Squamous Cell Carcinoma of the Gastroesophageal Junction as an Unusual Underlying Cause of Pseudoachalasia

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Introduction

Achalasia is a rare esophageal motility disorder characterized by progressive dysphagia to solids and eventually liquids.[1-3] Pseudoachalasia, termed by Ogilvie in 1947, refers to an achalasia secondary to an underlying etiology, such as masses, post-operative complications, or paraneoplastic syndromes.[1,3,4] The estimated annual incidence rate is 1 in 100,000 individuals.[1,2] Pseudoachalasia arising from malignant neoplasms make up to 4% of achalasia-like syndromes.[1-3,5] We discuss a case that deviates from the typical etiology of esophageal cancer, emphasizing the need for a comprehensive workup for dysphagia including radiology, high resolution manometry, and upper endoscopy.

Case Presentation

A 71-year-old male with PMH for deep vein thrombosis due to Factor V Leiden deficiency, on anticoagulation with rivaroxaban, initially presented with intermittent dysphagia to solid food for many years, rapidly progressive to both solids and liquids over the month preceding presentation. No reported history of tobacco use, drank 6 to 8 alcoholic beverages daily until limiting to social drinking 3 years prior. No family history of gastrointestinal malignancy. Initial barium esophagram evaluation revealed decreased esophageal peristalsis, distention, and smooth narrowing at the gastroesophageal junction with “bird beak” appearance consistent with possible achalasia.

On examination he was hemodynamically stable. His abdomen was soft and nontender, and there was no lymphadenopathy. Initial blood count and chemistries was unremarkable. The barium esophagram report contained findings consistent with achalasia. CT chest and abdomen revealed a 4cm x 4cm hypoattenuating infiltrative lesion in the gastroesophageal junction (GEJ) with loss of fat plane with serosal irregularity [Fig. 1a and 1b], and acute pulmonary embolus in the distal right pulmonary artery.

Esophagogastroduodenoscopy to evaluate for pseudoachalasia revealed severe narrowing at the GEJ [Fig. 2a] which could only be traversed after esophageal balloon dilation at 8-9-10 mm through the scope, at which point, a 4 cm submucosal mass was found at the GEJ extending into the gastric cardia; its epicenter was at 2-3 cm from the GEJ [Fig. 2b and 3]. Deep biopsies were performed and histopathology revealed invasive squamous cell carcinoma (SCC) with submucosal involvement. Endoscopic ultrasound revealed an intramural lesion at the gastric cardia with deep margins limited to the submucosal layer and one enlarged 14 mm x 6 mm lymph node in the subcarinal mediastinum; further imaging did not reveal pathologic lymphadenopathy. Biopsy of the lesion revealed SCC. Fine needle aspiration of the lymph node was negative for malignancy, leading to a diagnosis of T2N0M0 SCC of the GEJ with gastric cardia involvement.

Ongoing patient care is being managed by medical oncology, surgical oncology, and radiation oncology.

Figures

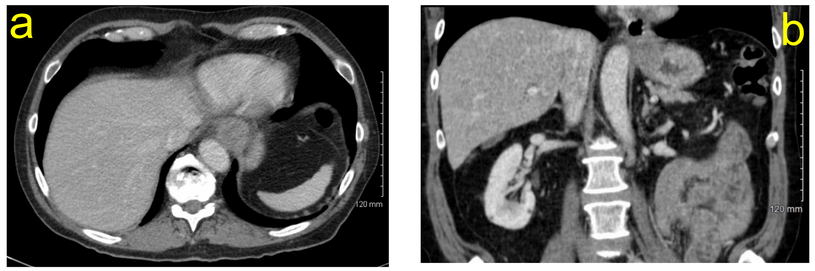

Figure 1: Computed tomography (CT) scan of the abdomen revealed hypoattenuating infiltrative lesion in the gastroesophageal junction (GEJ), axial image (a) and coronal (b)

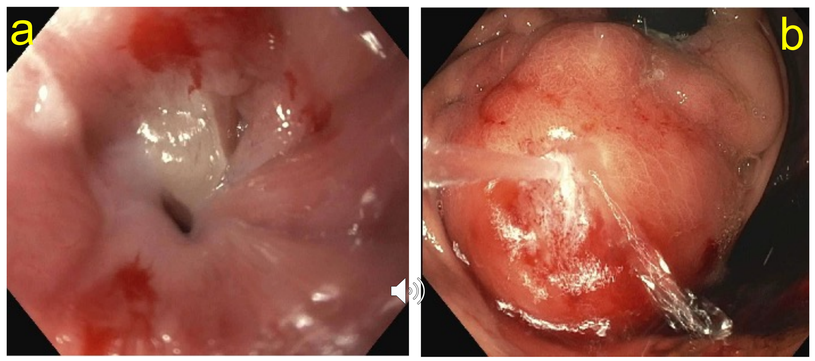

Figure 2: Esophagogastroduodenoscopy revealed severe stricture at the GEJ (a), a 4 cm submucosal mass at the GEJ with extension to the gastric cardia (b).

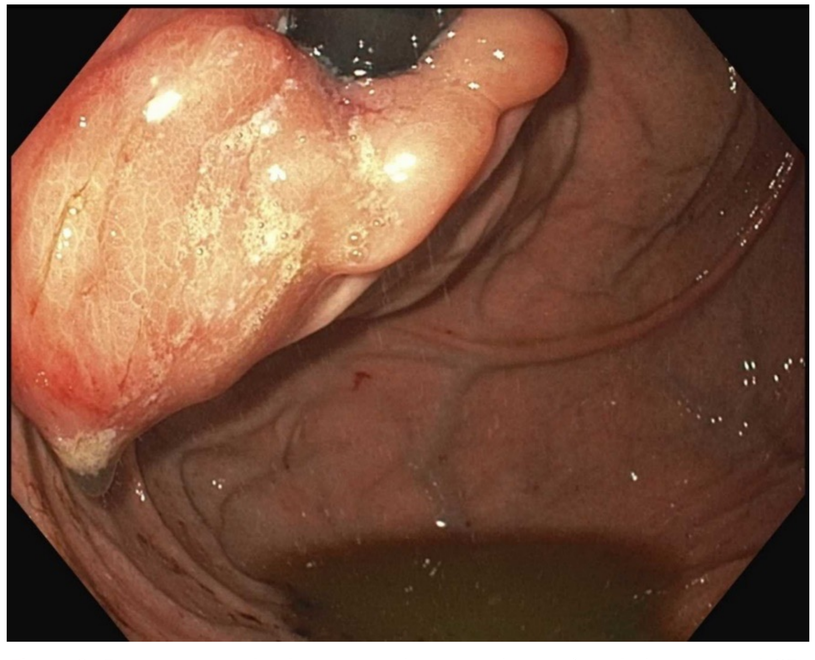

Figure 3: Esophagogastroduodenoscopy with a gastric retroflexed view revealed GEJ submucosal mass extension into the gastric cardia.

Discussion

Pseudoachalasia is often misdiagnosed for primary achalasia because of its rarity and indistinguishable symptomatology.[1,5] Patients present with dysphagia, regurgitation, chest pain, or weight loss.[1] Furthermore, imaging studies may also mimic primary achalasia.[1,3-5] In both cases, barium swallow shows narrowing of the distal esophagus, esophageal manometry shows decreased or absent distal peristalsis with impaired relaxation of the lower esophageal sphincter, and endoscopy reveals distal esophageal stenosis.[3] Simple mucosal biopsies from upper endoscopy have a 25% false negative rate, and therefore are not a reliable means of excluding pseudoachalasia.[1,3]

CT findings suggestive of pseudoachalasia over primary achalasia include asymmetric esophageal wall thickening or presence of a mass.[3,5] Endoscopic ultrasound can confirm pseudoachalasia by providing more detail of any abnormal tissue beneath the mucosa of esophageal and gastric wall layers.[3-5] Fine-needle aspiration can also be performed if endoscopic biopsies are inconclusive.[3]

Nearly 75% of pseudoachalasia cases are due to adenocarcinoma of the gastric cardia.[2-5] Esophageal squamous cell carcinoma (SCC) has an incidence rate of 1.2 per 100,000 individuals and is primarily found in the middle third of the esophagus.[6] SCC of the distal esophagus or GEJ as compared to the mid-esophagus is rare. Pseudoachalasia due to SCC of the distal esophagus has been reported once[7]; we present our patient as the first reported case of pseudoachalasia due to gastroesophageal junction squamous cell carcinoma.

Conclusion

Squamous cell carcinoma of the gastroesophageal junction is an extremely rare esophageal malignancy and has never previously been reported as an underlying cause of pseudoachalasia. Other cases have often misdiagnosed malignancy for primary achalasia in the setting of normal findings on CT, endoscopy, and/or biopsy.[1-3] This emphasizes the need for comprehensive workup to distinguish primary achalasia and pseudoachalasia to avoid overlooking a potential malignancy.

References

1. Campo SM, Zullo A, Scandavini CM, Frezza B, Cerro P, Balducci G. Pseudoachalasia: A peculiar case report and review of the literature. *World J Gastrointest Endosc*. 2013;5(9):450-454. doi:10.4253/wjge.v5.i9.450
2. Park JH, Park DI, Kim HJ, et al. An unusual case of submucosal invasion of esophageal squamous cell carcinoma mistaken as primary achalasia. *J Neurogastroenterol Motil*. 2010;16(2):194-198. doi:10.5056/jnm.2010.16.2.194
3. Roushan N, Zolfaghari A, Asadi M, Taslimi R. Pseudoachalasia: a diagnostic challenge. *Med J Islam Repub Iran*. 2014;28:54. Published 2014 Jul 12.
4. Robertson CS, Griffith CD, Atkinson M, Hardcastle JD. Pseudoachalasia of the cardia: a review. *J R Soc Med*. 1988;81(7):399-402.
5. Choi MK, Kim GH, Song GA, et al. Primary squamous cell carcinoma of the liver initially presenting with pseudoachalasia. *Gut Liver*. 2012;6(2):275-279. doi:10.5009/gnl.2012.6.2.275
6. Thrift, A. P. (2016). The epidemic of oesophageal carcinoma: Where are we now? *Cancer Epidemiology*, *41*, 88–95. https://doi.org/10.1016/j.canep.2016.01.013
7. Kahrilas, P., Kishk, S., Helm, J., Dodds, W., Harig, J. and Hogan, W., 1987. Comparison of pseudoachalasia and achalasia. *The American Journal of Medicine*, 82(3), pp.439-446.

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