The great mimicker; esophageal sarcoidosis masquerading as malignancy

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INTRODUCTION

BACKGROUND

Sarcoidosis is a chronic, multi-system disorder that most commonly affects the lungs. Luminal gastrointestinal involvement including the esophagus is rare, occurring in less than 2% of patients. We describe a case not previously reported of esophageal sarcoidosis presenting as a cratered, esophageal ulcer concerning for malignancy.

CASE PRESENTATION

A 43-year-old woman presented to clinic with three months of solid food dysphagia and epigastric pain. Her medical history was notable for laparoscopic Roux-en-Y gastric bypass surgery two years earlier.

Esophagogastroduodenoscopy (EGD) demonstrated a large, cratered ulcer with irregular borders in the mid esophagus concerning for malignancy (Figure A).The ulcer was deep with concern for an underlying fistula, and esophageal biopsies were deferred until further imaging was performed.

Esophagram showed a 3 cm ovoid filling defect in the anterolateral mid-thoracic esophagus with surrounding edema (Figure B). Computed tomography scan of the chest demonstrated a subcarinal soft tissue mass adjacent to the right esophageal wall measuring 21x27x47 mm, difficult to distinguish from surrounding posterior mediastinal lymph nodes. There was prominent mediastinal lymphadenopathy.

Esophagogastroduodenoscopy with endoscopic ultrasound showed a 17x40 mm non-circumferential, hypoechoic, and heterogeneous mass with invasion of the esophageal wall from the adventitia through the mucosa, which appeared consistent with a large lymph node (Figure C).

Pathology showed multiple non-necrotizing granulomas consistent with sarcoidosis (Figure D). The patient was referred to Rheumatology and due to prior intolerance to prednisone (severe psychosis), treated with mycophenolate mofetil and close follow-up in clinic.

FIGURE A

Image from esophagogastroduodenoscopy of a large, cratered ulcer (arrow) in the middle esophagus.

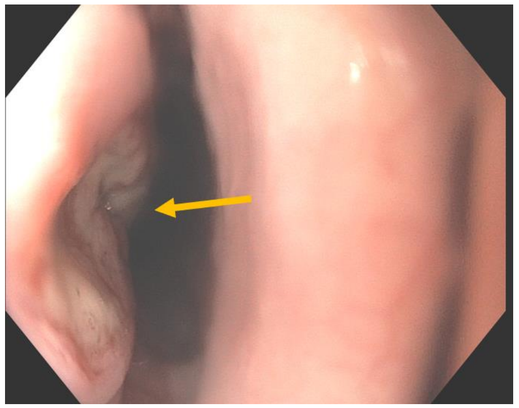


FIGURE B

Fluoroscopic images from esophagram demonstrating an irregular, ovoid shaped filling defect consistent with ulcer (arrow) in the mid esophagus.

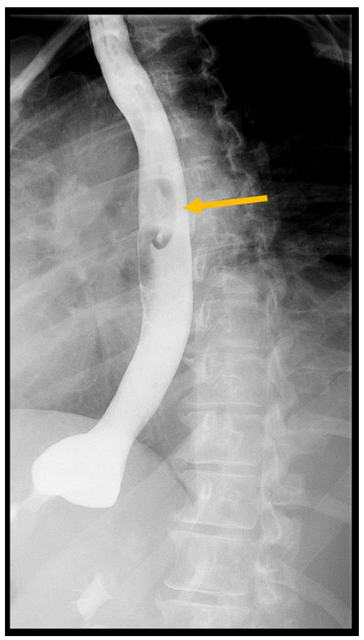


FIGURE C

Endosonographic image demonstrating a hypoechoic and heterogeneous mass (blue dotted lines) infiltrating the esophageal wall. The mass measured 17 mm x 40 mm in cross section.

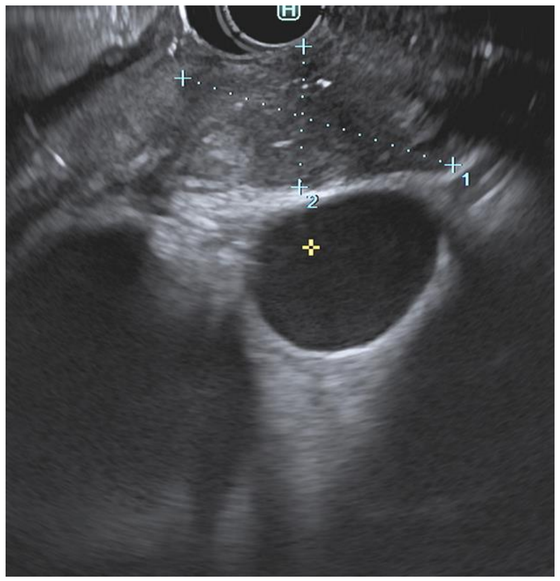
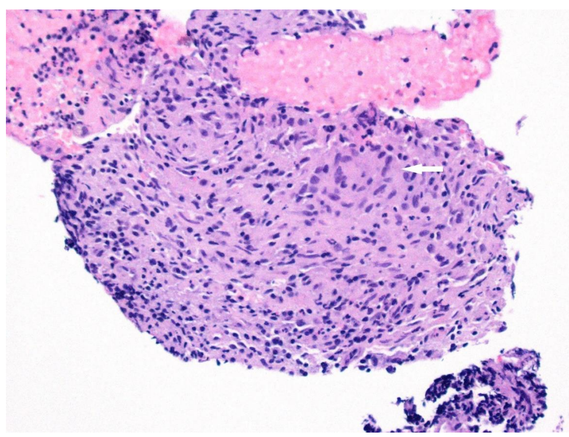


FIGURE C

Microscopic image of the cell block preparation shows a nonnecrotizing granuloma with a multinucleated giant cell (arrow). Hematoxylin and eosin stain; original magnification x 200.



DISCUSSION

Sarcoidosis is a condition encountered in clinical practice, and gastrointestinal involvement may be the first sign of this systemic disease in patients.

Esophageal sarcoidosis was first reported in 1948, and as of 2019 only 43 cases had been reported in the medical literature.

Mucosal infiltration leads to the formation of strictures or nodules, and myenteric involvement leads to malfunction of the affected esophagus.

Extrinsic compression may occur secondary to mediastinal lymphadenopathy and can result in a traction esophageal diverticulum or even perforation.

Rarely, an achalasia-like presentation can occur owing to direct involvement of the Auerbach's plexus requiring myotomy.

The most common presenting symptom is dysphagia, followed by weight loss. Other presenting symptoms include abdominal or chest pain, nausea, vomiting, heartburn/reflux, and odynophagia.

Given the rarity of esophageal sarcoidosis, there are no clinical trials evaluating different treatment modalities.

The mainstay of treatment is corticosteroids, and the duration of therapy depends on the clinical response.

The goal of therapy is to relieve symptoms and prevent the progression of disease and organ damage.

Steroid-sparing agents can be utilized in refractory cases or in patients with contraindications to corticosteroids such as this case presentation

Steroid-sparing agents include methotrexate, mycophenolate mofetil, azathioprine, infliximab, and cyclosporine.

Sarcoid should be considered in the differential diagnosis of patients with esophageal symptoms and atypical findings.