion Thomas J. Malignant peripheral nerve sheath tumours associated with von Recklinghausen's neurofibromatosis. Eur J Surg Oncol EJSO. 1999 Apr;25(2):190–3.
14. Anghileri M, Miceli R, Fiore M, Mariani L, Ferrari A, Mussi C, et al. Malignant peripheral nerve sheath tumors: prognostic factors and survival in a series of patients treated at a single institution. Cancer. 2006 Sep 1;107 (5):1065–74.
15. Zou C, Smith KD, Liu J, Lahat G, Myers S, Wang W-L, et al. Clinical, pathological, and molecular variables predictive of malignant peripheral nerve sheath tumor outcome. Ann Surg. 2009 Jun;249(6):1014–22.

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