Case Report

# A rare presentation of jejunal GIST: A case report

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## ABSTRACT

Gastrointestinal stromal tumors (GISTs) are rare tumors that constitute 1% of all GI tract tumors. Jejunal GISTs are the rarest subtype. We present a middle-aged gentleman, who presented with pain in the right lower abdomen. On abdominal examination, a lump was palpable in the right iliac fossa (RIF). Contrast-enhanced computed tomography of the abdomen revealed an extraluminal soft-tissue mass in the ileum. Computed Tomography-guided core needle biopsy from the lump was consistent with GIST, which was confirmed on immunohistochemistry. Mutation analysis revealed exon 11 mutations. Due to the proximity of GIST to the rectum and urinary bladder, the patient was started on imatinib therapy. After 3 months of treatment, imatinib therapy had to be stopped due to skin reactions. Restaging was done with a positron-emission tomography scan, which demonstrated a soft-tissue mass likely arising from the ileocecal region in the RIF abutting the ascending colon without any significant decrease in size. On exploration, a well-circumscribed mobile extraluminal lobulated mass was seen arising from the antimesenteric border of the jejunum. Histopathologic examination showed GIST, which was confirmed on immunohistochemistry. We report this case to emphasize keeping small bowel GIST as an unusual differential diagnosis of a RIF lump. Furthermore, not all patients can tolerate imatinib treatment, hence exon mutation study is important, and surgery should be considered if it is deemed resectable.

**Key words:** Gastrointestinal stromal tumors, Jejunal tumors, Right iliac fossa mass

Gastrointestinal stromal tumors (GISTs) are the most common type of mesenchymal neoplasm that occurs in the gastrointestinal (GI) tract. These tumors originate from pluripotential mesenchymal cells that are committed to becoming interstitial cells of Cajal, which are the pacemaker cells situated between the circular and longitudinal layers of the muscularis propria along the GI tract. Two-thirds of GISTs occur in the stomach, whereas one-fourth develops in the small intestine. Jejunal GISTs are extremely rare, representing only 0.1–3% of all GIST tumors [1]. GISTs are usually asymptomatic, and they are often diagnosed with non-specific symptoms. The symptomatology of GISTs depends on the location of the tumor. Abdominal pain and bleeding into the GI tract are more common in gastric GISTs (61% and 58%, respectively), while acute abdominal symptoms are more common in jejunal and ileal GISTs (40% and 60%, respectively) [2]. Although there are a few reported cases of jejunal GISTs presenting as an abdominal mass, their initial presentation as a right iliac fossa (RIF) lump is extremely uncommon. Neoadjuvant tyrosine kinase inhibitor (TKI) therapy is often considered for locally advanced GISTs to reduce the tumor size, which may facilitate margin-negative resection [3]. Mutation study is essential and helps to guide the appropriate therapy. Although the drug is generally well-tolerated, up to 3.7% of patients have to discontinue treatment due to adverse events [4].

Here, we report an abnormal presentation of jejunal GIST masquerading as a RIF mass in a 67-year-old gentleman, where imatinib therapy had to be stopped due to skin rash. This case report emphasizes keeping small bowel GIST as an unusual differential diagnosis of RIF mass. Furthermore, imatinib treatment may not be well-tolerated by all patients, leading to its discontinuation. If the tumor is deemed resectable, surgery should be considered and a mutation study should be done before starting imatinib therapy.

## CASE REPORT

A 67-year-old gentleman, non-alcoholic and non-smoker, without any prior surgical history or family history of malignancy, presented to our center with pain in the right lower abdomen for 2 months, which was insidious in onset, continuous, non-radiating, and relieved by analgesics. He did not have nausea, vomiting, fever, hematochezia/melena, abdominal distension, weight loss,

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altered bowel habits, or trauma to the abdomen before the onset of the symptoms.

On general examination, vitals were stable and pallor was present. On abdominal examination, an approximately 5 cm × 5 cm ill-defined tender, firm, mobile lump with a smooth surface was palpable in the RIF region. The rectal examination was normal.

Laboratory tests were normal. Ultrasound of the abdomen reported a large 6.4 cm × 6.8 cm heterogeneous solid space-occupying lesion noted beneath the urinary bladder, abutting the rectum without ascites. The contrast-enhanced computed tomography abdomen revealed a heterogeneously enhanced complex lobulated exophytic extraluminal soft-tissue mass in relation to the ileum with few peripheral calcifications and no features of gut obstruction (Fig. 1). A computed tomography (CT)-guided core needle biopsy from the lump was consistent with GIST. Immunohistochemistry was positive for CD117 and DOG1, confirming the diagnosis of GIST. Due to the proximity of GIST to the rectum and urinary bladder, the patient was started on imatinib therapy at a dose of 400 mg once daily. Mutational analysis revealed a KIT gene (exon 11) mutation.

However, after 3 months of starting imatinib therapy, the patient developed severe itchy, desquamative skin lesions over the whole body except the face, due to which treatment had to be stopped. The patient underwent restaging with a positron-emission tomography scan, which demonstrated metabolically inactive circumscribed heterogeneously enhancing soft-tissue mass of 6.8 cm × 6.3 cm × 6.5 cm with peripheral nodular calcification likely arising from the ileocecal region in the RIF abutting the ascending colon.

After a multidisciplinary tumor board discussion, a decision was made to plan for laparoscopy with or without laparotomy. A mass was found in the RIF on laparoscopy. A small infra-umbilical midline incision was made. On exploration, around 9 cm × 7 cm × 6 cm well-circumscribed mobile extraluminal lobulated mass was seen arising from the antimesenteric border of the jejunum, approximately 15 cm from the duodenojejunal flexure, and lying in the RIF region (Fig. 2). He underwent wedge resection of GIST with primary anastomosis. The abdomen was closed in layers.

Histopathologic examination of the excised mass revealed a 9 cm × 7 cm × 6 cm mass arising from the antimesenteric border of the jejunum with features of a low-grade spindle cell type, with 20% viable tumor cells and >80% necrosis with free margins. Mitotic figures constitute 20/50 high-power fields. CD117 (c-KIT) and DOG1 immunostains were strongly positive in the tumor cells (Fig. 3), confirming the diagnosis of GIST, spindle cell type, and ypT3Nx. On risk stratification, GIST was categorized as a high-risk group.

The post-operative period was uneventful, and was discharged on the 12$^{th}$ post-operative day after the removal of stitches. After a multidisciplinary tumor board discussion, the patient was started on imatinib therapy with a decreased dose of 200 mg once daily, considering the large size of the tumor and the high-risk assessment. The patient remains disease-free at 6 months of follow-up with better tolerance to the treatment and is advised to have regular follow-up and continuation of imatinib therapy.

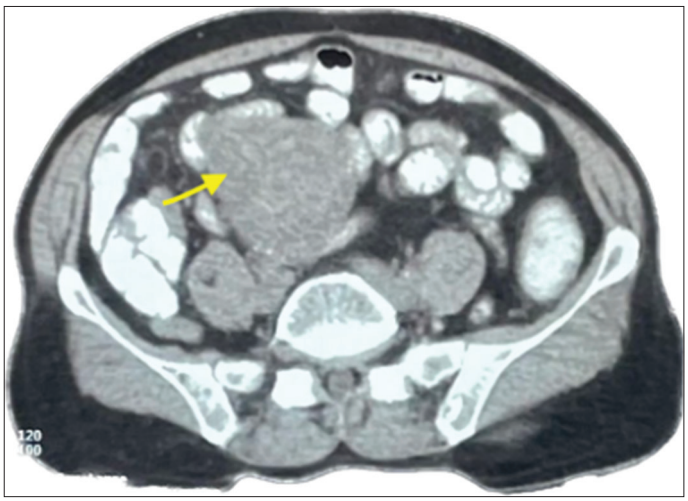

**Figure 1: Contrast-enhanced computed tomography abdomen (axial section) showing a complex lobulated mass in relation to the ileum (yellow arrow depicting the mass)**

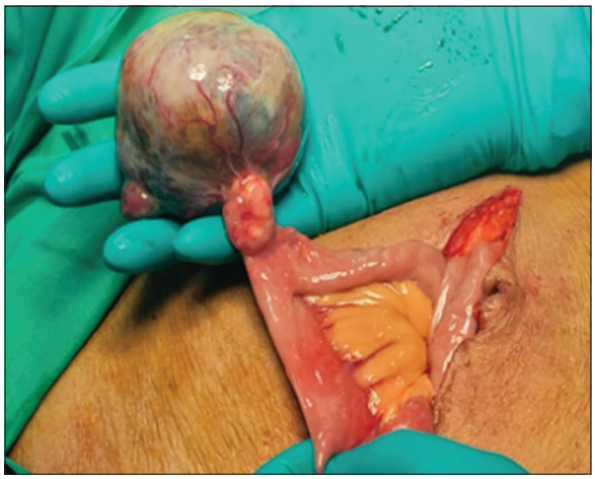

**Figure 2: Huge mass arising from the jejunum**

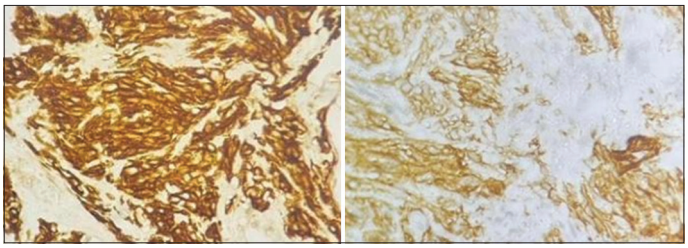

**Figure 3: Tumor cells showing strong cytoplasmic positivity for CD 117 (left) and DOG-1 (right)**

## DISCUSSION

The majority of GISTs are diagnosed in adults over 40 years of age, with a peak age of 60–65 years, and show a slight male preponderance. GISTs can develop anywhere along the GI tract, from the esophagus to the rectum; however, the stomach (60%) and small intestine (30%) are the most common locations for GIST. Only 10% of GISTs are found in the esophagus, mesentery,

omentum, colon, or rectum. Jejunal GISTs are the rarest subtype [1].

Jejunal GISTs are typically asymptomatic while small and may be diagnosed incidentally from CT, during surgery, or from symptomatic liver metastases. Enlargement causes variable symptomatology; GI bleeding or non-specific GI symptoms such as bloating or early satiety. Around 40% are associated with ulceration, and 28% present with overt GI bleeding. Bleeding may be acute (hematemesis or melena) or chronic (anemia). Around 20% grow large enough to present with pain, a palpable mass, or obstruction secondary to intussusception [2]. There are only a few documented cases of GIST presenting as a RIF lump. Jejunal GIST has been reported to masquerade as an appendicular mass [5], ovarian mass [6], intratumoral abscess [7], or a huge abdominal lump [8]. In our case, jejunal GIST presented as a RIF mass.

Because GISTs do not have a distinct appearance on ultrasound, a contrast-enhanced CT scan is the preferred imaging modality for the evaluation of patients with suspected GIST. There are no specific CT findings for GIST tumors, although they typically appear as an inhomogeneous mass with areas of necrosis and hemorrhage (usually in the center), while viable tumor areas show contrast enhancement (usually at the periphery) [9]. The definitive diagnosis of GIST is based on histology and immunohistochemistry. On histopathology, tumor cells demonstrate mainly three types of histological patterns, the most common of which is spindle cell type (60–70%), followed by epitheloid type (30–40%), and mixed type (10–20%) [10]. In our case, GIST was of spindle cell type. Positive immunohistochemical staining for CD117 is a defining characteristic of GISTs, and currently, it is considered the most specific and sensitive marker for GIST [11]. DOG-1 is a sensitive immunohistochemical marker for GIST, comparable with KIT, with the additional benefit of detecting 36% of KIT-negative GISTs. DOG-1 is also a sensitive marker for unusual GIST subgroups lacking KIT or PDGFRA mutations [12]. In our case, GIST was positive for CD117 and DOG-1 and even though we had a pre-operative diagnosis of GIST, the exact location and the operability of the GIST were confirmed only during exploration.

The treatment modality for non-metastatic GISTs is surgical resection with negative margins without lymph node resection. However, if the tumor is large and suspected to have infiltrated the nearby organs, neoadjuvant imatinib therapy results in tumor shrinkage, thus increasing the chances of margin-negative resection with decreased chances of tumor rupture, thereby decreasing the local recurrence rates [3]. Mutational analysis is essential to making an adapted decision about TKI therapy. Patients with exon 11 mutations are more likely to benefit from imatinib therapy than those with the less common KIT exon 9 mutations, which needs dose escalation to 800 mg/day. Patients who harbor a KIT mutation-negative tumor with a D842V mutation in the PDGFRA gene, succinate dehydrogenase-deficient GIST, or neurofibromatosis-related GIST should not be offered TKI therapy. Skin rashes may frequently be associated with the intake of imatinib (66.7%). However, grade 3 or 4 skin rashes are observed in only a small proportion of these cases (3.8%) [13], and in such cases, imatinib therapy should be discontinued urgently. Imatinib-associated rashes are often pruritic. The lesions are typically erythematous or maculopapular and appear mainly on the forearms or body, but can also occur on the face. In our case, the patient developed grade 3 skin lesions according to the criteria of the National Cancer Institute (Table 1) [14]. As a result, the medication was stopped.

**Table 1: Adverse skin reactions according to the National Cancer Institute criteria**

| Grade | Skin Lesions |
|---|---|
| 1 | Macular or papular eruption or erythema without associated symptoms |
| 2 | Pruritic macular or papular eruption or erythema with covering <50% of body surface area |
| 3 | Symptomatic, generalized erythroderma or macular, papular or vesicular eruption, or desquamation covering more than 50% of body surface area |
| 4 | Generalized exfoliative dermatitis or ulcerative dermatitis |

The Armed Forces Institute of Pathology classification and the Modified-National Institutes of Health classification system are the most commonly used for the prognostication of GIST. It divides patients into very low-risk, low-risk, medium-risk, and high-risk groups according to mitotic rate, tumor size, tumor site, and tumor rupture. Adjuvant therapy with imatinib given at a dose of 400 mg/day for 3 years is the standard treatment for patients with a high risk of relapse, based on a randomized trial with relapse-free survival and overall survival advantages [15].

## CONCLUSION

When diagnosing a mass in the RIF, it is important to consider the possibility of GIST arising from the cecum or small bowel. If the mass can be operable upfront, surgery should be considered. If the mass is a GIST that cannot be removed surgically with negative margins, neoadjuvant therapy with imatinib should be considered. Mutational testing should be performed to determine if imatinib therapy would be effective. Adverse effects from imatinib therapy may require discontinuation. GISTs in the jejunum are rare, and those presenting as a mass in the RIF have not been reported before.

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