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Gastrointestinal Stromal Tumour (GIST) of Duodeno-Jejunal (DJ) Flexure with Gastrointestinal Bleeding: A Rare Case Report

Nagesh Madnoorkar[1], Pramod Shinde[2], Aarti Anvikar[3], Saylee Madnoorkar[4]

[1]Kaushalya Hospital, Surgical Oncology, Nashik, India.

[2]Kaushalya Hospital, General Surgery, Nashik, India.

[3]Government Medical College, Pathology, Miraj, India.

[4]Government Medical College, Pathology, Miraj, India.

Correspondence: Dr. Nagesh Madnoorkar.

ABSTRACT: GIST - Gastrointestinal stromal tumor is the most common mesenchymal tumor of the gastrointestinal tract with an annual incidence of 11-20% per 1 million people.

In GIT, the most common site of presentation is the stomach (70%) followed by the small intestine (20-30%), remaining (10%) in the colon, rectum, and oesophagus.

GIST occurs due to mutations in the receptor tyrosine kinase KIT gene. Few cases have mutations in the PDGFRA gene.

Here we report a rare case of Duodeno-Jejunal (DJ) flexure GIST with a history of pain in the abdomen and melena.

KEYWORDS: Gastrointestinal bleeding, Gastrointestinal stromal tumour (GIST), DJ flexure, CD 117, C KIT

INTRODUCTION

There is a unique subset of gastrointestinal mesenchymal neoplasms that are referred to as gastrointestinal stromal tumors (GISTs). GISTs are a prominent pathological entity within the realm of gastrointestinal malignancies, with an estimated annual incidence of 11% to 20% per 1 million people[1]. Abdominal pain, bleeding, blockage, or incidentally detected masses are just few of the many clinical manifestations of these malignancies [2-3]. The diagnosis and therapy of GISTs depend on a thorough understanding of the disease's distinct genetic basis.

Mutations in the KIT proto-oncogene, which codes for the receptor tyrosine kinase KIT (also known as CD117) [4-6], are the primary cause of GIST pathogenesis. Mutations that activate KIT lead to constitutive kinase activity and unchecked cell proliferation, both of which contribute to the development of tumors. Although KIT mutations account for the vast majority of genetic alterations in GISTs, mutations in the platelet-derived growth factor receptor alpha (PDGFRA) gene are present in a small percentage of cases (about 5-10%) [7]. These PDGFRA-mutated GISTs have different clinicopathological characteristics than their KIT-mutated counterparts and may have different responses to targeted therapy.

Management decisions for gastrointestinal stromal tumors (GISTs) are heavily influenced by the tumors' clinical appearance and anatomical location. Approximately 70% of these neoplasms are found in the stomach, making it the major site for diagnostic and therapeutic management [8]. Small intestinal GISTs are seen at prevalence rates between 20% and 30%, making it the second most prevalent site after the stomach. Duodeno-jejunal (DJ) flexure-localized GISTs are extremely uncommon and underreported in the current literature [2]. The diagnosis and treatment strategy for a tumor depend heavily on its anatomical location. The heterogeneous clinical behavior of GISTs, ranging from indolent tumors with a low risk of recurrence to those displaying aggressive and metastatic tendencies, further highlights the need for individualized approaches to treatment [4]. The complexity of the tumor's clinical behavior calls for a comprehensive knowledge of the tumor's anatomical location and related dangers.

This case offers insight on the extremely rare occurrence of a Gastrointestinal Stromal Tumor (GIST) at the Duodeno-Jejunal (DJ) flexure, an anatomical position with its own set of diagnostic problems. Despite their rarity, GISTs should always be considered a possible underlying cause of gastrointestinal bleeding due to the patient's clinical history of chronic stomach pain and melena (bloody stools). Immunohistochemical staining for CD117, a marker for GISTs, and mutation analysis, which can reveal particular genetic abnormalities like KIT or PDGFRA mutations, can aid in making a correct diagnosis. Accurately diagnosing GIST is critical since it not only helps to confirm the diagnosis but also directs subsequent treatment. Imatinib and other tyrosine kinase inhibitors have become indispensable tools for improving patient outcomes in the treatment of advanced or irresectable GISTs [5]. It is clear

from this case that doctors need to be more alert to the possibility of GISTs occurring in non-typical sites, and that prompt and precise identification is crucial for providing the best care possible to patients.

MATERIALS AND METHODS

A 51-year-old male presented with breathlessness, generalized weakness, and melena for 5 days. On examination: The patient was pale but hemodynamically stable. Systemic examination showed no significant findings. Per rectal examination was normal except for black-colored stools. Blood investigations were normal except for low haemoglobin (7 gm%)

Ultrasound Abdomen Pelvis: DJ flexure Mass abutting superior mesenteric vessels anteriorly & aorta posteriorly.

CECT scan of Abdomen Pelvis showed 6.2 x 4.7 x 4.3 cm well defined heterogenous exophytic lesion arising from Duodeno-Jejunal flexure with foci of calcifications & luminal narrowing. The lesion was in close proximity to the uncinate process of the pancreas.

CT scan-guided biopsy confirmed the diagnosis of GIST. Tumor cells are positive for C-KIT (CD 117), DOG-1 and SMA on Immunohistochemistry.

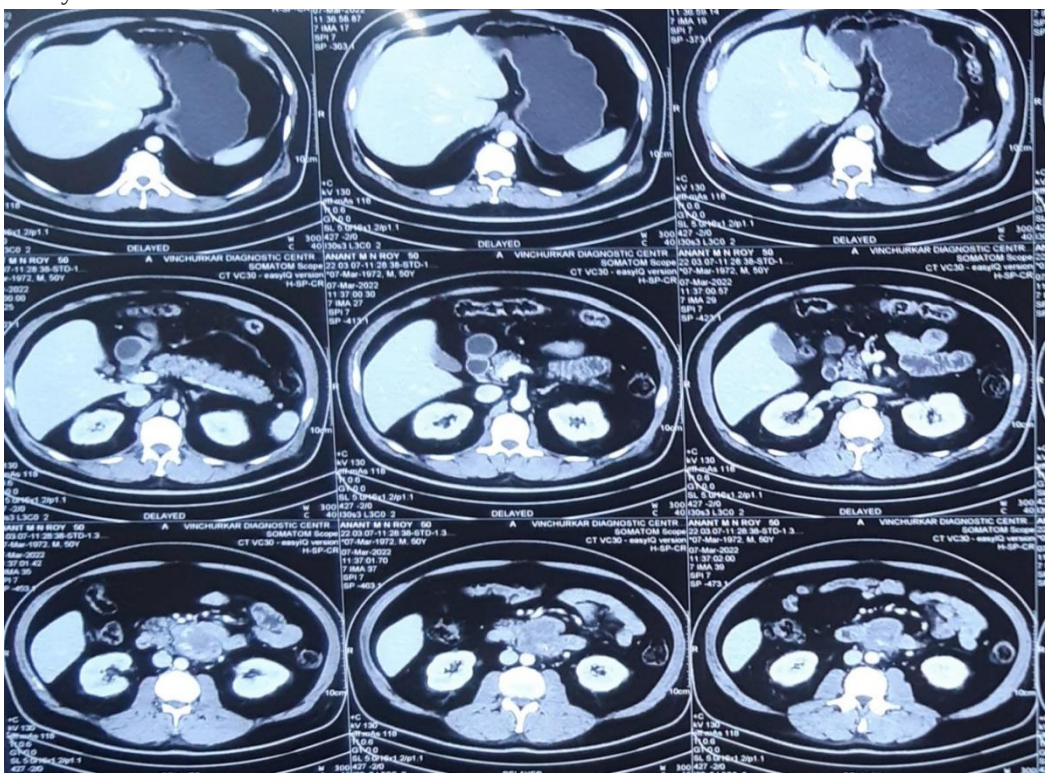

Figure 1: CT SCAN ABDOMEN WITH CONTRAST

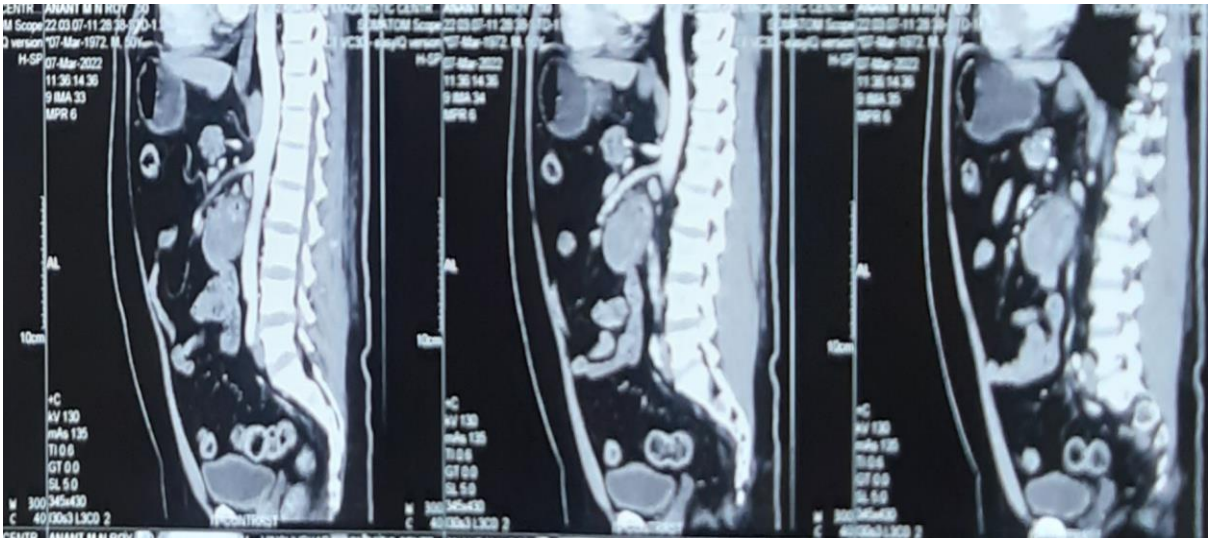

Figure 2: CT SCAN ABDOMEN WITH CONTRAST

RESULTS

After stabilization of general condition with blood transfusion, Surgery was planned because of impending duodenal obstruction & ongoing melena.

Intraoperative findings: 6.2 x 4.7 x 4.3 cm large mass involving DJ flexure with luminal obstruction. Mass was adherent to the uncinate process of the pancreas & Left inferior mesenteric vein.

Third & fourth parts of Doudenum were resected in toto followed by Duodeno-Jejunostomy side to side anastomosis.

The final histopathology report confirmed the diagnosis of Duodeno-Jejunal Flexure Gastrointestinal Stromal Tumour with clear margins. In the postoperative period, the patient developed mild pancreatitis which was managed conservatively.

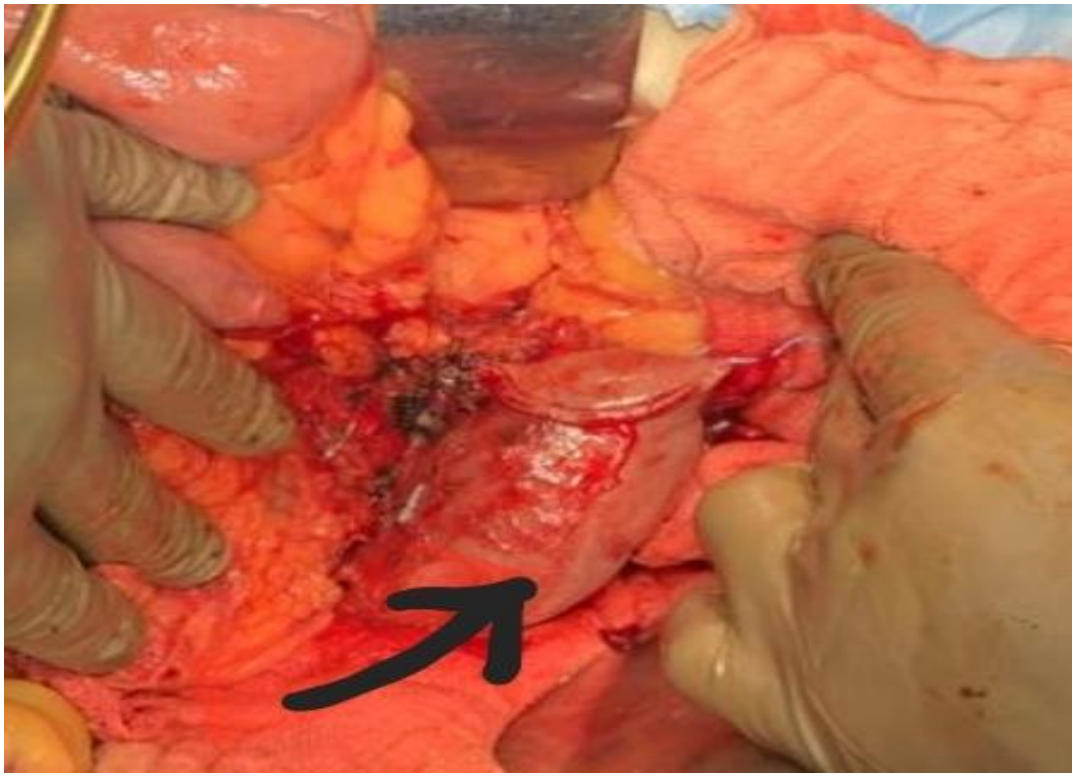

Figure 3: Intra- Operative Picture with Arrow showing Tumour site

CONCLUSIONS

Bleeding Duudeno-Jjejunal (DJ) flexure GIST is a rare cause of GI bleeding and can be extremely difficult to diagnose due to the inaccessibility of endoscopy.

The mainstay of management for DJ Flexure GIST is complete surgical excision. The technical difficulty of complete resection because of unusual location, ongoing intra-operative bleeding from the tumour and the chances of vascular injury with an anastomotic leak made this case a surgical challenge. The survival rate is increased with the use of Imatinib Mesylate.

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