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Esophageal Gastrointestinal Stromal Tumor: A Rare Case Report

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Abstract **Case Report**

Esophageal gastrointestinal stromal tumor (GIST) is an exceptionally rare entity. We report the case of a 60-year-old patient investigated for solid-food dysphagia, in whom the diagnostic workup revealed a large mass involving the lower third of the esophagus with metastatic disease at presentation. The diagnosis was established through histopathological and immunohistochemical examination, demonstrating a mesenchymal proliferation positive for CD117, DOG1, and CD34, consistent with an esophageal GIST. Owing to the advanced stage of the disease, treatment with imatinib was initiated. Through this case, we highlight the rarity of this location, the diagnostic value of immunohistochemistry, and the essential role of targeted therapy in advanced forms.

Keywords: Esophageal GIST; dysphagia; immunohistochemistry; imatinib.

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INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are the most frequent mesenchymal tumors of the gastrointestinal tract and are thought to arise from, or share differentiation with, the interstitial cells of Cajal. Their diagnosis is based on the integration of morphology and immunohistochemistry, most notably CD117, DOG1, and CD34 expression, with molecular testing for KIT or PDGFRA mutations whenever available [1,2]. Most GISTs occur in the stomach or small intestine, whereas the esophagus is an exceptional location, accounting for less than 1% of all GISTs in published series [3,4].

Esophageal GISTs are clinically challenging because symptoms are often nonspecific and may mimic more common submucosal lesions, particularly leiomyoma. Dysphagia, retrosternal discomfort, bleeding, or weight loss can occur when tumors become large, and metastatic disease may be present at diagnosis in aggressive forms [5,6]. Cross-sectional imaging helps assess local extension and metastatic spread, while endoscopic evaluation and tissue sampling are required for definitive diagnosis. We report the case of an esophageal GIST presenting with dysphagia and diagnosed at a metastatic stage at initial presentation, illustrating the potential severity of this rare localization and the need for multidisciplinary management.

CASE PRESENTATION

We report the case of a 60-year-old patient with no significant past medical history, who had been experiencing solid-food dysphagia for one year, without any other associated symptoms. On clinical examination, the patient was afebrile and hemodynamically stable. Abdominal examination revealed a soft, non-tender abdomen, with no palpable mass. Laboratory investigations showed no significant abnormalities.

A thoraco-abdomino-pelvic computed tomography scan revealed a large, well-circumscribed posterior-inferior mediastinal mass measuring 13 × 9 × 13.5 cm and displacing the thoracic esophagus. The same examination showed secondary metastatic localizations, including peritoneal nodules in Morrison's pouch, multiple hepatic nodules, bilateral adrenal involvement, and deep lymphadenopathy (Figure 1).

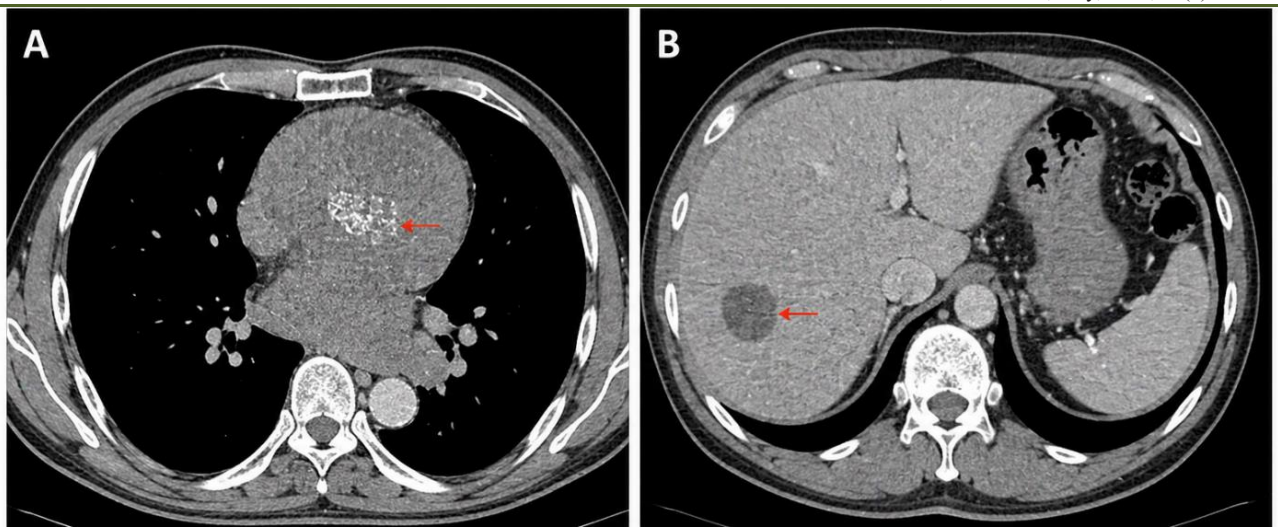

Figure 1: Thoraco-abdomino-pelvic computed tomography scan. (A) Axial thoracic section showing a large posterior-inferior mediastinal mass arising from the esophagus with central calcification (red arrow), displacing the thoracic esophagus. (B) Axial upper abdominal section showing hepatic metastatic involvement (red arrow). Patient-identifying information has been removed

An upper gastrointestinal endoscopy was performed, revealing a large submucosal-appearing mass, highly vascularized and friable, extending above the gastroesophageal junction from 40 cm to 30 cm from the dental arches, over an approximate length of 10 cm (Figure 2).

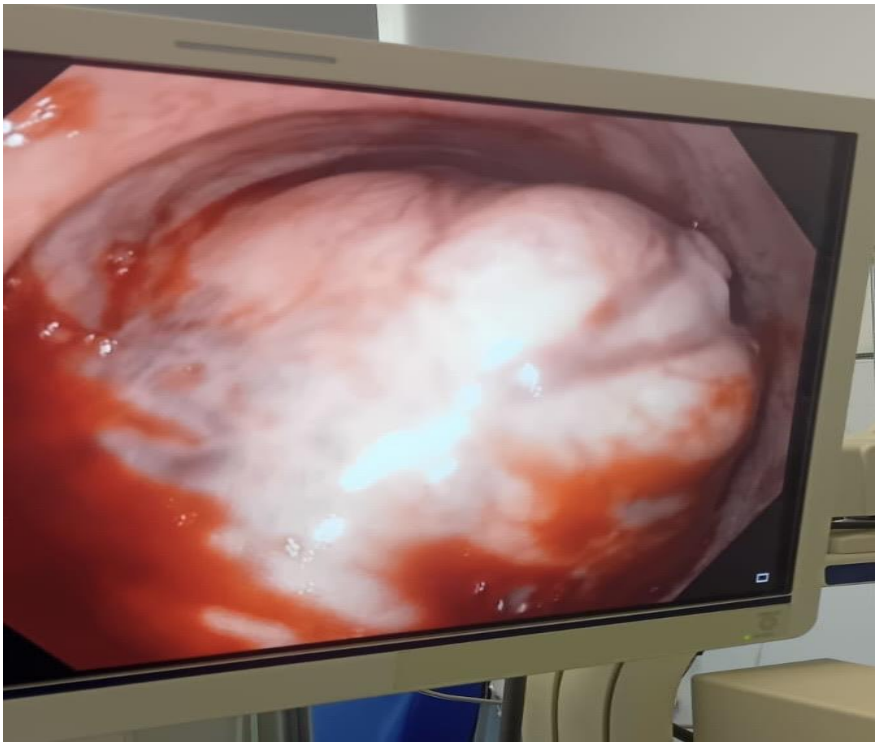

Figure 2: Large submucosal-appearing mass involving the lower third of the esophagus on endoscopic examination

Histological examination revealed a mesenchymal tumor proliferation with fascicular architecture. The tumor cells were spindle-shaped and occasionally epithelioid, with regular nuclei. Mitotic activity was moderate, and no tumor necrosis was observed. Immunohistochemical analysis demonstrated positivity for CD117, DOG1, and CD34. These findings were consistent with a gastrointestinal stromal tumor (Figure 3).

After oncological assessment, treatment with imatinib was initiated at a dose of 400 mg/day.

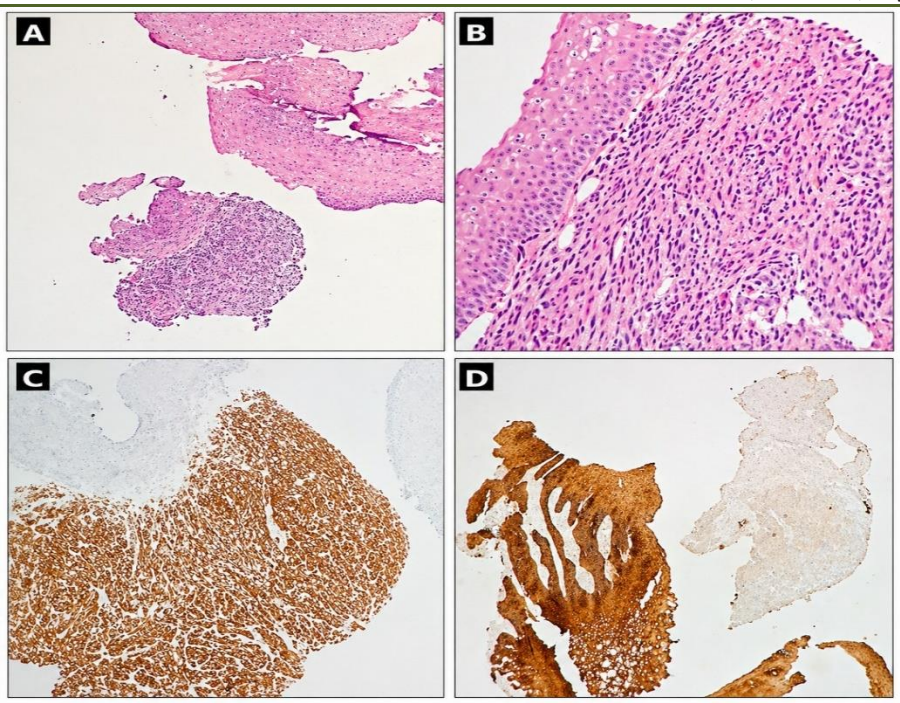

Figure 3: Histopathological and immunohistochemical findings. (A) Low-power H&E section showing a well-circumscribed submucosal spindle-cell proliferation. (B) Intermediate-power H&E view demonstrating fascicular proliferation of spindle cells with elongated nuclei. (C) Immunohistochemical staining showing diffuse strong cytoplasmic positivity for c-KIT (CD117). (D) Additional immunohistochemical profile excluding epithelial differentiation and supporting the diagnosis of esophageal gastrointestinal stromal tumor

The expected therapeutic result was symptom control and tumor stabilization under first-line targeted therapy. At the time of manuscript preparation, the obtained result was initiation of imatinib with planned clinical and radiological reassessment; objective radiological response could not yet be evaluated because follow-up imaging was not available.

DISCUSSION

Gastrointestinal stromal tumor can occur at any age, with a peak incidence between 50 and 60 years. Regarding esophageal GISTs, the median age at diagnosis is estimated to be 63 years [3].

GISTs most commonly arise in the stomach (60–70%) and small intestine (25–30%). Colorectal and extra-gastrointestinal forms are rare, while esophageal involvement remains exceptional, accounting for less than 1% of cases [4].

The clinical presentation is variable and often nonspecific. The most common symptoms include dysphagia, gastrointestinal bleeding, and abdominal pain. Approximately 20% of cases are incidentally discovered during endoscopic or radiological examinations, or during surgery performed for other indications [5].

Differentiating GIST from leiomyoma can be challenging. None of the currently available imaging modalities, including CT scan, endoscopic ultrasound, or FDG-PET, provide sufficient specificity to establish a definitive distinction [6]. Biopsy, once debated due to the presumed risk of tumor seeding, is now considered an essential step in the pre-therapeutic evaluation of GISTs [5].

Currently, the European Society for Medical Oncology (ESMO) recommends the use of CD117 and DOG1 as key diagnostic markers. However, approximately 5% of GISTs are negative for CD117 [7].

In our case, dysphagia revealed a large esophageal tumor that was already metastatic at the time of diagnosis, highlighting the potentially aggressive behavior of this localization. The diagnosis was confirmed by histopathological and immunohistochemical analysis, demonstrating positivity for CD117, DOG1, and CD34.

From a morphological standpoint, Gastrointestinal stromal tumor are most commonly composed of spindle cells, while the epithelioid component, although less frequent, is also well recognized. The coexistence of both components in our case illustrates the histological heterogeneity of these tumors. The originality of our observation lies in the large size and the initially metastatic presentation of this esophageal GIST, associated with a mixed spindle and epithelioid histological pattern.

Surgery is reserved for localized forms. Enucleation of esophageal GISTs is recommended for small tumors, generally between 2 and 5 cm [8], whereas esophagectomy is more often indicated for large lesions, particularly those exceeding 9 cm [9]. Complete en bloc resection is essential due to the malignant potential of these tumors, and intraoperative tumor rupture must be strictly avoided.

In locally advanced unresectable or metastatic disease, Imatinib is the standard first-line treatment at a dose of 400 mg/day. In the presence of a KIT exon 9 mutation, a higher dose of 800 mg/day is recommended due to improved progression-free survival. In cases of progression on imatinib or intolerance, second-line treatment is based on sunitinib [7]. However, the lack of detailed follow-up data under treatment in this case limits the assessment of therapeutic response.

The prognosis of esophageal Gastrointestinal stromal tumor depends mainly on tumor size and mitotic index, both of which are associated with more aggressive behavior. Esophageal GISTs therefore appear to have a poorer prognosis compared with gastric GISTs [2].

Their management should be discussed in a multidisciplinary setting, ideally within referral centers specialized in sarcomas and GISTs [10].

CONCLUSION

Esophageal gastrointestinal stromal tumors represent an exceptionally rare location and often present with nonspecific clinical features, which may delay diagnosis. This case highlights the importance of a multidisciplinary approach combining imaging, endoscopy, histopathological examination, and immunohistochemistry to establish an accurate diagnosis and differentiate this entity from other esophageal mesenchymal tumors. It also emphasizes the central role of staging assessment in guiding therapeutic decisions and determining prognosis. Because of their rarity, these tumors remain both a diagnostic and therapeutic challenge, requiring individualized management in experienced centers. Reporting additional cases remains essential to improve understanding of this uncommon presentation, refine management strategies, and increase clinician awareness of this rare disease.

REFERENCES

1. Costa F, Casaca R, Monteiro C, Ramos P. Oesophageal GIST. BMJ Case Rep. 2020;13(10): e238058. doi:10.1136/bcr-2020-238058.
2. Lott S, Schmieder M, Mayer B, Henne-Bruns D, Knippschild U, Agaimy A, *et al.,* Gastrointestinal stromal tumors of the esophagus: evaluation of a pooled case series regarding clinicopathological features and clinical outcome. Am J Cancer Res. 2015;5(1):333-343.
3. Bouggar M. Tumeurs stromales (GIST) de l'œsophage thoracique: à propos de 02 cas [thèse de médecine]. Rabat: Université Mohammed V, Faculté de Médecine et de Pharmacie; 2018.
4. Robb WB, Bruyere E, Amielh D, Vinatier E, Mabrut JY, Perniceni T, *et al.,* Esophageal gastrointestinal stromal tumor: is tumoral enucleation a viable therapeutic option? Ann Surg. 2015;261(1):117-124. doi:10.1097/SLA.0000000000000505.
5. Nemeth K, Williams C, Rashid M, Robinson M, Rasheed A. Oesophageal GIST—A rare breed case report and review of the literature. Int J Surg Case Rep. 2015; 10:256-259. doi: 10.1016/j.ijscr.2015.02.023.
6. Zaiem A, Ben Mahmoud A, Sebai A, Atri S, Daghfous A, Kacem JM. Diagnosis and surgical management of a rare esophageal GIST: A case report and review of literature. Int J Surg Case Rep. 2025; 134:111664. doi: 10.1016/j.ijscr.2025.111664.
7. Casali PG, Blay JY, Abecassis N, Bajpai J, Bauer S, Biagini R, *et al.,* Gastrointestinal stromal tumours: ESMO–EURACAN–GENTURIS Clinical Practice Guidelines for diagnosis, treatment and follow-up. Ann Oncol. 2022;33(1):20-33. doi: 10.1016/j.annonc.2021.09.005.
8. Yamamoto H, Ebihara Y, Tanaka K, Matsui A, Nakanishi Y, Asano T, *et al.,* Robot-assisted thoracoscopic esophagectomy for gastrointestinal stromal tumor of the esophagus: A case report. Int J Surg Case Rep. 2021; 86:106335. doi: 10.1016/j.ijscr.2021.106335.
9. Vogli S, Papadakos SP, Argyrou A, Schizas D. Expanding the role of endoscopic resection in esophageal gastrointestinal stromal tumors: insights and challenges. World J Gastroenterol. 2025;31(20):106441. doi:10.3748/wjg. v31.i20.106441.
10. Nakano A, Akutsu Y, Shuto K, Uesato M, Kono T, Hoshino I, *et al.,* Giant esophageal gastrointestinal stromal tumor: report of a case. Surg Today. 2015;45(2):247-252. doi:10.1007/s00595-014-0929-x.