An Unusual Presentation of a Large Gastrointestinal Stromal Tumor (GIST)

Jamil Shah, Praneeth Bandaru, Vijay Gayam, Vikash Kumar, Philip Xiao, Madhavi Reddy, Denzil Etienne

Introduction

- Gastrointestinal stromal tumors (GISTs), although the most common mesenchymal neoplasms of the digestive tract, are relatively rare primary GI cancers.
- They originate from the interstitial cells of Cajal, which are cells of the intestinal autonomic nervous system.
- They function as electrical pacemakers, controlling motility as well as regulating peristalsis.
- Individuals with GISTs can present with overt or occult GI bleeding, but, more frequently, they present with nonspecific symptoms, such as vague abdominal pain or discomfort, early satiety, or bloating.
- Other individuals may be asymptomatic, and the GISTs are detected incidentally during upper endoscopy (where they usually appear as subepithelial masses) or on cross-sectional imaging studies performed for a different reason.
- Here, we present an interesting case of an elderly woman discovered to have a large GIST.

Case Description

- A 69-year-old woman, with a PMH of bilateral unprovoked pulmonary embolisms (submassive with evidence of right heart strain) in August 2017 on anticoagulation (Warfarin), HTN, and DM, underwent EGD and EUS in 2018 that showed a 3-4 cm umbilicated, firm, round mass along the posterior wall/greater curve of the stomach. FNB was performed (Figure 1).

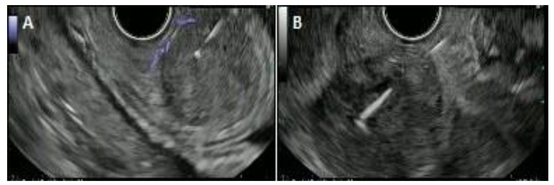

Figure 1. A, B) A submucosal GIST was seen on the posterior wall of the proximal gastric body, approximately 3 cm from the GE junction, during EUS. FNB was performed.

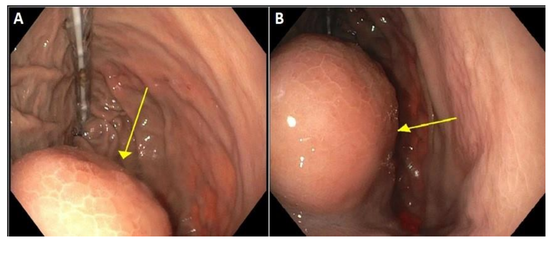

Figure 2. A, B) A 3-4 cm umbilicated, firm, round mass along the posterior wall/greater curve of the stomach seen during EGD.

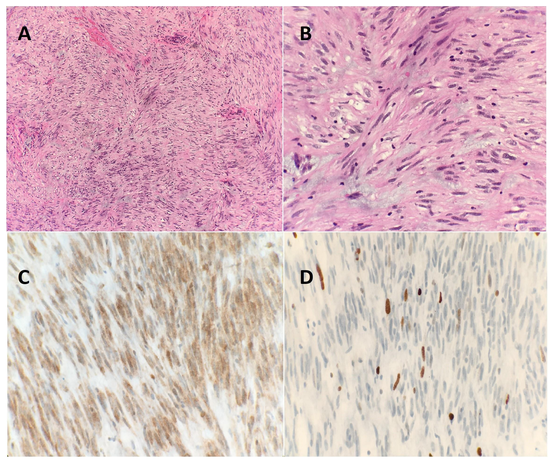

Figure 3. Mediastinal mass needle biopsy:
-Minute fragments of malignant neoplasm with necrosis.
-Tumor cells are positive for AE1/3, CAM5.2, Chromogranin, Synaptophysin, CD56 and negative for p40, CD3, CD20, and CD45. Ki-67 demonstrates about 60% positivity. Combined with morphological features, this immunoprofile supports the diagnosis of small cell carcinoma.

A) Hematoxylin and eosin (H&E) stain, low power. B) Hematoxylin and eosin (H&E) stain, high power. C) CKit. D) Ki-67.

Case Description continued…

- The pathology from FNB at the time was suggestive of gastrointestinal stromal tumor (GIST) (+ C-kit, CD117).
- She subsequently failed to follow up with GI.
- Then, recent CT A/P showed a small hiatal hernia, and a filling defect in the stomach, 3.2 x 3.1 cm, arising from the lesser curvature immediately distal to the GEJ.
- The patient underwent EGD in January 2022 for intraoperative evaluation of the GIST tumor (Figure 2), followed by robotic-assisted surgical wedge resection of the gastric tumor by the surgical team (Figure 3).

Discussion

- GISTs are identified primarily by the expression of the KIT protein and often carry activating mutations in either the KIT or the platelet-derived growth factor receptor alpha (PDGFRA) genes.
- These neoplasms are frequently discovered in the stomach (40 to 60 percent) and the small intestine (30 to 35 percent).
- However, they can arise in any part of the digestive tract.
- Resectable GISTs can be completely, or almost completely, removed by surgery.