

Case Report

Jejunal gastrointestinal stromal tumors masquerading as an appendicular mass: an unusual presentation

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are a rare variety of tumors of mesenchymal origin found in the gastrointestinal (GI) tract forming about 1% of all GI tumors. These originate from the interstitial cells of Cajal. Small bowel GISTs have been shown to present as obscure GI bleeding, obstruction and perforation in literature. We report a 57 years old female patient presenting with pain abdomen, fever and vomiting and palpable right iliac fossa (RIF) mass diagnosed as an appendicular mass and managed conservatively. She was planned interval appendicectomy and was discovered to have a jejunal GIST at laparotomy treated with resection and anastomosis. There are case reports of small bowel GISTs presenting as sources of obscure or overt GI bleeding and luminal or extra luminal mass causing small bowel obstruction. Surgery is mainstay of treatment with imatinib for adjuvant or neoadjuvant therapy. This case highlights an unusual presentation of a jejunal GIST with a sealed off perforation mimicking an appendicular mass in the RIF treated by surgical resection followed by adjuvant Imatinib therapy. GIST being an uncommon tumor with varied presentations can lead to misdiagnosis and delays in treatment. This differential should be kept in mind while evaluating small bowel pathologies to aid a timely diagnosis.

Keywords: Jejunal GIST, Appendicular abscess, RIF

INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are rare tumors forming about 1% of all gastrointestinal (GI) tumors and find their origin in the interstitial cells of Cajal.¹ These can present as benign or malignant and can be symptomatic in up to 80% of the patients. The commonest site of presentation is gastric 55%, small bowel 32%, colonic 6%, extra-intestinal (E-GISTs) 6% and least in the esophagus 1%. Management of GISTs has evolved significantly in the past decade with newer modalities of treatment and improved outcomes.

The problem with GISTs lies not in treatment but diagnosis itself due to the myriad variety of presentation and subtle symptoms. Small bowel GISTs have been

shown to present as obscure GI bleeding, obstruction and perforation in literature.^{4,6} Here we present an unusual case of a jejunal GIST that initially presented as a right iliac fossa mass and managed as an appendicular abscess, only to be discovered at interval appendicectomy to be arising from the jejunum.

CASE REPORT

A 57 years old woman presented with lower abdominal pain of 2 weeks duration associated with low grade fever and vomiting. There was no history of constipation or abdominal distension. She did not have any known comorbidities or previous surgeries. Her general physical examination was unremarkable except for fever and tachycardia. On examination of the abdomen, we discovered a mass in the right iliac fossa (RIF) measuring

12 cm x 10 cm with tenderness. Per rectal and per vaginal examination was unremarkable.

Haematological investigations revealed leucocytosis (13,200 cells per cu.mm) with neutrophilia. Ultrasonography of the abdomen revealed a 9x7 cm lesion with ill-defined borders in the RIF with air foci within. The origin of the mass could not be delineated. An abdominal contrast enhanced computerised tomographic (CT) scan showed a complex mass measuring 8x6 cm in the RIF containing fluid and air and a tubular structure within and mild ascites (Figure 1). A diagnosis of ruptured appendix with appendicular abscess was made and conservative management was planned. She was treated as per the Ochsner Sherren regimen and her condition improved with resolution of tachycardia, fever, pain and decrease in size of the mass. She was discharged after completion of 14 days of antibiotic therapy for interval appendicectomy.

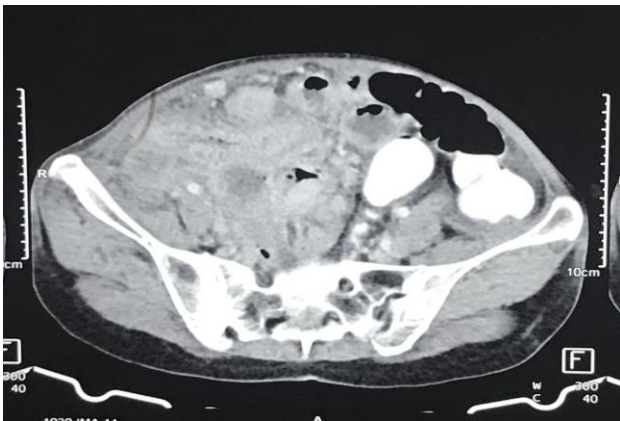


Figure 1: Computed tomography axial section through the lower abdomen showing a mass in the right iliac fossa with containing fluid and air within.

At review after 4 weeks she was asymptomatic with a clinically palpable mass in the RIF. She underwent ultrasonography for confirmation of the findings and was planned for appendicectomy. Intra-operatively she was found to have a mass in the right iliac fossa with multiple small bowel loops clumped together. Adhesiolysis was done and there was a mass was found to be arising from the jejunum with surrounding inflammation and adhesions with a sealed off perforation of adjacent loop of jejunum (Figure 2). She underwent resection and anastomosis of the jejunal segment with adequate margins and the specimen was sent for histopathology.

Her post-operative recovery was uneventful. Histopathology of the surgical specimen showed a partially encapsulated tumour with spindle cells and immunohistochemical profiling with S-100, CD117 and DOG-1 confirmed the diagnosis of gastro-intestinal stromal tumor. There were <5 mitoses/50 high power fields and the proliferative index (Ki-67) was <5%. The tumor was graded as low risk. As per the

multidisciplinary tumor board opinion she was advised adjuvant Imatinib therapy and was started on the same after 3 weeks of surgery. She was found to be symptom free and tolerating the treatment well at 6 weeks follow up.

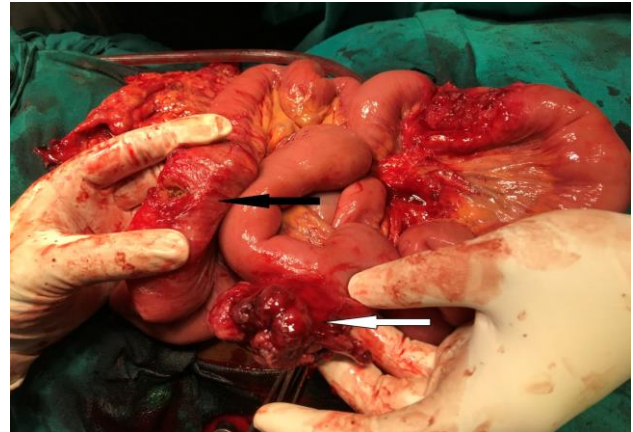


Figure 2: Mass arising from the jejunum (white arrow) with surrounding inflammation and adhesions with a sealed off perforation of adjacent loop of jejunum (black arrow).

DISCUSSION

GISTs are rare albeit the most frequent tumor of mesenchymal origin found in the GIT.¹ The various locations are gastric 55%, small bowel 33% and rest of GIT. About 10% of these tumors present in the extra-intestinal locations such as mesentery, pelvis, retroperitoneum and omentum.^{1,2} These tumors start in the muscularis externa of the GIT and tend to grow extraluminally more often than intra-luminal. 80% of these tumors present with symptoms such as bleeding, pain, mass per abdomen or intestinal obstruction while the rest 20% are asymptomatic and incidentally detected.²

Radiologically GISTs can have a varied presentation depending on size and location but usually present as a well-defined mass with a predominantly exophytic component. They may demonstrate a homogenous or heterogenous enhancement and necrotic cavities or foci of cystic degeneration within. Differentials include tumors of other origins such as adenocarcinoma, lymphoma, secondaries and other mesenchymal neoplasms.⁷

On gross pathological examination, GISTs appear to be well circumscribed round to ovoid masses with fleshy appearance on cut section and areas of cystic degeneration or haemorrhage. Both benign and malignant types can have similar appearance and hence, distinction is not possible.⁸ Histopathologically, there are three subtypes of spindle 70%, epithelioid 20%, or mixed 10%.² This can resemble any mesenchymal tumors and immunohistochemistry is required for confirmation of diagnosis with CD117 being the most common marker

followed by other markers like CD34, DOG-1 and S-100.^{2,8}

Location in areas other than stomach, larger dimensions and greater mitotic index have been shown to be the factors associated with higher recurrence. Risk stratification of GISTs is based on mitotic index and size and is used to guide adjuvant therapy.^{1,3} Feng et al showed that, within small bowel, jejunal GISTs tend to be more common, larger in size and carry more chance of being high risk than ileal but prognosis for both is similar.⁹

Management of GISTs has evolved significantly in past decades. Surgical excision which was previously thought to be the only management option has now been challenged with discovery of immunohistochemical markers like CD117 and KIT and PDGFRA mutations. Targeted therapy with Imatinib has become feasible and mainstay in advanced and metastatic tumors.³ Resection followed by adjuvant Imatinib is considered gold standard of treatment for intermediate to high risk tumors is a well-tolerated treatment and increases recurrence free survival.^{3,10,11} Unresectable and borderline resectable tumors are treated with neoadjuvant Imatinib followed by surgery.^{11,12} Treatment of metastatic GISTs is still a matter of debate and there are no strict guidelines for surgical. Currently, these are treated with Imatinib and surgical resection is carried out if the tumor shrinks and becomes amenable to resection and metastasectomy.¹¹

Jejunal GISTs can present with variety of symptoms and occasionally, GI bleeding with anemia and fatigue can be the presenting complaint. Bleeding from the small bowel is usually missed during routine evaluation by an esophagogastroduodenoscopy and colonoscopy and this causes significant delays in the diagnosis and treatment. A useful modality to evaluate for bleeding from the small intestine and to look for lesions is a double balloon enteroscopy and capsule endoscopy.¹³ GISTs may not present as an intraluminal mass but will be seen as telangiectasias causing continuous occult or overt bleed.^{4,13} In a case reported by Mujawar et al, a similar presentation of a patient presenting with bleeding per rectum, leading to severe anemia and shock, was treated with exploratory laparotomy to reveal a jejunal GIST as the source of bleeding.¹⁴

Occult GI bleeding can sometimes be the predominant symptom and the mass can grow to a conspicuous size up to several centimetres before being recognised by the patient.¹⁵ These tumors can also present as a luminal mass causing small bowel obstruction.⁵ Pandit et al reported a mesenteric GIST from jejunal mesentery with impending perforation misdiagnosed as a right colonic growth with sealed off perforation due to its location on CT.⁶

In our case, there was an unusual presentation of a jejunal GIST that caused a sealed off perforation and was

initially misdiagnosed as an appendicular mass leading to delay in definitive treatment. As the tumor was low risk and patient was in good general condition, her final outcome was favourable.

CONCLUSION

This case highlights an instance of a jejunal GIST causing a sealed off perforation of an adjacent loop which was misdiagnosed as an appendicular mass leading to a delayed definitive treatment. GISTs arising from the small bowel can present with varying degree of symptoms which may not co-relate with size of the tumor and this often leads to delay in diagnosis. A cautious thought should be given to the varied atypical presentations, when evaluating small bowel pathologies to clinch this diagnosis at an early stage. Surgery is the cornerstone of management for localised and resectable tumors aided with adjuvant treatment with Imatinib for successful outcomes.

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