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Gastrointestinal Stromal Tumor Of The Oesophagus- A Rare Case Report.

Sumiti Gupta[1], Ankush[2]*, Mahak[3], Akanksha[4], Veena Gupta[5], and Sunita Singh[6].

[1]Professor, Department of Pathology, Pt. B. D. Sharma Post Graduate Institute of Medical Sciences,Rohtak, Haryana, India.
[2]Junior resident, Department of Pathology, Pt. B. D. Sharma Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.
[3]Senior resident, Department of Pathology, Pt. B. D. Sharma Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.
[4]Junior resident, Department of Pathology, Pt. B. D. Sharma Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.
[5]Professor, Department of Pathology, Pt. B. D. Sharma Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.
[6]Senior Professor and Head, Department of Pathology, Pt. B. D. Sharma Post Graduate Institute ofMedical Sciences, Rohtak, Haryana, India.

ABSTRACT

Gastroesophageal stromal tumors (GIST) often arise in stomach and small intestine, while esophageal GISTs are rare. Due to their rarity, there is always a lack of clarity in concerning optimal surgical management for esophageal GIST. It is difficult to distinguish esophageal GIST from leiomyoma, the most frequent esophageal mesenchymal tumor.Case report: A 53-year-old male presented to surgery department with chief complaint of dysphagia for six months and weight loss for one month. CT scan imaging of the patient showed circumferential long segment asymmetrical mural wall thickening in lower esophagus for about a length of 60 mm corresponding to D7 to D10 level causing luminal narrowing and mild proximal dilatation of esophagus, mitotic. Surgical exploration and pathologic specimens resulted in diagnosis of malignant mesenchymal tumor of esophagus. Conclusion: Esophageal GISTs pose a diagnostic challenge because they lack specific findings that distinguish them from more common leiomyomas during preoperative assessments such as endoscopy, endoscopic ultrasound, or CT scan. Both GISTs and leiomyomas present as hypoechoic lesions originating from the muscularis propria or muscularis mucosa on endoscopic ultrasound. Therefore, a definitive diagnosis can only be established through histopathological examination of tissue samples obtained during surgical exploration or biopsy. This underscores the importance of histopathological correlation in accurately diagnosing esophageal GISTs.
Keywords: Gastrointestinal stromal tumor(GIST), Esophagus

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*Corresponding author

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INTRODUCTION

Esophageal mesenchymal neoplasms, including GISTs, particularly malignant one are indeed rare compared to the more common malignant tumors of the esophageal tract, such as squamous carcinoma and adenocarcinoma. Leiomyomas are more commonly found in the esophagus compared to other parts of the gastrointestinal tract. Gastrointestinal stromal tumors (GISTs), which exhibit phenotypic features of interstitial cells of Cajal, are the most prevalent malignant mesenchymal tumors of the gastrointestinal tract [1]. Gastrointestinal stromal tumors (GISTs) can be classified pathologically into three types: spindle cell, epithelioid cell, and mixed cell type. Immunohistochemical panels, including markers like KIT(CD117), DOG-1, CD34, smooth muscle actin (SMA), desmin, and S100 protein, are crucial for distinguishing GISTs from other tumors [2]. While rare, GISTs predominantly affect specific regions of the digestive tract, with approximately 60.3% arising in the stomach, 33.2% in the small bowel, 3.1% in the rectum, and less frequently in the colon, esophagus, and appendix [3].

Clinically, esophageal GISTs typically present with a male predominance. Patients commonly experience dysphagia or odynophagia, followed by symptoms like gastroesophageal reflux and epigastric pain. Some tumors may be asymptomatic and discovered incidentally during oesophagoscopy or through imaging or endoscopy. Most esophageal GISTs are located in the distal esophagus [4]. Understanding these clinical and pathological features is essential for accurate diagnosis and appropriate management of esophageal GISTs.

In the case of a submucosal tumor found in the esophagus, the potential differential diagnosis for an esophageal GIST includes both malignant and benign tumors, such as leiomyoma, hemangioma, schwannoma, leiomyosarcoma, and papillary epithelioma [5]. Unfortunately, distinguishing between esophageal leiomyoma and GIST prior to resection is challenging because they appear similar on CT scans and endoscopic examinations [6].

Case Presentation

Here, we report a case of 53-year-old male who presented to the surgery OPD with chief complaint of dysphagia and weight loss for 6 months. On CT imaging,it showed a circumferential long-segment asymmetrical mural wall thickening seen in the lower esophagus for about a length of 60 mm corresponding to D7 to D10 level causing luminal narrowing and mild proximal dilatation of esophagus, mitotic. On the upper GI endoscopy, the patient showed, on the upper GI endoscopy, it showed ulcerated nodule lesion at 30 cm from incisor and the esophageal ulcerated lesion was likely to be malignant on upper GI endoscopy.

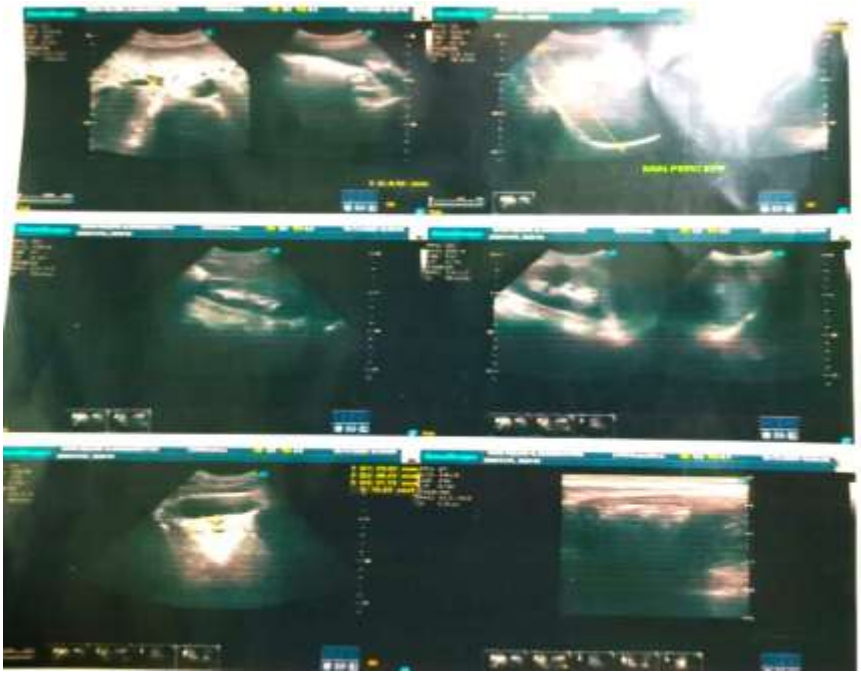

Figure 1: Ultrasonography of patient showing well defined hypoechoic lesion in lower esophagus.

Furthermore, the patient's esophageal biopsy was done and sent to the pathology department. Biopsy was received in multiple tissue grey brown to tan color bits measuring 0.5*0.3*0.2 cm. Histologically, the tumor cells were spindle shaped with eosinophilic cytoplasm arranged in short fascicles and syncytia, having elongated nuclei with inconspicuous nucleoli, indistinct cell borders, with >5 mitotic activity per 50 high power field making the diagnosis of malignant mesenchymal tumor. On immunohistochemistry these tumor cells showed positivity for vimentin CD117, DOG1 along with Ki67- 25 to 30% while CK and SMA were negative.

Patient had been put on Imatinib therapy 400 mg OD and will be on regular follow up.

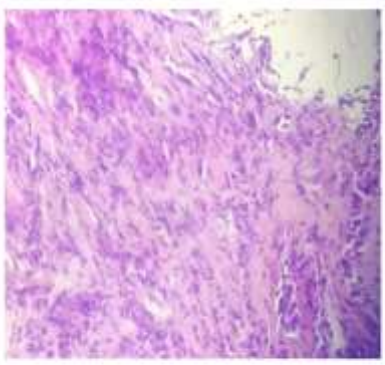

Figure 2: H&E stained section showing interspersed tumor cells along with necrosis. (100X)

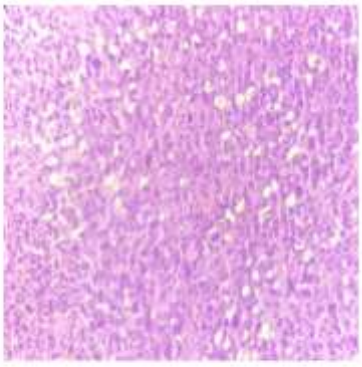

Figure 3: H & E stained section showing spingle shaped tumor cells with mitosis. (400X)

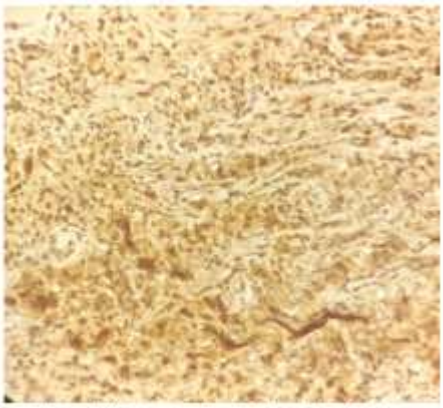

Figure 4: IHC cytoplasmic positivity for DOG1(100x)

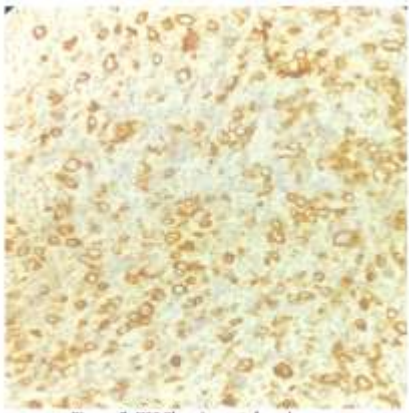

Figure 5: IHC Showing cytplasmic VIMENTIN positivity

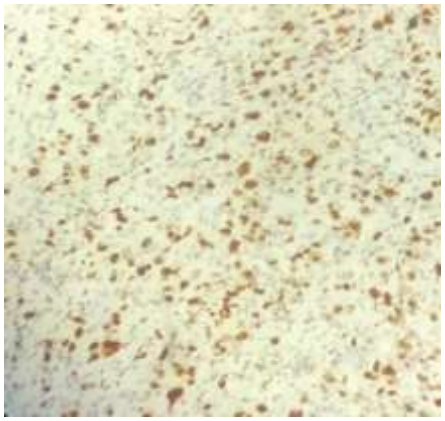

Figure 6: IHC positivity showing Ki67 25-30%

DISCUSSION

Esophageal GISTs are indeed rare tumors that typically affect middle to late-age males and are commonly located in the distal esophagus. They often present with symptoms such as dysphagia and odynophagia Distinguishing esophageal GIST from leiomyoma, the most common esophageal mesenchymal tumor, prior to resection is indeed challenging due to their similar appearances on imaging modalities such as MDCT, endoscopic ultrasound, and 18-fluorodeoxy glucose positron emission tomography (FDG-PET). While fine needle aspiration biopsy under endoscopic ultrasound can provide a definitive diagnosis, it is often avoided due to the risk of scarring and potential tumor dissemination [7].

Diagnosing esophageal mesenchymal tumors, such as gastrointestinal stromal tumors (GISTs), can be challenging due to their rarity and histological variability. GIST histology can range from relatively benign-looking spindle cell tumors to large sarcomatous tumors with high mitotic activity. Misdiagnosis can occur when further immunohistochemical (IHC) or molecular testing is not performed after identifying myogenic markers like SMA or desmin. Both gastrointestinal stromal tumors (GISTs) and leiomyomas typically appear hypoechoic on endoscopic ultrasound imaging, and they originate from the muscular layers of the gastrointestinal tract, specifically the muscularis propria for GISTs and the muscularis mucosae for leiomyomas. This similarity in appearance can make it challenging to differentiate between the two solely based on ultrasound findings. However, lipomas, another type of mesenchymal tumor, typically have a characteristic hyperechoic appearance, which can aid in their differentiation from GISTs and leiomyomas on endoscopic ultrasound [9].

Immunohistochemical panels, including markers like KIT (CD117), DOG1, CD34, smooth muscle actin, desmin, and S100, play a crucial role in distinguishing GIST from other tumors [10]. Additionally, prognostic factors such as tumor size, mitotic counts per 50 high-power fields (HPF), and anatomical location are significant in assessing the severity and potential outcomes of GIST.

Therefore, a comprehensive approach involving clinical, radiological, and immunohistochemical evaluations is necessary for accurate diagnosis and appropriate management of esophageal mesenchymal tumors, especially GISTs.

CONCLUSSION

Making a definitive clinical and pathological diagnosis is crucial for determining the appropriate treatment approach.For small tumors, tumor enucleation may be an option, while larger GISTs or those with a high mitotic rate may necessitate esophagectomy. The use of adjuvant imatinib therapy serves several purposes, including downsizing the GIST to reduce the extent of resection and decrease the risk of intraoperative complications, such as tumor rupture.

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