Gastrointestinal stromal tumors (GISTs): A case report

Shuvekcha Thapa[1]*, Sarjan Kunwar[1], Bijay Pandey[1], Deependra Man Shrestha[2]

[1]Department of General Surgery, Gandaki Medical College Teaching Hospital & Research Center, Pokhara, Nepal, [2]Department of Oncology, Gandaki Medical College Teaching Hospital & Research Center, Pokhara, Nepal

ABSTRACT

Gastrointestinal stromal tumors (GISTs) are rare malignant tumors, rarely found in jejunum. Diagnosis mainly relies on histopathological examination and immunohistochemistry. Management typically includes surgery and chemotherapy. Herein, we present a case of an older adult female with abdominal pain and a palpable lump and managed by small bowel resection and adjuvant imatinib therapy. The excised tumor showed positive immunoreactivity for CD117, DOG-1 and SMA. This case report highlights the importance of considering GISTs in abdominal masses and the role of CT imaging and Immunohistochemistry in diagnosis and management.

Keywords: CD117, DOG-1, diagnostic imaging, gastrointestinal stromal tumor, imatinib, jejunal tumor, surgical management.

***Correspondence:**

Dr. Shuvekcha Thapa
Department of General Surgery
Gandaki Medical College Teaching Hospital and Research Center, Pokhara, Nepal
Email: thapashuvekcha@gmail.com

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INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are mesenchymal tumors originating primarily in the gastrointestinal tract, with the stomach being the most commonly affected site (56%), followed by the small intestine (32%), colon and rectum (6%), and esophagus (<1%).[1]

The annual incidence of 10–15 cases per 1,000,000 patients, a rate that has exhibited a rising trend in the last few years.[2] Although relatively rare, jejunal GISTs make up about 0.1-3% of all GIST tumors.[3] These tumors arise from the intestinal cells of Cajal, located in and around the nerve plexuses in GIT and express CD 117 (c-kit), a proto-oncogenic protein.[4] Additionally, mutation in the PDGFRA gene encoding for platelet-Derived Growth Factor Receptor Alpha protein, also playing a significant role in tumor growth and progression.[5]

Here, we report a case of a 70-year-old female presenting with right upper abdominal pain and a gradually enlarging palpable mass.

This case is unique due to the exceptionally large size of the jejunal GIST with a low mitotic index and absence of metastasis, demonstrating a rare discordance between tumor size and biological aggressiveness.

CASE PRESENTATION

A 70-year-old female presented to the outpatient department of Gandaki Medical College Teaching Hospital and Research Center with a one-year history of insidious-onset lower abdominal pain. Over the past four months, she developed a gradually enlarging right upper abdominal mass, black-colored stools, loss of appetite, and a weight

loss of 6 kg. She had no significant past medical or surgical history and no family history of gastrointestinal disorders.

On examination, she was pale but hemodynamically stable, with a firm, non-tender 10 × 7 cm^2 mass in the right hypochondrium. Laboratory findings were normal.

Contrast enhanced computed tomography of abdomen and pelvis showed a long segmental circumferential thickening measuring approximately 32.7 mm in maximum thickness and length of approximately 98 mm of small bowel (jejunum) with homogenous enhancement in posterior contrast study and surrounding fat stranding (Figure 1a). The thickened loops were abutting ascending colon laterally, pylorus of stomach superiorly, superior mesenteric vein medially. Other parts of bowel were normal (Figure 1b).

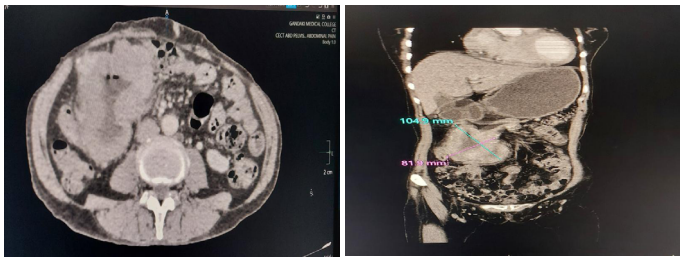

Figure 1a and 1b: Axial section (a) and coronal reformat (b) from CECT abdomen and pelvis showing long segmental circumferential thickening

Provisional diagnosis of large jejunal mass was made. The patient was admitted and the decision to go for an operative procedure was made. Resection of jejunal mass with jejunojejunal anastomosis done on 23-10-2024, the specimen was sent for histopathological examination. The post-operative period was uneventful.

Operative findings were- A highly vascular mass arising from the jejunum tumor around 7 cm distal to duodenojejunal flexure. Tumor size of approx. 10x8x10 cm^3, tumor length of approx. 7 cm of the jejunal wall. Tumor was adherent to the mesentery of colon. No lymphadenopathy was noted. Grossly other bowel loops were normal(Figure 3).

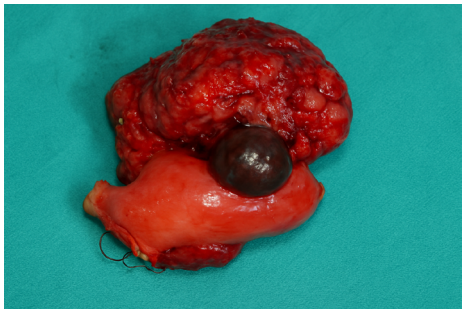

Figure 2: Gross specimen of Duodenojejunal (DJ) Junction - Highly vascular tumor mass arising from the jejunum

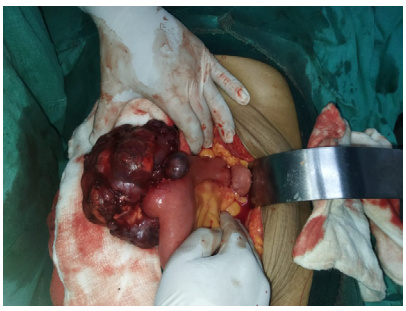

Figure 3: Intraoperative picture showing: Jejunal mass distal to duodenojejunal flexure adherent to the mesentery of colon.

Histopathology reports- Grossly, the duodenojejunal (DJ) Junction segment with mass protruding from the lumen altogether measuring 10x9x8 cm^3. The DJ segment measures 9x3x1.5 cm and the mass measures 10x9x7 cm^3 and was 1 cm^3 away from the capsular area. The mass was dark brown with irregular surface, appears lobulated and congested vessels visible on the surface (Figure 2). The cut section of the mass shows variegated appearance, necrosis and cystic spaces filled with hemorrhage.

Microscopically, the tumor cells are composed of spindle-shaped cells arranged in intersecting fascicles with a vague whorled pattern (Figure 4a and 4b). Mitotic activity was 3 per 50 high-power fields. Focal necrosis and hemorrhage were noted, but no atypia, lymphovascular, or perineural invasion was identified (Figure 4a). On this basis, a benign mesenchymal tumor of the intestine was considered, with differential diagnoses including gastrointestinal stromal tumor and leiomyoma. Tissue was sent for immunohistochemistry to establish the diagnosis.

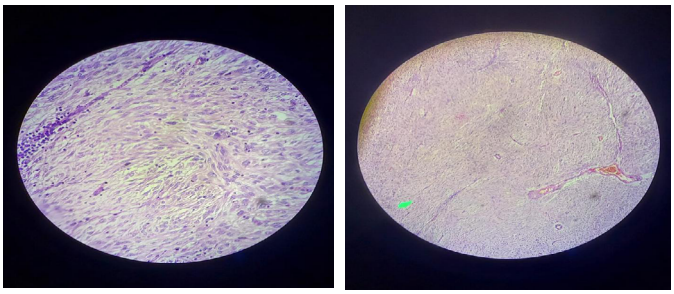

Figure 4a and 4b: Photomicrograph showing cells arranged in intersecting fascicles and vague whorled pattern. Individual cells are uniformly spindle-shaped. (original magnification 10X and 40X).

Immunohistochemistry report shows a cellular spindle cell neoplasm arranged in fascicles. Individual tumor cells are spindled with elongated blunt end vesicular nuclei and moderate eosinophilic cytoplasm. Mitosis is < 5/50hpf.

Necrosis is not seen, the tumor cells are positive for CD117, DOG-1, SMA and negative for H-caldesmon, Desmin, S100, CD34, CK. Ki-67 is 8-10 %.

Final diagnosis based on findings of histopathology and immunohistochemistry favours the diagnosis of a low-grade, spindle cell GIST.

The patient was managed with resection of the small bowel due to the size of the tumor. Based on the National Comprehensive Cancer Network (NCCN) Guidelines 2024, it was categorized as T4N0, stage IIIA, indicating a moderate risk of metastasis.

Microscopic evaluation revealed a Grade 1 tumor with three mitoses per 50 high-power fields (HPF), consistent with moderate risk. Accordingly, adjuvant imatinib therapy, a targeted tyrosine kinase inhibitor, was initiated at a dose of 400 mg once daily.

DISCUSSION

Although uncommon, jejunal GISTs make up about 0.1 to 3% of all GIST tumors.[2] Jejunal GISTs are often asymptomatic when small and usually discovered incidentally through imaging and larger tumors may cause nonspecific gastrointestinal symptoms, bleeding, or, in 20% of cases, a palpable mass or obstruction.[3] Over 90% of GISTs occur in adults over 40 years old with a median age at diagnosis is between 55 and 65 years.[6]

Moreover, GISTs lack specific CT features but typically appear as inhomogeneous masses with central necrosis or hemorrhage and peripheral contrast enhancement. Simultaneously, it also plays a crucial role in identifying and characterizing the tumor.[7]

In addition to imaging, the definitive diagnosis of gastrointestinal stromal tumors (GISTs) is established through a combination of histopathological assessment and immunohistochemical profiling. The morphology of jejunal GISTs is varied: tumors may be composed of spindle cells (70%), epithelioid cells (20%) or mixed spindle and epithelioid cells (10%).[8]

Furthermore, prognosis depends on tumor size, mitotic index, and site of origin, with mitotic count being a key predictor of malignancy.[9] A mitotic rate ≥ 5/50 HPF predicts higher recurrence and poorer overall survival.[10] Immunohistochemically, CD117 is expressed in approx. 95% of cases while DOG-1, SMA, CD34, S100, desmin and BRAF are variably expressed.[11] DOG-1 positivity rate is almost identical to CD117 positivity and currently considered the most specific and sensitive marker for GISTs.[12] As seen in this case, the tumor showed a low mitotic index, minimal atypia, limited necrosis, and Ki-67 of 8–10%, indicating a low-grade spindle cell tumor with favorable prognosis.

Regarding treatment, the primary 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.[13] Given the tumor's size and moderate-risk features, adjuvant imatinib was initiated, with ongoing follow-up necessary to monitor long-term safety and efficacy.

Finally, the limitation of this case was the unavailability of original immunohistochemistry (IHC) photomicrographs, as the analysis was performed at a referral laboratory outside the country, where image release was restricted. Nevertheless, the patient remains under regular follow-up, and longer observation is required to assess long-term outcomes.

CONCLUSIONS

Jejunal GISTs are rare tumors that often remain asymptomatic until advanced, leading to delayed diagnosis. In this case, a large jejunal GIST presenting with abdominal mass and pain was identified on CT imaging and confirmed by histopathology and immunohistochemistry, with CD117 and DOG-1 positivity being highly specific. Complete surgical resection followed by adjuvant imatinib was undertaken due to the tumor's size and moderate metastatic risk, with good early tolerance. This case underscores the need for early consideration of GISTs in patients with unexplained abdominal masses and highlights the importance of early detection, accurate pathological assessment, and appropriate integration of surgery with targeted therapy to optimize outcomes.

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PATIENT CONSENT: Informed consent for publication of this case had been obtained from the patient.

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