Case Report

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Cystic Esophageal Gastrointestinal Stromal Tumor (GIST): A Rare presentation

Junaid Iqbal, Sadia Rashid, Hina Pathan, Sorath Murtaza

ABSTRACT

Gastrointestinal stromal tumors (GIST) are solid tumors of the gastrointestinal tract. The stomach is the most common site while the esophagus is the least common site for gastrointestinal stromal tumors. The cystic Gastrointestinal stromal tumor has never been reported in the past. On CT scan of a young male patient, we reported an exophytic cystic structure of 2.5 x 2.2 cm along the esophagus as an esophageal duplication cyst then he underwent an image-guided biopsy that finally turned out to be a Gastrointestinal stromal tumor on histopathology. He underwent surgical resection; he is now symptoms-free and on 6 monthly CT scan follow-ups.

How to cite this Article:
Iqbal J, Rashid S, Pathan H, Murtaza S. Cystic Esophageal Gastrointestinal Stromal Tumor (GIST): A Rare presentation. J Bahria Uni Med Dental Coll. 2022; 11(3):169-171 DOI: https://doi.org/ 10.51985/JBUMDC202139

INTRODUCTION:

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors of the gastrointestinal tract. They account for up to 3% of all GIT tumors and up to 5.7% of sarcomas.[1] GIST arises from any part of the gastrointestinal tract (GIT), the most common site is stomach followed by the small bowel, colon, and rectum. The esophagus is the least common site, accounting for <1% of all GISTs.[1, 2, 3] The annual incidence of gastrointestinal stromal tumors is 7 to 20 per million. Esophageal GISTs occurred significantly more frequently in men, as well as in patients younger than 60 years at diagnosis. The most common location for esophageal GISTs is the lower esophagus, followed by the middle esophagus , whereas GISTs in the upper esophagus are rare. The clinical features of esophageal GISTs are not well-known due to rarity of esophageal GIST. [4]

The literature on esophageal GISTs is limited, mainly comprising case reports and only a few case series.[1]

CASE PRESENTATION:

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Received: 14-Jun-2021
Accepted: 11-Apr-2022

A 35-year-old male came to OPD with chest pain, dysphagia, and odynophagia for two months. CT scan was done which showed an exophytic cystic lesion along the right posterolateral wall of the cervical esophagus. It was a well-defined hypodense mass not showing enhancement on post-contrast images (Fig 1 & 2 white arrow), measuring 2.5 x 2.2 cm. Initially it was diagnosed as a duplication cyst. MRI was done as presurgical planning which showed well defined exophytic lesion. It was hypointense on T1W, slightly hyperintense on T2W, and showed minimal enhancement on post-contrast images (Fig 3 & 4 white arrowhead). Endoscopy was not performed as the patient did not give consent. Due to minimal enhancement, an ultrasound-guided trans-thyroid biopsy was performed and it came out to be an esophageal gastrointestinal stromal tumor (GIST), spindle cell type with the expression of Ki67 and CD117 on histopathology. The patient underwent an esophagectomy with end-to-end anastomosis. The tumor did not involve the esophageal sphincter and was located approximately 15 cm proximal to the gastro esophageal junction. The histopathology of the surgical specimen showed that, it is low-grade gastrointestinal stromal tumor (GIST). He recovered and was discharged successfully after 8 days. He is currentlyon imatinib (tyrosine kinase inhibitor). The patient is now asymptomatic and is on 6 monthly follow-up.

DISCUSSION:

Gastrointestinal stromal tumors (GISTs) were previously classified as smooth muscle tumors but now proved that they arise from the interstitial cells of Cajal and on immunohistochemistry they express KIT protein-CD117. GIST can arises from any part of the gastrointestinal tract (GIT) including the omentum. They usually originate from the muscularis propria. The tumor growth is exophytic rather than intraluminal or intramural.[5]

GISTs of the esophagus are usually asymptomatic especially when they are small but as the tumor grows, patients present

with different symptoms due to pressure effects like dysphagia, chest pain, reflux, and heartburn[6]. Larger tumors may cause obstruction of the gastrointestinal lumen.[7] GISTs are usually reported in middle-aged and elderly people, and they range in size from sub centimeters up to 40cms.[8] Smaller GISTs (< 2cm) have almost no chance of invasion or infiltration, however, GIST cannot be labeled as benign.[9] On Computed Tomography scan, GISTs appear as a large exophytic, hypervascular, enhancing solid mass and sometimes appear heterogeneous due to necrosis, hemorrhage, or cystic degeneration.[10] The GISTs are slow-growing tumors and have a prolonged survival period that's why imaging has great importance, not only for diagnosis but also to see the response of treatment. Various imaging techniques are available for the detection of GISTs including fluorine 18 fluorodeoxyglucose (FDG) positron emission tomography (PET), magnetic resonance imaging (MRI), and ultrasonography (US) but the imaging modality of choice is Computed Tomography (CT). Although FDG PET is most sensitive for the detection of GISTs, however, it is not easily available everywhere. [11, 12]

The differential diagnosis for esophageal GISTs on imaging depends upon the location and size. Smaller tumor limited to the wall of the esophagus most likely represents leiomyomas with differentials of duplication cyst, GIST, lipoma, and hemangioma. The endoluminal ultrasonography (EUS) with tissue biopsy is useful for histopathological diagnosis and further management of esophageal lesions.[11]

As esophageal gastrointestinal stromal tumors are very rare that is why there is a lack of clear recommendations regarding their surgical management. The surgical options range from the highly invasive esophagectomy to the much less invasive surgical tumor enucleation. Due to anatomical peculiarity of the esophagus, segmental-wedge resections are not usually performed.

Preoperatively it is difficult to categorize an esophageal tumor into benign or malignant by imaging examinations, like endoscopic ultrasound (EUS), computed tomography (CT), magnetic resonance imaging (MRI) andFluorodeoxyglucose positron emission tomography (FDG-PET). [18] Endoscopic or image-guided biopsy provides important information preoperatively and helps in surgical planning, but biopsy is considered a controversial technique due to the risk of tumor rupture and seeding. The difficulty in preoperative diagnosis makes it difficult for surgeons to select the surgical method. Imatinib, a tyrosine kinase inhibitor (TKI), has been shown to be highly effective in metastatic and neoadjuvant therapy. However, due to the rarity of esophageal GISTs, there is limited literature available regarding the neoadjuvant administration of Imatinib in patients with esophageal GISTs.[4] The aim of Imatinib therapy is downsizing the GIST to reduce the extent of resection and reduce the risk of intraoperative complications, including tumor rupture and reduce the chances of recurrence.

CONCLUSION

Esophageal gastrointestinal tumors are rare. Endoscopic or image-guided biopsy gives a definite diagnosis, but there is a risk of tumor rupture or tumor dissemination. Surgical resection is the only curative treatment for localized GISTs with the use of imatinib preoperatively and postoperatively. In addition to the most common imaging appearance of GIST as a solid tumor, it can also present as a cystic lesion and needs to be considered in the differential diagnosis of cystic esophageal lesions.

Figure.1 CT scan non enhanced

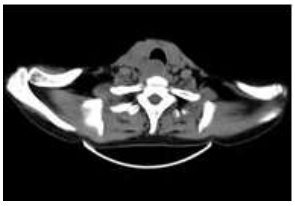


Figure.2 CT scan contrast enhanced

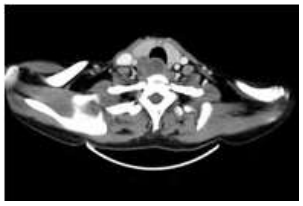


Figure.3 MRI T1W

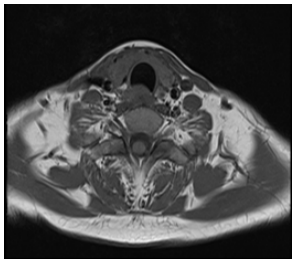
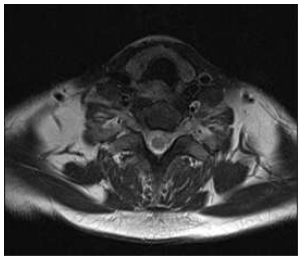


Figure.4 MRI T2W



Authors Contribution:
Junaid Iqbal: Conception
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