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CASE REPORT | ESOPHAGUS

Gastrointestinal Stromal Tumor Presenting as Food Impaction and Pseudo-Achalasia

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are one of the most common mesenchymal tumors of the gastrointestinal tract. Studies report the incidence of GIST to be 14.5 per million, with 18% being diagnosed incidentally. The most common location is the stomach while the esophagus is the rarest, representing only 0.7% of cases. The clinical manifestations of GISTs vary. Most patients present with bleeding and gastric discomfort. Some may present with life-threatening hematemesis or melena. We present a unique case of a GIST presenting as pseudo-achalasia and food impaction.

KEYWORDS: esophageal impaction; pseudoachalasia; stromal tumors

INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are, in general, rare tumors but some of the most common mesenchymal tumors of the gastrointestinal tract. One study in Sweden estimated the incidence of GIST to be approximately 14.5 per million.[1] One systematic review estimates that 18% of patients had an incidental diagnosis of GIST, with the location of the primary tumor varying significantly. The stomach is the most common (55.6%) location of GISTs while the esophagus is least common (0.7%).[2] The clinical manifestations of a GIST vary. Up to 19% of patients are asymptomatic on presentation. The most common presenting symptoms are abdominal pain, intestinal obstruction, and GI bleeding.[2] The prognosis of GIST in patients depends on a variety of factors. The most widely accepted prognostic factors by multiple guidelines are mitotic rate, tumor size, tumor site, and rupture. Other prognostic factors, such as depth of invasion and histological type, have been used less often.[3] There are multiple therapeutic options for GIST depending on the prognostic factors. The standard of care for patients without metastases is surgical resection. For the patients with metastases or who are poor surgical candidates, the standard of care is treatment with a tyrosine kinase inhibitor such as imatinib.[4] We present a unique case of a GIST presenting as pseudo-achalasia.

CASE REPORT

An 87-year-old White man with medical history significant for atrial fibrillation and diabetes presented with respiratory failure. The patient was eating a chicken dinner when he felt food got stuck in his chest. This had never happened to him before, and he had no history of dysphagia and food impaction. He was unable to tolerate solids or liquids after he presented to the emergency department. He underwent an emergent endoscopy that was aborted because of resistance and being unable to retrieve the food bolus. He remained intubated after the first endoscopy strictly for airway protection.

He had 2 further endoscopies that were unsuccessful because of persistent resistance and were only notable for pools of old blood and clots in the esophagus. After the third upper endoscopy, the advanced therapeutics gastroenterology team was consulted and performed another upper endoscopy. This time, upper endoscopy showed no evidence of frank food impaction, but showed concern for mucosal trauma and esophageal diverticula concerning for achalasia vs an external mass effect (Figure 1). Because no food impaction was visualized, the decision was made to extubate the patient, which was performed without incident.

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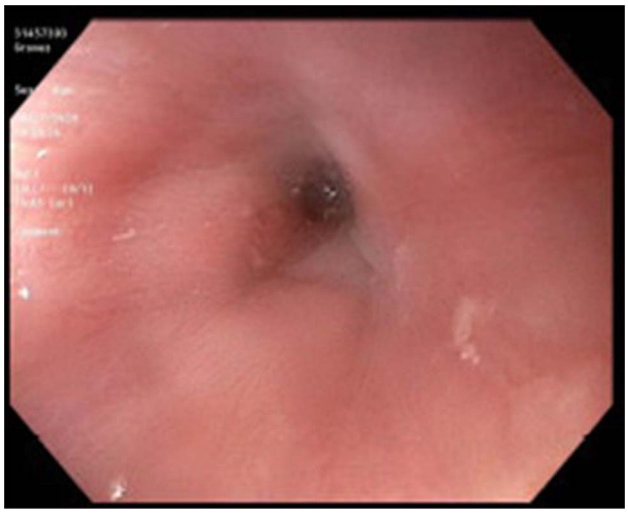

Figure 1. Upper endoscopy showing stenosis with suspected extrinsic compression at a level of 37 cm from incisors.

To evaluate for the possible mass effect, a computed tomography (CT) scan of the chest, abdomen, and pelvis with contrast showed a 3.8 × 3.1 cm low-attenuation structure in the distal esophagus, approximately 5 cm proximal to the gastroesophageal junction imparting mass effect upon the esophageal lumen with severe luminal stenosis. The differential provided by the radiology team included esophageal duplication cyst, distal esophageal mass including a leiomyoma, or fluid-filled diverticulum. Of note, no mass was visualized in the abdomen or pelvis.

The patient subsequently underwent a water-soluble single contrast esophagram, which showed multiple suspected strictures and concern for a mass effect in the distal esophagus. Overall findings were less suspicious of true achalasia. He subsequently underwent an endoscopic ultrasound of the esophagus, which showed a well-circumscribed mass at the level of 37 cm from the incisors concerning for GIST, and a fine-needle aspiration was performed for pathology evaluation (Figure 2).

The aspiration cytology confirmed a spindle cell tumor with positive CD117, CD34, and smooth muscle actin immunostains, consistent with GIST.

The patient was subsequently discharged from the hospital with close oncology follow-up for further management. An outpatient 18-FDG-PET/CT was performed, which redemonstrated the 3.8 cm esophageal mass and showed a nonenlarged hypermetabolic lymph node anterior to the proximal superior vena cava concerning for a reactive or metastatic lymph node. No other concerning foci of hypermetabolism were identified.

On thoracic surgery evaluation, he was deemed a poor surgical candidate because of poor functional status; thus, the patient agreed to initiate treatment with imatinib.

DISCUSSION

Pseudo-achalasia is a clinical entity that presents with progressive dysphagia to solids and liquids and is usually secondary to extrinsic compression secondary to a gastroesophageal junction mass, as opposed to primary denervation in the case of true achalasia. A systematic review reported that the most common cause of pseudo-achalasia is because of compression of the esophagus secondary to gastric adenocarcinoma.[5] The nonmalignant causes of pseudo-achalasia were reported to be sarcoidosis, amyloidosis, and Chagas disease.[5] It is important to note the patient's history when comparing pseudo-achalasia with achalasia, in which patients with pseudo-achalasia have very similar presentations but were found to have a shorter duration of dysphagia. A study found that both groups have difficulty with endoscopic procedures passing through the gastroesophageal junction and that CT scans are limited in their ability to diagnose many of the compressive tumors.[6] Diagnostic modalities, such as manometry, barium esophagram, endoscopy with numerous biopsies, and CT scans, should be used judiciously and in a complementary manner to achieve a diagnosis.

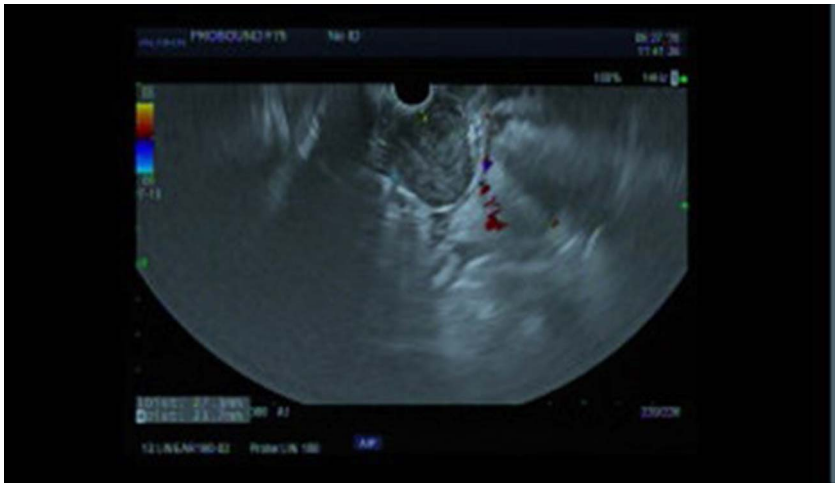

Figure 2. Endoscopic ultrasound showing well-circumscribed homogeneous mass at the level of stenosis (37 cm from incisors).

Numerous other causes of pseudo-achalasia have been reported in the literature, but on review, no cases of GIST pseudo-achalasia with acute dysphagia were found. One case report noted a patient presenting with an esophageal perforation, which was later diagnosed as being caused by a GIST.[7] Another study commented on a lower esophageal stromal tumor that presented with a 10-year progressive history of dysphagia that was later diagnosed as a stromal tumor.[8] This highlights the variable presentations previously discussed of GIST and the importance of a thorough history and evaluation for causes of dysphagia. As discussed, dysphagia can be a multifactorial disease and must be diagnosed with a thorough history and multidisciplinary evaluation.

Physicians should have several differentials when a patient with suspected pseudo-achalasia and food impaction presents. After stabilization of the patient and protection of the airway, workup and evaluation should include common and rare causes for food impaction. Although generally uncommon, and even rarer in the esophagus, GIST should be considered in the differential diagnosis of any gastrointestinal tract malignancy. Prompt diagnosis by tissue sampling from fine-needle aspiration is crucial to ensure expedited treatment. This is particularly important in cases such as this where symptoms significantly affect quality of life and may be life-threatening.

DISCLOSURES

Author contributions: MS Obri: lead author and researcher. F. Nimri: literature review and manuscript writing. W. Kamran: radiological review and literature review. R. Nimri: literature review and manuscript writing. R. Pompa: endoscopy performer, literature review, and supervising physician. T. Zuchelli: supervising author, endoscopy performer, and literature review. MS Obri is the article guarantor.

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REFERENCES

1. Nilsson B, Bümming P, Meis-Kindblom JM, et al. Gastrointestinal stromal tumors: The incidence, prevalence, clinical course, and prognostication in the preimatinib mesylate era: A population-based study in western Sweden. *Cancer.* 2005;103(4):821–9.
2. Søreide K, Sandvik OM, Søreide JA, Giljaca V, Jureckova A, Bulusu VR. Global epidemiology of gastrointestinal stromal tumours (GIST): A systematic review of population-based cohort studies. *Cancer Epidemiol.* 2016; 40:39–46.
3. Zhang H, Liu Q. Prognostic indicators for gastrointestinal stromal tumors: A review. *Transl Oncol.* 2020;13(10):100812.
4. Akahoshi K, Oya M, Koga T, Shiratsuchi Y. Current clinical management of gastrointestinal stromal tumor. *World J Gastroenterol.* 2018;24(26):2806–17.
5. Schizas D, Theochari NA, Katsaros I, et al. Pseudoachalasia: A systematic review of the literature. *Esophagus.* 2020;17(3):216–22.
6. Tracey JP, Traube M. Difficulties in the diagnosis of pseudoachalasia. *Am J Gastroenterol.* 1994;89(11):2014–8.
7. Sjogren PP, Banerji N, Batts KP, Graczyk MJ, Dunn DH. Rare presentation of a gastrointestinal stromal tumor with spontaneous esophageal perforation: A case report. *Int J Surg Case Rep.* 2013;4(7):636–9.
8. Mohamed A. Education and imaging. Gastrointestinal: Pseudoachalasia caused by a lower esophageal stromal tumor. *J Gastroenterol Hepatol.* 2009; 24(6):1152.