

Case Report
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Gastrointestinal Stromal Tumor of Esophageal Location: A Rare Case with Surgical Discussion. Case Report and Literature Review

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms. They arise from the intestinal wall and usually present as subepithelial neoplasms in the stomach and small intestine; however, they can appear anywhere in the gastrointestinal (GI) tract. Most of these tumors have mutations in KIT or platelet-derived growth receptor alpha (PDGFRA), while mutations in succinate dehydrogenase (SDH) or other genes are less frequent. New molecules have shown promising results in the therapy of these tumors.

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Introduction

Gastrointestinal stromal tumor (GIST) is a rare tumor that accounts for about 3% of all tumors of the gastrointestinal (GI) tract [1]. Gastric location corresponds to 50 to 60% of cases, midgut to 30 to 35% and other sites of gastrointestinal tract are less common, such as colorectal and esophagus [2]. GISTs likely originate from the interstitial cells of Cajal (ICCs), sometimes referred to as the GI pacemaker cell and represent a distinct entity from other mesenchymal tumors of the GI tract [3]. The development of effective therapies is focused on the molecular profile of these tumors, where the KIT proto-oncogene mutation is the most common and on which therapies with better results have been sought [4].

Case Report

Female, 69 years old, with dyspeptic symptoms such as nausea and vomiting for 1 year. In upper digestive endoscopy, nodulation was visualized without alteration of the mucosa, not biopsied (Figure 1). During the period, she was diagnosed of pulmonary infection secondary to COVID and a chest X-ray identified an opacity in the power mediastinum (Figure 2). Chest CT scan showed heterogeneous expansive formation in the posterior mediastinum, located in the distal esophageal wall/esophageal-gastric transition, indeterminate, admitting among the differential diagnoses inflammatory pseudotumor and neoplasm, including benign lesions (leiomyoma) or malignant (GIST, for example).

In endoscopic ultrasound (EUS), a hypoechoic formation was seen involving distal esophagus with a predominant exophytic component. Diagnostic puncture with pathological report showed spindle cell proliferation without significant atypia, suggestive of GIST versus leiomyoma; then underwent distal esophagectomy.



Figure 1: Chest X-ray showing enlargement in the superior mediastinum

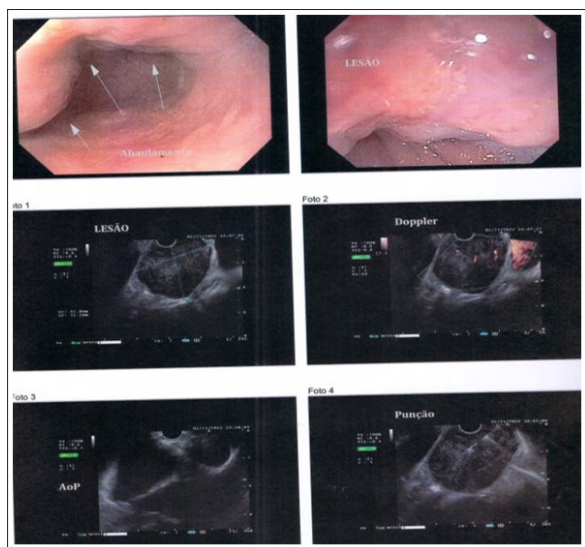


Figure 2: Upper digestive endoscopy image showing bulging mucous due to submucosal growth. At ultrasound (images below), evidence of submucosal expansive lesion

Pathological report confirmed spindle cell epithelioid neoplasia, measuring 6.0 x 4.0 x 3.5 centimeters (fragmented piece), mitotic count 9 mitoses per 5 mm, high cellularity, moderate cell pleomorphism, present necrosis at 15%, grade 2, positive angiolymphatic and perineural invasion, free margins, lymph node metastasis in 4/6 TEG lymph nodes (largest deposit measuring 10 mm), in addition to 2 nodules in the submucosa of the esophagus both measuring 7 mm. Immunohistochemistry was consistent with GIST due CD 117, DOG-1 and CD34 positive, S100 negative (Figure 3). Pathological staging pT3 pN1, stage IV.



Hospital discharge after 10 days of hospitalization with good evolution. The molecular profile of the GIST demonstrated positive cKIT mutation in Exon 11. PET-CT scan showed uptake in liver segments V/VI (SUV 3.3). Magnetic resonance imaging (MRI) of the abdomen confirmed multiple small liver lesions suggestive of secondary involvement [5-16]. Started Imatinib 400mg/day as a first line systemic treatment for metastatic GIST.

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