

Gastrointestinal Stromal Tumor- An Unusual Presentation

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ABSTRACT

Gastrointestinal stromal tumor (GIST) is the most common mesenchymal tumor of the gastrointestinal tract which earlier was classified as leiomyomas, leiomyosarcomas and leiomyoblastomas. These are known to have specific markers and are important to diagnose as targeted chemotherapy is available. Our case represents one of the ends of the spectrum of clinical presentation of gastrointestinal tumors – an aggressive large mass at a younger than the usual age of presentation. Though challenging, imaging diagnosis of GIST is very important as it can guide the confirmation of the same by identification of the markers on pathology.

Keywords: Gastrointestinal stromal tumors, GIST, Mesenchymal tumors.

INTRODUCTION

Gastrointestinal stromal tumor (GIST) is the most common mesenchymal tumor of the gastrointestinal tract which earlier was classified as leiomyomas, leiomyosarcomas and leiomyoblastomas. Diagnosis of GIST is restricted to specific c-kit expressing and Kit-signal driven mesenchymal tumors.¹ Our case represents the presentation of GIST as an aggressive large mass arising from stomach at a younger age than usual.

CASE REPORT

A 19 year old female presented to the hospital with few episodes of hematemesis and features

of severe anemia which was confirmed on the blood examinations. Endoscopic examination showed three large submucosal proliferative growth in antrum, lesser and greater curvature, one with ulceration in the body of stomach, biopsy of which revealed mild chronic active gastritis (Image 1). On contrast enhanced CT scan of the patient, approx. 10.9 x 7.2 x 19.8 centimeter sized heterogeneously enhancing large exophytic mass arising from the body of stomach abutting the surrounding structures with maintained fat plane (Image 2). Imaging diagnosis was given as suspicious of malignant GIST of gastric origin with minimal pelvic ascites. Patient also had cardiomegaly, cardiogenic pulmonary changes and minimal bilateral pleural effusion which could be attributed to anemia. USG guided biopsy of the mass was done which showed multiple fragments of tissue composed of diffuse proliferation of spindle cells having eosinophilic cytoplasm with positive CD117, negative for desmin and S100. A final diagnosis of gastric GIST was

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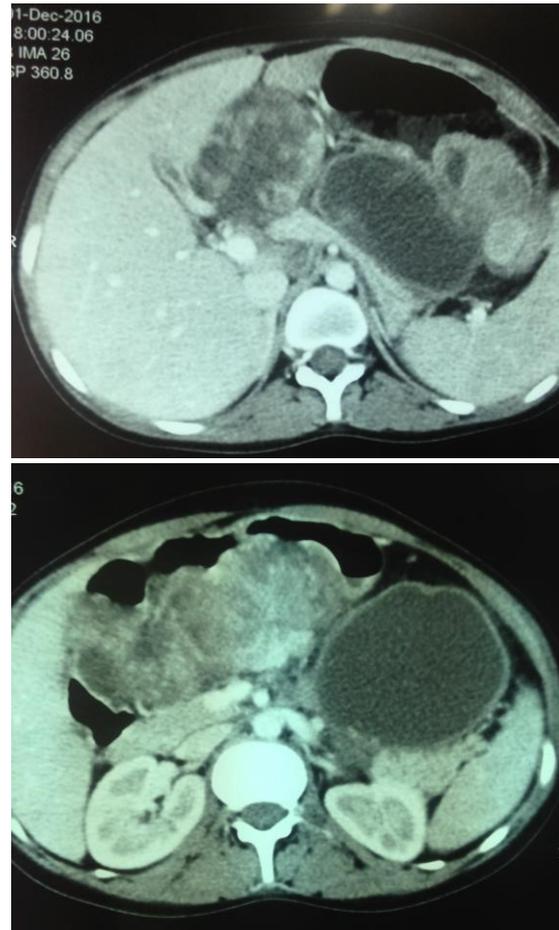


Image 1: Endoscopic images with stomach fundus and antrum showing submucosal lesion. Areas of mucosal ulceration are also seen in the lesion in the antrum.

Image 2: Contrast enhanced axial CT images showing heterogeneously enhancing large exo-endophytic mass in the region of gastric body and antrum.

made .

DISCUSSION

GIST is the most common mesenchymal tumor of the gastrointestinal tract which originates from the intestinal interstitial cells of Cajal. Majur and Clark in 1983 gave the term GIST because they lacked immunohistochemical features of Schwann cells and did not have ultrastructural characteristics of smooth muscles.² Functional mutations of KIT protooncogene is present in about 95% of GIST. GIST are positive for KIT (CD117) and about 70% express CD34. The intracytoplasmic portion of KIT functions as a tyrosine kinase and hence Imatinib Mesylate which inhibits tyrosine kinase

is highly effective for treatment of GIST.³ Rarely, GIST may be familial with a mutated KIT more often affecting young women, as a part of the Carney triad of gastric GIST, functioning extra-adrenal paraganglioma and pulmonary chondroma, first described in by Carney in 1977.⁴ GIST does not show significant racial or gender bias occurring only slightly more in males (55%), usually between 40-80 years with median age of 60 years with only 3% of GIST diagnosed before age of 21 years. The most common location of GIST is the stomach (60%) followed by small intestine (30%), duodenum (5%), colorectum (<5%), esophagus and appendix (1%) and rarely, peritoneum, its reflections and retro peritoneum. The diagnosis may be incidental or patient may be symptomatic, most common symptoms being vague abdominal

pain and discomfort or severe like blood loss, perforation, etc.⁵

Usual origin of GIST is from the muscularis propria of intestinal wall with propensity for exophytic growth, size ranging from few millimeters to 30 cm. Mucosal ulceration may be seen in upto 50% of cases. Most important prognostic factors are size > 5 cm, tumor necrosis, infiltration and metastasis to other sites, mitotic counts >1-5 per 10 high-powered fields and mutation in the c-kit gene.⁶

Abdominal radiography may show a nonspecific soft tissue mass rarely with calcification. In barium studies, GISTs have the characteristic features of submucosal masses - that is, make an obtuse angle with the wall of the bowel on profile view and smoothly circumscribed on en face view with focal area of ulceration in 50% of cases.⁷

On cross sectional imaging, the tumor has an intramural with usually a larger exophytic component with predominantly peripheral enhancement and central non enhancing areas of hemorrhage and necrosis. Small lesions may have homogenous enhancement. Sometimes, bulk of the tumor may be extraluminal, attached to the organ of origin by a thin pedicle or wall thickening at the attachment site. Locoregional spread, presence of ascites and liver metastasis need to be evaluated. Calcification and nodal metastasis are rare. MRI is similar in sensitivity to CT scan for the diagnosis and evaluation of the locoregional relationships of the tumor. The peripheral area of the tumor shows low signal intensity on T1 and high signal intensity on T2 weighted sequence, while the signal of the central area is variable depending upon presence of hemorrhage and necrosis.⁸

PET scan is a useful for the detection of metastatic deposits of the tumor and early detection of clinical response of the tumor to therapy with imatinib mesylate.⁹ Endoscopy

is done in the early workup of patients with gastrointestinal symptoms where GIST may be seen as a smooth submucosal mass displacing the overlying mucosa, few with ulceration or bleeding of the overlying mucosa.⁶ Endoscopic ultrasound allows localization of the lesions and their characterization as well as obtaining tissue for histopathological diagnosis. The typical ultrasound appearance of GIST is that of a hypoechoic mass situated in the layer corresponding to the muscularis mucosa.¹⁰

Other mesenchymal neoplasms such as true leiomyomas, schwannomas, neuroendocrine neoplasms etc, which may also arise in the gastric wall may simulate gastric GISTs, are however, not so frequent. The other differentials for gastric

GIST may be adenocarcinoma and lymphoma which predominantly have mural location or an intraluminal component and rarely demonstrate marked exophytic growth. Lymphadenopathy may be seen in both these which are unusual in GISTs.

CONCLUSION

There are very few reported cases of such a large gastric GIST in a young female. It must be considered in the differential diagnosis of a gastric mass with a predominant exophytic component. Histopathological confirmation is necessary as large masses, which may not be amenable to surgery show good response to Imatinib, a tyrosine kinase inhibitor.

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