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A case report of perforated small intestine exophytic growth, a rare manifestation of Gastrointestinal Stromal Tumor (GIST)

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Abstract

Gastrointestinal stromal tumors (GISTs) are rare, but most common soft tissue neoplasm of the gastrointestinal tract, which represent 0.1% -3% of the gastrointestinal malignant tumors. GIST can affect any part of the gastrointestinal tract, but most common sites are stomach, jejunum/ileum and rarely occur at duodenum and colorectum. Usually, GISTs are asymptomatic but on the basis of site of origin, it can cause abdominal pain, bleeding, and mechanical obstruction. Herein, we report the case of GIST in 55 year old male patient, who was admitted with a history of RIF pain for 4 days and fever for 15 days. The clinical examination revealed the patient having no history of vomiting, anorexia, abnormal urinary or bowel habits, no bleeding per rectum and no weight loss. The histopathological finding suggested malignant GIST with infiltration into omentum. IHC study for CD117/c-kit was positive, thus confirming the case of GIST. Preoperative difficulty in diagnosis and management is discussed in this case report.

Keywords: GIST, C-KIT, CD117/ CD34

Introduction

Gastrointestinal stromal tumors (GISTs) although rare but most common primary mesenchymal tumor of the gastrointestinal tract, represents 0.1-3% of all the gastrointestinal malignancies [1]. The frequency of these tumors is estimated to 10 - 20/1,000,000 population [2, 3]. GIST can affect any part of the gastrointestinal tract, but most common sites are stomach (40-60%), jejunum/ileum (30%) and rarely occur at duodenum and colorectum (5%) [4]. GIST can even affect the extra-gastrointestinal sites such as omentum, mesentery or peritoneum [5]. GIST commonly occurs in adults between ages 40-70 but rarely children and young adults develop these tumors [6, 7]. Most GISTs may cause no

signs or symptoms. However, some cases patient may experience abdominal pain or swelling, nausea, vomiting, weight loss. Sometimes, bleeding may lead to low RBC counts, weakness and tiredness.

Several genes mutation involved in the formation of GISTs. But 80% of cases mutation in the C-KIT gene (CD117, CD34) and 10% in PDGFRA are associated with both familial and sporadic GISTs. Mutation in these genes results in constant activation of protein and signaling pathways, which increases the proliferation and survival of cells leads to GIST formation [8]. Surgical resection remains the standard treatment for all localized GISTs and in patients with metastatic neoplasm, imatinib mesylate is the standard chemotherapy. Histopathology and CD117/ CD34 detection by immunohistochemistry are the basis of the diagnosis of GISTs.

Case report

A 55- year old male patient presented with the case of fever for 15 days and pain RIF for 4 days. There was no history of vomiting, anorexia, abdominal urinary or bowel habits, bleeding per rectum and loss of weight. On clinical examination, pulse was 100/min, B.P. was 110/70 mm of Hg and spo2 97% at room air. There was an intraperitoneal lump of size 8x8 cm oval firm consistency occupying RIF extending up to right lumbar and umbilical region. Tenderness and guarding was observed in RIF.

Haematological investigations revealed hemoglobin 12.1 gm/dl, TLC 18000, neutrophilia, BUN 8, creat. 0.54, TSB 0.97, Direct Bilirubin 0.32 and serum albumin 3.4. Urine examination was within normal limit.

Ultrasonography revealed large heterogenous intra-abdominal cystic mass with internal fine moving echoes. Contrast-enhanced computed tomography (CECT) of the whole abdomen revealed irregularly marginated thick-walled SOL with peripheral thick enhancing wall 15mm measuring 18x15x13 cm occupying RHC and RIF with high-density contents and air fluid level in it. Fat stranding with surrounding mesentery observed. Appendix separately not seen. CECT also revealed localized collection near distal ileum might be the possibility of distal ileal perforation with localized collection contained rupture.

Exploratory laparotomy of the patient was planned. Intraoperative findings revealed a lump of size 10x20 cm in RIF crossing middle with omental wrapped around it. Growth was adherent to the mesentery of the small bowel and peritoneal fold in the pelvis and parietal peritoneum and anterior abdominal wall. Adhesiolysis was performed to make growth free. There was a loop of proximal ileum about 3 feet from ICJ entering into growth. About 1L foul smelling faeculent fluid aspirated. Growth separated from the ileal loop. There were two perforations over antimesenteric border over the ileal loop. Ileal resection and anastomosis were done. Rest of the gut was normal.

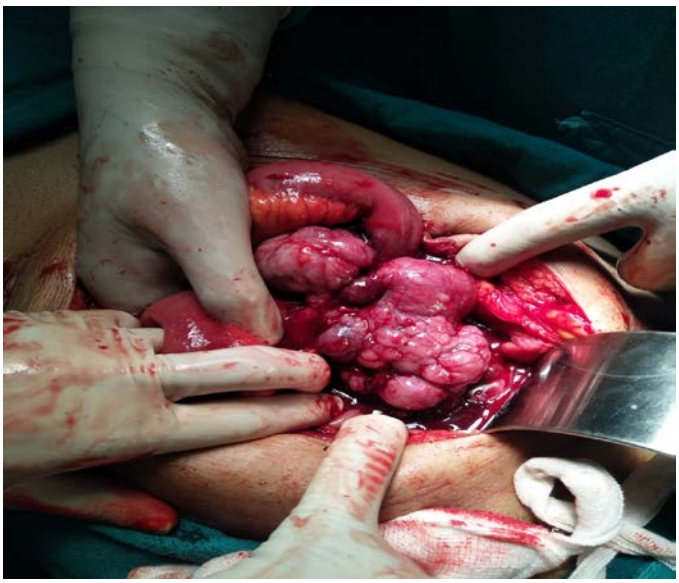

Figure 1

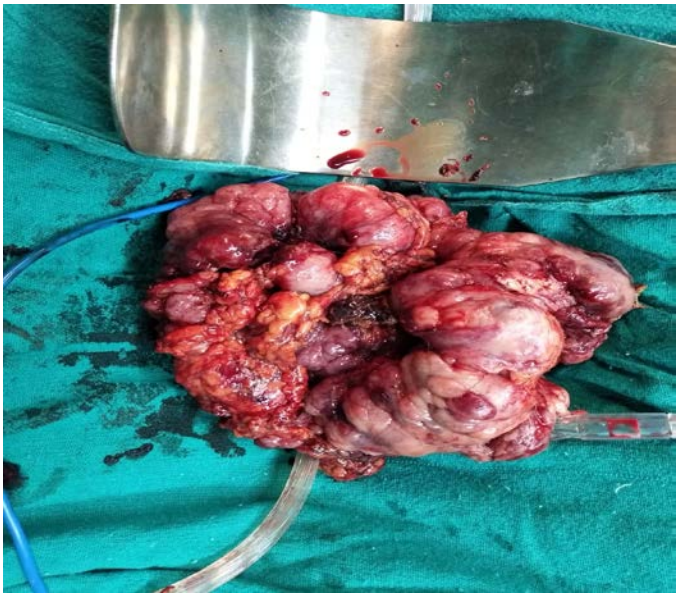

Figure 2

Growth resected, sent for histopathological examination and immunohistochemistry. On macroscopic examination grape-like, a cystic mass of size 20x10 cm was present. Histopathological examination revealed malignant gastrointestinal stromal tumor with infiltration into omentum. Immunohistochemistry revealed tumor cells were positive for CD117/C-KIT and negative for CD34. Postop period was uneventful and the patient discharged on 8th OPD.

Discussion

Among the acute clinical presentations of primary small bowel GIST, tumour mass perforation with peritonitis has been infrequently reported in the literature. GIST mostly affect individuals aged > 50 yrs with male to female ratio of 1.5 [9]. Most common sites Stomach (60%), Jejenum and Ileum (30%), Duodenum (5%) and colorectum (<5%) [10]. Small bowel GIST may present as minute incidental nodule to large tumours. Clinical signs and symptoms are usually nonspecific. Acute abdomen arises from bleeding, intestinal obstruction, tumour rupture with intra-abdominal haemorrhage, pelvic mass and acute appendicitis like acute pain [11]. Investigation of choice is CECT Abdomen. Small tumour <2cm if symptomatic, complete surgical resection is recommended. If asymptomatic can be managed conservatively with endoscopic surveillance at 6 to 12 months intervals. Surgery is the primary treatment of choice in localized or potentially resectable GIST. If tumour is unresectable preoperative Imatinib should be considered. Imatinib is effective in reducing the size of the tumour prior to resection. Increasing the likelihood of negative margin without significant morbidity. Adjuvant imatinib has been shown to improve OS in postsurgical patients [12]. Complete surgical resection is connected with 48-65% five-year survival [13]. Patients with localized or locally advanced tumors have 46% five-year survival. The prognosis is dismal in tumour with perforation, multifocal location or metastatic lesions. Patients with metastatic tumors or multifocal tumors in whom the five-year survival is 0%. Perforation of the tumor lowers the five-year survival to 24%, due to peritoneal dissemination [14].

Conclusion

Understanding that GIST constitute a distinctive GI tract lesion is critical in managing these rare and frequently aggressive tumors, which, given the risk variables currently recognised, have a strong tendency for recurrence and behave as a malignant lesion. A comprehensive strategy, postoperative targeted molecular treatment in intermediate and high-risk patients, and ongoing surveillance are all required for a successful outcome.

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