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ATYPICAL PRESENTATION OF GASTROINTESTINAL STROMAL TUMOR AT THE GASTRO ESOPHAGEAL JUNCTION

Gastroenterology

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ABSTRACT

Gastrointestinal stomal tumors are the most common mesenchymal neoplasms of the GIT. Originally thought to be distinct type of smooth muscle sarcoma they are now known to be derived of interstitial cells of cajal. They can occur anywhere in the GIT, stomach being the most common and esophagus being the least common. They can manifest in any age, usually present in individuals older than 50 years and most commonly are asymptomatic. Cross sectional imaging and endoscopic ultrasound are useful for diagnosis. Complete surgical resection is the primary modality of treatment. The below case highlights the uncommon presentation of this common tumor at the gastroesophageal junction, an atypical location in a young female who presented with chronic abdominal pain she was subject to routine investigations and cross-sectional imaging which revealed the presence of the lesion at the GE junction following which she was subject to laparoscopic removal of the lesion.

KEYWORDS

Gastrointestinal Stromal Tumor, Gastro Esophageal Junction.

Case Report:
A 26 year old female patient presented with the chief complaint of abdominal pain since 1 year which was insidious in onset, gradually progressive. Aggravated on eating food. Associated with bloating sensation.

No history of vomiting/diarrhea/constipation
No history of hematemesis
Patient has no co morbidities and no significant family history
On examination per abdomen was soft, with no tenderness, no guarding or rigidity and no palpable lump or organomegaly.

The patient underwent laparoscopic transgastric resection of the lesion which was intra operatively found to be a 3*2 cm soft polypoidal lesion at the gastro esophageal junction.

HPE and IHC staining revealed CD117 postivity which confirmed the diagnosis of GIST.

Post operative period was uneventful. Patient was kept on regular follow up.

BLOOD INVESTIGATIONS:	Hb- 11.6 g/dl WBC- 9,870/UL Plt- 3,65,000/UL Sr. creatinine – 0.51mg/dl Na- 136 meq/lit K- 4.1 meq/lit TBIL – 0.43mg/dl DBIL – 0.08 mg/dl ALP – 51 U/L AST- 12.06 U/L ALT- 19.14 U/L BGT- O positive
Viral markers:	HIV – negative HBsAg – negative HCV - negative
UGI Endoscopy:	Polypoidal lesion with wide base seen in the fundus/ cardia of stomach
Endoscopic ultrasound:	Heteroehoic lesion measuring 23*14mm noted at the GE junction arising from the fourth layer, it has cystic spaces and minimal increased vascularity. There are no obvious peri lesional lymphnodes.
CECT of abdomen and pelvis:	Well defined polypoidal soft tissue dense lesion with minimal enhancement measuring 22*23*29 mm arising from the GE junction

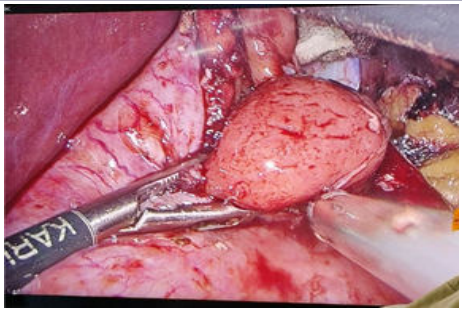

Figure 1: Intraoperative picture showing the lesion

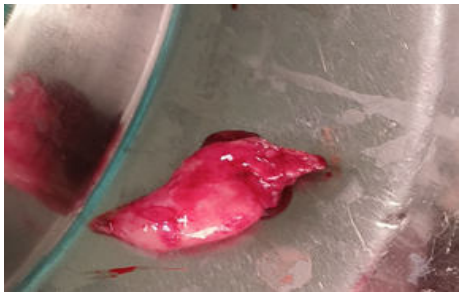

Figure 2: Picture of the excised specimen

DISCUSSION:
Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal (GI) tract. They are derived from interstitial cells of Cajal, a GI pacemaker cell. They may be found anywhere in the GI tract, from the esophagus to the internal anal sphincter.

The most common GI location is the stomach (56%), followed by the small intestine (32%), colon and rectum (6%), and esophagus (<1%). The remaining 5% of lesions occur in other less common locations, including the mesentery, pelvis, pancreas, liver, omentum, and genitourinary tract.

GISTs are diagnosed based on morphologic features supported by immunohistochemical (IHC) studies.

Histologically, GISTs can occur in three different types: spindle cell, which accounts for approximately 70% of tumors, epithelioid, and mixed, which contains a variable combination of spindle cells and epithelioid cells.

The basic underlying pathophysiology in the majority of lesions involves a gain of function mutation leading to increased activity of the

KIT tyrosine kinase receptor on the cell membrane. Some lesions may also develop due to gain of function mutations in platelet-derived growth factor receptor A (PDGFRA), or BRAF.

CD117 IHC staining has now been widely accepted as the primary criterion for a pathologic diagnosis of GIST, with a reported sensitivity greater than 95% and a high specificity. Additional IHC markers that can be used are PDGFRA, c- KIT and DOG1.

GISTs are often asymptomatic and discovered only as incidental findings on imaging studies or endoscopy performed for other indications. Patients with a symptomatic GIST often have symptoms that are related to the size and location of the tumor. Early in disease progression, symptoms may be very nonspecific, such as mild pain, bloating, or dyspepsia. More significant symptoms generally do not occur until much later in the course of the disease, when they have grown quite large. This is likely due to their submucosal location, exophytic growth, and propensity to displace rather than invade adjacent organs. Patients presenting with large GISTs may have palpable tumors and present with pressure or pain or symptoms. Patients may also present with acute GI blood loss or symptoms of chronic anemia and associated fatigue resulting from overlying mucosal ulceration, or with peritonitis related to tumor perforation.

Suspected GISTs should be evaluated with contrast-enhanced computed tomography (CT) and/or magnetic resonance imaging (MRI). Large GISTs appear as hypervascular, enhancing masses and are often heterogeneous because of necrosis, hemorrhage, or cystic degeneration. They often displace but rarely invade adjacent organs. Smaller GISTs are usually more homogeneous and polypoid in appearance.

On endoscopy a submucosal mass may be seen that is smooth in appearance and often appears to form a bulge into the lumen of the stomach. Ulceration may be seen in lesions that present with bleeding or anemia.

Biopsy of a suspected GIST is not required if it appears surgically resectable.

Endoscopic ultrasound (EUS) may also aid in the differentiation of a submucosal gastric mass versus impingement from surrounding organs (i.e., pancreatic mass, pseudocyst). It is also useful in biopsy of these lesions due to their submucosal location. It is particularly important in the management of very small (<2 cm) gastric GISTs. Chest imaging, PET/ CT scans are useful in the staging of the disease and assessing the response to therapy.

American Joint Committee on Cancer (AJCC) staging of GISTs includes tumor size, presence of nodal metastases, distant metastases, and grade (high grade is > 5 mitoses/50 high-power field). GISTs rarely metastasize to lymph nodes, but distant metastases, particularly to the liver and peritoneum, are not uncommon.

Complete resection with at least a 1- to 2-cm gross margin should be the goal of surgical treatment. Care must be taken not to violate the tumor pseudocapsule or rupture the tumor because violation of the tumor pseudocapsule causes risk for subsequent tumor seeding into the peritoneum. Lymphadenectomy is not indicated unless enlarged or pathologic nodes are seen on imaging or intraoperatively because GISTs rarely spread to regional nodes.

Current guidelines do not generally recommend laparoscopic resection for tumors greater than 5 cm in diameter.

A 2008 study proposed a classification schema to identify three distinct minimally invasive approaches to resecting gastric GISTs based on tumor location in the stomach. Type I tumors were located in the fundus or greater curvature and were treated using a laparoscopic stapled partial gastrectomy. Type II tumors were located in the antrum/prepyloric region and were approached using laparoscopic distal gastrectomy. Type III tumors were located in the lesser curvature near the gastroesophageal junction and were resected by laparoscopic trans gastric resection.

Follow-up after surgical resection of GISTs generally consists of history and physical exam every 3 to 6 months for 5 years, then annually, as well as serial CT scans every 3 to 6 months for 3 to 5 years, then annually. Approximately 40% to 50% of GISTs will recur or develop metastases, even after potentially curative resection.

Imatinib is used for recurrent, locally invasive, or metastatic GIST. Sunitinib and Regorafenib can also be used.

Patients who present with locally advanced disease, large tumors, or tumors at difficult anatomic sites may benefit from preoperative treatment with imatinib. This may decrease the size of the tumors, allowing a more limited surgical procedure to be performed, improving organ preservation and enabling complete resection.

Current guidelines mainly advocate the use of radiation only in the palliation of bone metastases from GISTs.

Imatinib remains the primary treatment for recurrent or metastatic GISTs.

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