A Rare Case of a Large Esophageal Gastrointestinal Stromal Tumor

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INTRODUCTION

- GISTs are rare mesenchymal neoplasms, 1-2% of all primary GI cancers
- Symptoms are location dependent but can include abdominal pain, nausea, vomiting, early satiety, bleeding, and dysphagia
- Majority of tumors found incidentally
- 40-60% in stomach, 25-30% in small bowel, 5-15% in colon and rectum
- We present a case of an elderly patient diagnosed with esophageal GIST after he was found to have an incidental esophageal mass on imaging.

CASE PRESENTATION

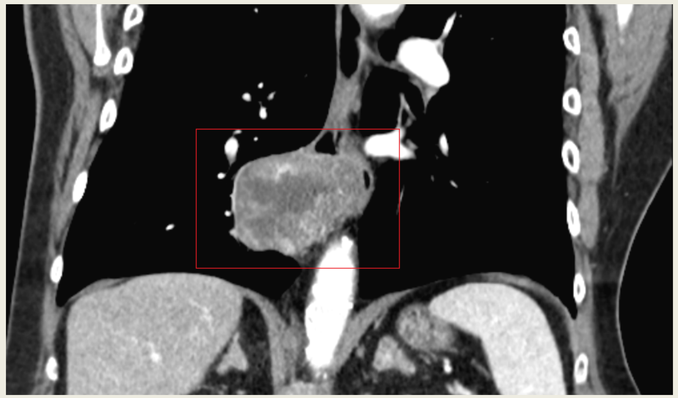
Background:
- 80-year-old male referred for an upper endoscopy for evaluation of an incidental finding on CT scan
- Past medical history notable for HTN, HLD, DM2, CKD Stage III, COPD, former tobacco user
- ROS negative for dysphagia, abdominal pain, nausea, vomiting, weight loss, hematemesis, or melena

Imaging:
- CT revealed a mass arising from the distal esophagus, measuring 6.5 x 7.5 cm (A)
- Endoscopy showed submucosal mass in the distal esophagus (B)
- Upper endoscopic ultrasound (EUS) showed an intramural lesion in the distal esophagus arising from the muscularis propria (C)

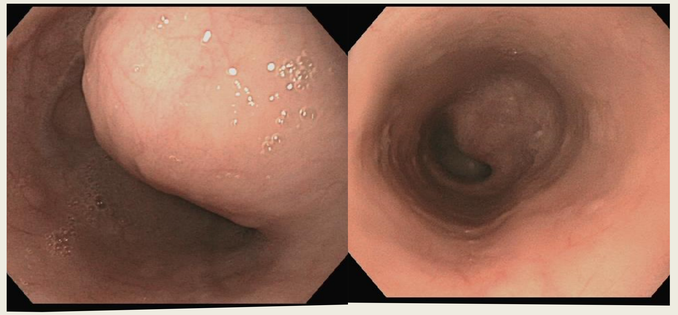
Additional Testing:
- Pathology results confirmed GIST, positive for CD117 and DOG-1
- Further testing revealed a mutation in the KIT gene exon 11

Treatment:
- Patient initiated on neoadjuvant therapy with imatinib mesylate with anticipated surgical resection.

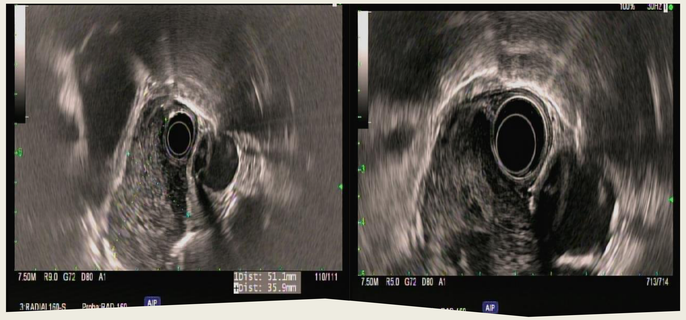
IMAGES



(A)



(B)



(C)

DISCUSSION

- Most mesenchymal esophageal tumors are leiomyomas, and GIST only accounts for 0.7% of them
- EUS with fine needle aspiration biopsy is the most accurate method for diagnosis and definite diagnosis is made histologically
- Majority of GISTs, expresses the CD117 antigen, a KIT receptor tyrosine kinase.
- Platelet derived growth factor (PDGFRA) tyrosine kinase gene is another commonly found mutation
- Tumor size and mitotic rate are used for prognostication, with Increased malignancy risk seen in tumor size > 3 cm with irregular margins
- Tumor location is also a key factor in risk stratification; non gastric primary location has higher rates of metastases
- Most common sites of metastases are liver and peritoneum
- Treatment is surgical resection and neoadjuvant therapy with imatinib

CONCLUSION

- Esophageal GISTs are extremely rare and difficult to diagnose and treat.
- Given significant implication for its management, esophageal GIST should be considered in differential diagnosis and distinguished from more common esophageal leiomyomas

REFERENCES

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