

Received: 2015.04.23
Accepted: 2015.06.24
Published: 2015.10.29

ISSN 1941-5923
© Am J Case Rep, 2015; 16: 768-773
DOI: 10.12659/AJCR.894444

Pseudoachalasia: Still a Tough Clinical Challenge

Department of Internal Medicine, Texas Tech University Health Sciences Center,
Paul L. Foster School of Medicine, El Paso, TX, U.S.A.

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

ABCDEF **Yi Jia**
ADEFG **Richard W. McCallum**





Corresponding Author: Richard W. McCallum, e-mail: richard.mccallum@ttuhsc.edu
Conflict of interest: None declared

Patient: Male, 81
Final Diagnosis: Pseudoachalasia
Symptoms: Dysphasia
Medication: —
Clinical Procedure: Endoscopy ultrasound
Specialty: Gastroenterology and Hepatology

Objective: Unusual clinical course
Background: Treatment of achalasia is focused on decreasing the resting lower esophageal sphincter by either pneumatic dilation or surgical myotomy. When patients symptomatically relapse after one or more pneumatic dilations, then one explanation is to consider the possibility of pseudoachalasia as the diagnosis.
Case Report: We present a rare case of an elderly patient with a presentation of chronic dysphagia and severe weight loss, who had diagnostic findings consistent with achalasia, and who also responded very well to a series of pneumatic dilations, but for only brief intervals. Further investigations finally uncovered esophageal adenocarcinoma, thus making our patient an example of the entity “pseudoachalasia”.
Conclusions: Pseudoachalasia secondary to an esophageal malignancy should be suspected when dysphagia progresses despite technically well-performed pneumatic dilations, and is particularly suspicious in the setting of an elderly patient with marked weight loss. Endoscopic ultrasound is a new diagnostic tool for detecting and staging malignancy by obtaining diagnostic tissue and allowing appropriate therapy to be planned.

MeSH Keywords: Endoscopy, Gastrointestinal • Esophageal Achalasia • Esophageal Neoplasms • Gastric Dilatation

Full-text PDF: <http://www.amjcaserep.com/abstract/index/idArt/894444>

 2176  —  4  11



Background

The patients with a presentation of dysphagia with or without noncardiac chest pain can have the diagnosis of achalasia. Treatment of achalasia can be accomplished by mechanical disruption of the muscle fibers of the lower esophageal sphincter (LES), by either pneumatic dilation (PD) or surgical myotomy. When patients symptomatically relapse after one or more PDs, then pseudoachalasia should be considered as another possibility diagnosis. Here, we present a rare case of a patient with a presentation of chronic dysphagia with an initial well documented diagnosis of achalasia and treated with PDs. However, further investigations were warranted in an elderly patient with marked weight loss when the PDs gave only a brief interval of relief. Esophageal adenocarcinoma was found and staged by Endoscopic ultrasound (EUS), thus making our patient an example of the entity “pseudoachalasia”.

Case Report

An 81-year-old Caucasian male was referred to the gastroenterology clinic with a 6 month history of progressive dysphagia. His referring physician had already made a diagnosis of achalasia and he had received a Botulinum toxin (Botox) injection into the LES through the endoscope about 3 weeks prior to his visit. He had responded well initially but then redeveloped dysphagia to both solids and liquids. He also has been having regurgitation of recently ingested food and in view of a more than 40 lbs weight loss history, he was admitted to hospital.

In reviewing his history, he had been experience dysphagia for solids and liquids for 4 years as well as regurgitation of food, particularly old food at night. On review of systems, patient denied fever, weight loss, chest pain, cough or dyspnea. He did not report any neurological or respiratory symptoms. He had no medications at home. Pertinent history included colon cancer with colectomy 6 years ago without other further chemoradiation therapy. He was an active smoker for the past 20 years but denied any alcohol or illicit drug use. Upon physical examination, the patient was afebrile with stable vital signs. He weighed 68 Kg with a BMI of 25. On abdominal exam, there was a lower midline surgical scar from the colectomy; his abdomen was soft, non-distended and non-tender, no masses or organomegaly were palpable. Bowel sounds were present and normal. Other exam findings were unremarkable. His laboratory results indicated a normal comprehensive metabolic panel, but his complete blood count showed mild anemia with hemoglobin 11.5 g/dL, hematocrit of 38.9%, and MCV of 81 fL, more indicative of an anemia of chronic disease.

Patient underwent a high resolution esophageal motility study which showed a classic achalasia pattern with resting LES

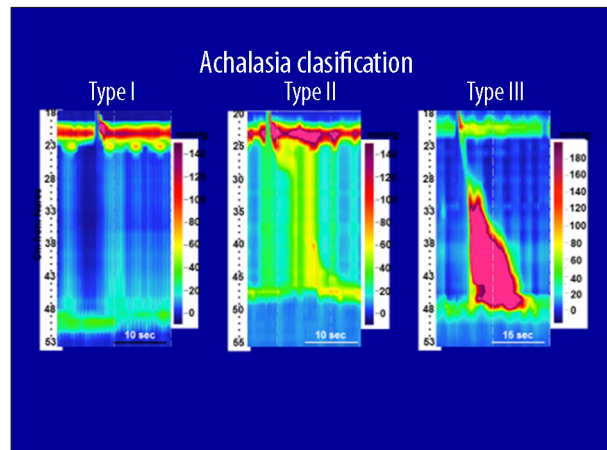


Figure 1. A motility study can distinguish achalasia from other motility disorders. The presence of aperistalsis in the distal two-thirds of the esophagus and incomplete LES relaxation on esophageal manometry, defined as a mean four-second integrated relaxation pressure (IRP) >15 mmHg are the manometric criteria required for the achalasia. Our patient's outpatient high-resolution manometry results indicated a Type 2 classification.

pressure of 36 mmHg (normal 10–40 mmHg), integrated relaxation pressure (IRP) >15 mmHg, inadequate (25%) relaxation of the LES with wet swallows; aperistalsis with wet swallows with low amplitude contractions (<30 mmHg), consistent with type 2 Chicago classification for achalasia (Figure 1). The upper esophageal sphincter and striated muscle function were intact. Esophagogastroduodenoscopy (EGD) found patchy candidiasis in the lower third of the esophagus and a dilated, atonic esophagus. A hypertonic lower esophageal sphincter was found with resistance to endoscopic passage into the stomach. The Z-line was irregular and located 37 cm from the incisors with a 3 cm hiatus hernia.

The therapy plan was to dilate the LES and improve esophageal emptying. Patient had pneumatic esophageal dilation using a Rigidflex 30 mm balloon dilator (90 French diameter). Post dilation, the esophagus was endoscopically examined and there was the usual mild mucosa trauma. An immediate post-dilation Gastrografin esophagram showed a long segment stricture of the distal esophagus with no evidence of leakage (Figure 2). No other complications were observed. He was able to tolerate a mechanical soft diet and discharged home.

Three weeks later, patient presented to the ER with a chief complaint of the return of dysphagia to solids and liquids with decreased oral intake and received a second esophageal dilation, this time with a 35 mm pneumatic balloon dilator. Patient tolerated the esophageal dilation well and was able to be advanced to a regular diet before being discharged home. During his outpatient follow up, a chest CT was obtained that showed concentric mural thickening of the mid and distal esophagus

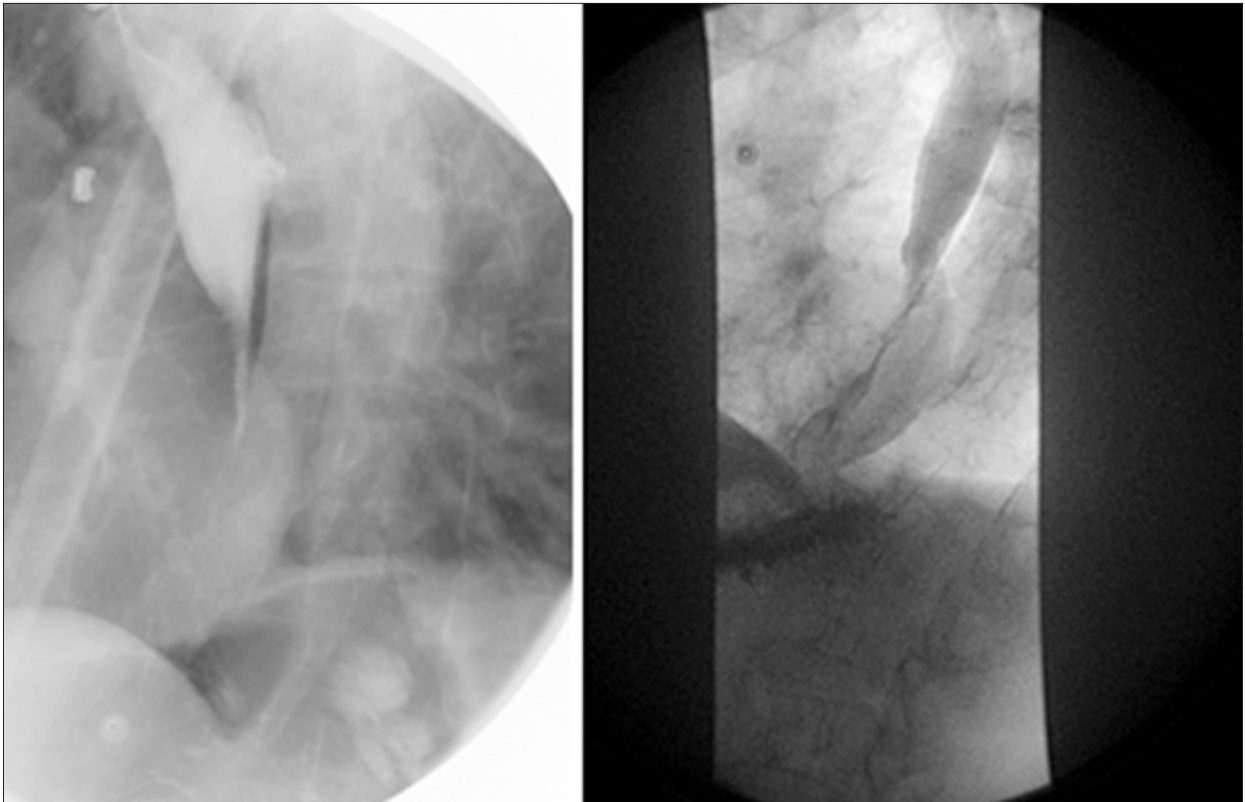


Figure 2. Gastrografin swallow of the esophagus immediately after pneumatic dilation showed long segment of stricture of the distal esophagus with suggestion of some irregularity without evidence of leakage.

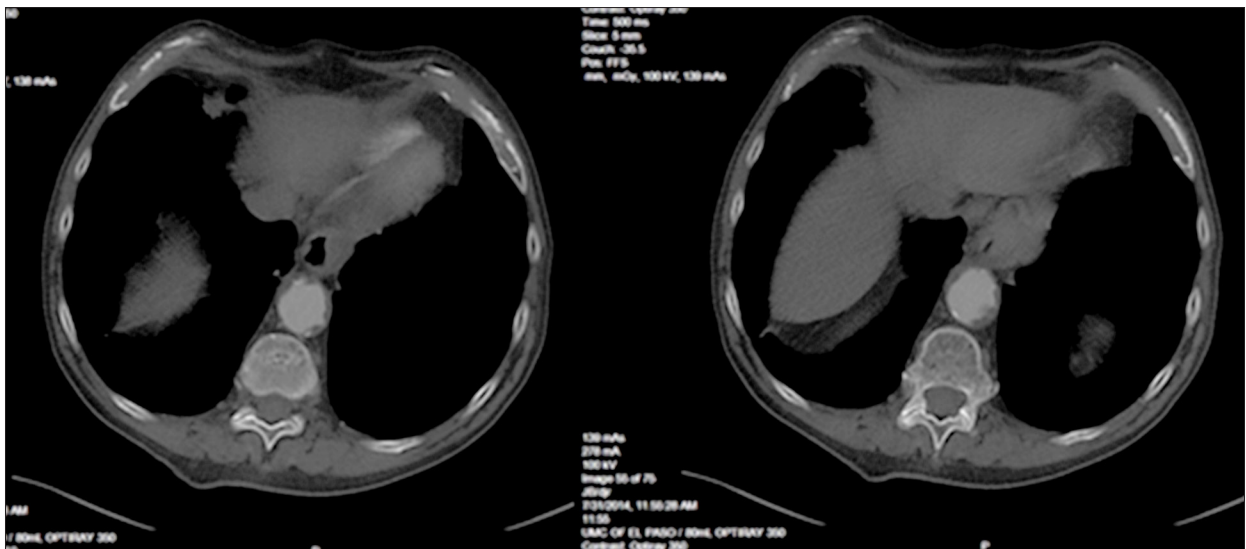


Figure 3. Follow up chest CT showed concentric mural thickening of the mid and distal esophagus which was out of proportion to that seen in a regular achalasia.

which was out of proportion to the usual length of “beaking” present at the lower esophageal sphincter in regular achalasia (Figure 3).

Six months later patient presented with return of dysphagia to both liquids and solids that had markedly progressed over the past month with about a 50 pounds weight loss over 3 months. Repeat EGD found moderately severe esophagitis, erosions, friability and a narrowing at 35 cm from the incisors

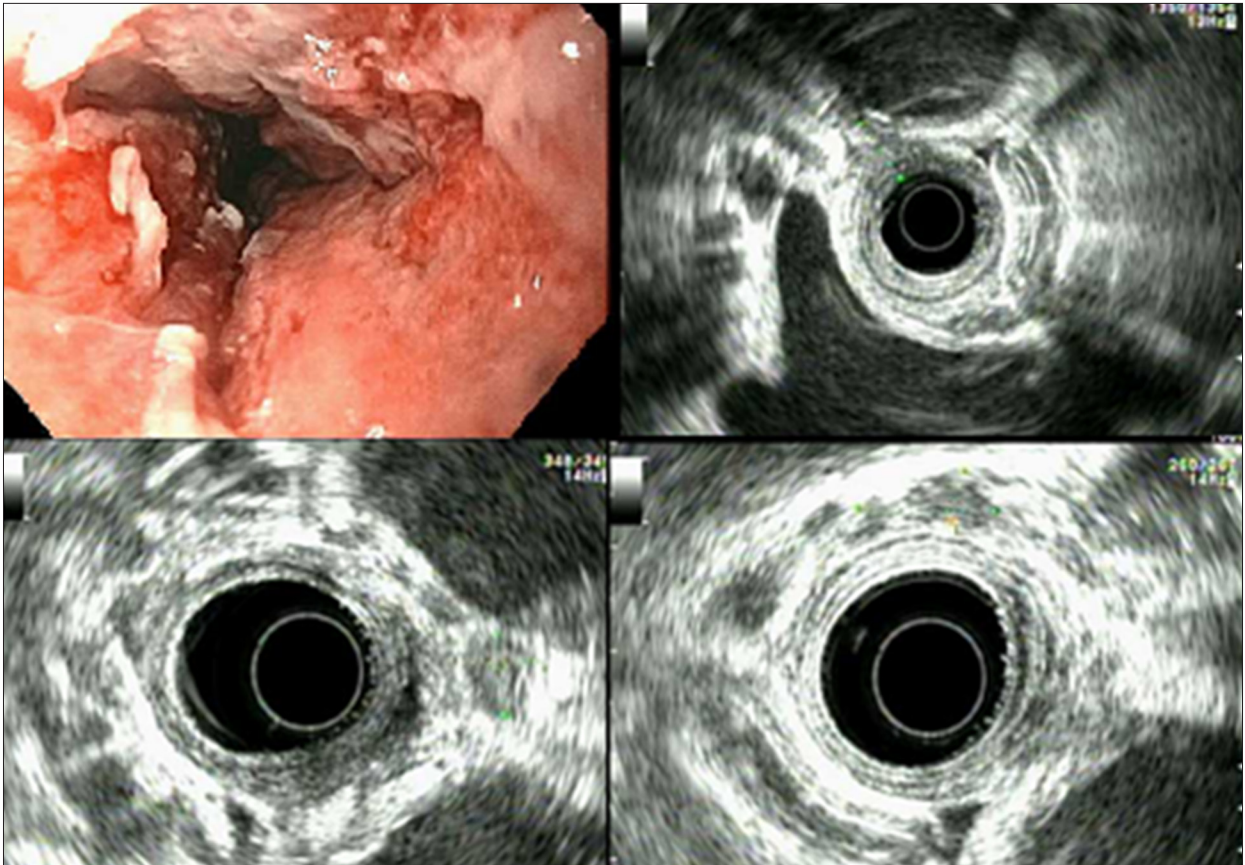


Figure 4. Endoscopic ultrasound (EUS) identified a hypochoic mass in the lower third of the esophagus, encountered at 35 cm from the incisors and extended to 38 cm which was the furthest the echoendoscope could be advanced due to the stenosis. The endosonographic borders were irregular, and a T3 lesion with multiple benign-appearing lymph nodes was detected. EUS guided biopsies showed poorly differentiate adenocarcinoma.

with granular irregular mucosa and moderate resistance to the passage of the endoscope. Pneumatic dilation was performed using a 40 mm balloon size over a guide wire for 5 total sessions. During the first 2 dilation attempts the balloon quickly migrated to the stomach. Hence extra attempts were required to ensure correct positioning. During the subsequent 3 dilations each of 1 minute duration, a lot of traction was required to keep the balloon in the lower esophageal sphincter and the impression was that the sphincter and distal esophagus were very tight and non-compliant. The post-procedure endoscopy and Gastrografin swallow confirmed no perforation and the patient was discharged home.

Because of the endoscopic findings of irregular and inflamed tissue present proximal to the LES suggesting a mass effect together with the pneumatic dilation experience as well as the chest CT findings in the distal esophagus, an upper endoscopic ultrasound (EUS) was performed 2 weeks later, to rule out the possibility of underlying cancer of the esophagus that could explain his refractoriness to the dilations. EUS identified a hypochoic mass in the lower third esophagus and several hypochoic lymph

nodes with poorly defined margins (Figure 4). The mass lesion was circumferential with irregular borders. There was one image that the mass appeared to protrude through the muscularis propria (layer 4) with intact interface between the mass and the adjacent structures. This was staged as T3 N0 Mx by endosonographic criteria. The biopsies of the mass indicated poorly differentiated adenocarcinoma. Tumor cells were diffusely positive for low molecular weight cytokeratin, focally positive for cytokeratin-7, and negative for high molecular weight cytokeratin, cytokeratin-20, P63, and leukocyte common antigen immunostains. This tumor is most compatible with poorly differentiated adenocarcinoma as the primary rather than metastatic tumor from the previous colon cancer. The adenocarcinoma was presumed to have developed in the setting of Barrett's esophagus. Patient had a PET scan which indicated no other metastasis. As a preventive measure to overcome any obstruction which could occur with the radiation therapy being planned preoperatively, the patient underwent esophageal stent placement and his nutrition status improved with ability to tolerate soft foods. He is currently being managed by the oncologist for his esophageal cancer with radiation and chemotherapy before surgery as the final goal.

Discussion

In patients presenting with esophageal dysphagia, it is important to determine the types of food that produce symptoms (solids, liquids, or both) and whether there is the progression of symptoms from solids to liquids. Dysphagia for both solid and liquids from the onset of symptoms is most likely due to an esophageal motility disorder, such as achalasia and scleroderma. On the other hand, progressive dysphagia, beginning with dysphagia to solids evolving to liquids, is usually caused by a mechanical problem such as cancer or a peptic stricture or Schatzki ring [1]. The symptoms due to a malignancy usually progress rapidly, while the peptic strictures are more insidious [2]. However, patients with the motility disorders of achalasia or scleroderma, may also exhibit a worsening of their dysphagia when there is no treatment intervention.

Achalasia results from the progressive degeneration of ganglion cells in the myenteric plexus in the lower esophageal sphincter and esophageal smooth muscle, leading to the loss of inhibitory innervation in the esophagus. Achalasia should be suspected in patients with dysphagia to solids and liquids with regurgitation of food from the esophagus, particularly occurring at night, sometimes accompanied by heartburn. Often this symptom triggers a trial of proton pump inhibitor (PPI) therapy because GERD is suspected by the primary care physicians. The heartburn accompanying the regurgitation is generally explained as the acidic pH of degraded food contents that have been retained in the esophagus [3]. Upper endoscopy indicates a dilated esophagus which appears to be atonic often with some retained food and a “tight” LES requiring pressure to push the endoscope into the stomach. A motility study is required to establish the diagnosis of achalasia and distinguish this entity from other motility disorders. The presence of aperistalsis following wet swallows in the distal two-thirds of the esophagus and incomplete LES relaxation on esophageal manometry, defined as a mean four-second integrated relaxation pressure (IRP) >15 mmHg in the setting of a normal to high pressure LES pressure are the manometric criteria required for the achalasia [3]. Other motility disorders, such as diffuse esophageal spasm and Jackhammer esophagus can also be detected in patients with nonstructural dysphagia [4]. Barium study is still helpful in confirming the classic “beaking” at the LES in achalasia. For our patient the “beaking” was more than 3 cm in length, exceeding the lower esophageal sphincter length, and somewhat asymmetric and irregular, all indicative that thus may not have been “garden-variety” achalasia.

Treatment of achalasia is focused on decreasing the resting LES to facilitate emptying of esophageal contents. This can be accomplished by mechanical disruption of the muscle fibers of the LES (e.g., pneumatic dilation (PD) or surgical myotomy). Biochemical reduction in LES pressure (e.g., injection

of botulinum toxin, or pharmacologically with oral nitrates, or calcium channel blockers) can be a short term initial therapy strategy [3]. Pneumatic dilation (PD) is the most cost-effective treatment for achalasia and has the advantage of being less invasive as compared with myotomy [5]. About 71-90% patients respond to PD initially. Approximately 50% of patients will relapse over a five-year period and will require additional dilation or surgery [6-9]. Type 1 and 2 achalasia by Chicago classification are the most appropriate for non-surgical intervention, while Type 3 is best managed by surgery, myotomy and a loose fundoplication.

Hence in our patient with a Type 2 classification, we would have expected a better response from repeated PDs. The friable pattern of the mucosa with the barium contrast study in our patient could have been attributed to some irregularities and erosions due to leftover food retention and/or *Candida* infection. General rule is not to biopsy at the same time as a pneumatic dilation in fear of increased risk of perforation. Soon after the 3rd dilation, abnormal CT and PD procedure findings led to EUS and defined the diagnosis of pseudoachalasia.

Pseudoachalasia is the term used when the manometric findings mimic “classic idiopathic achalasia”, but when some clinical characters are different and expected treatment outcomes are not achieved. Pseudoachalasia has a number of possible contributing etiologies: Chagas disease (*Trypanosoma cruzi* parasite), adenocarcinoma of the gastric fundus or distal esophagus/GE junction, lymphoma, and metastatic malignancies from breast, pancreas, or lung [10]. Pseudoachalasia in our patient is explained by adenocarcinoma of the lower esophagus involving the lower esophageal sphincter and microscopically infiltrating the myenteric plexus of the esophageal body and the LES [11]. The likelihood of pseudoachalasia in our patient was heightened by the following clinical characteristics: short duration of symptoms (in our patient the onset was less than six months); advanced age (our patient is over age 80); rapid and marked weight loss (our patient had a 50 pounds weight loss during 6 months); resistance to passing the endoscope through the gastroesophageal junction, and refractoriness to aggressive pneumatic dilation. In retrospect, our patient is atypical in that he had been experiencing dysphagia for up to 4 years. The biopsy might be done before or after pneumatic dilation. Pathology result is necessary to confirm the diagnosis of pseudoachalasia, especially since the patient had lost weight or anemia (probably due to iron deficiency, in old age, and disease did not last for long). Esophageal Biopsy is appropriate when abnormal or suspicious tissue to biopsy is identified. This was not apparent until the last pneumatic dilation in our patient as the atypical presentations and late onset of lost weight.

Hence, while the idiopathic achalasia may have been present initially the refractories, we witnessed was due to cancer occurring in underlying Barrett's esophagus. In such cases of refractories, a CT scan can be key to closely examine the esophagus and mediastinum. Endoscopic ultrasonography with fine-needle aspiration (EUS-FNA) is a new diagnostic tool to further investigate suspected etiologies of pseudoachalasia involving identifying and biopsying enlarged lymph nodes and masses [11]. In our patient, Barrett's esophagus was the underlying diagnosis with a history of GE reflux and predisposing to esophageal adenocarcinoma.

References:

1. Boeckxstaens GE, Zaninotto G, Richter JE: Achalasia. *Lancet*, 2014; 383(9911): 83–93
2. Lahbabi M, Ihssane M, Sidi Adil I, Dafr Allah B: Pseudoachalasia secondary to metastatic breast carcinoma mimicking radiation stenosis. *Clin Res Hepatol Gastroenterol*, 2012; 36(6): e117–21
3. Vaezi MF, Pandolfino JE, Vela MF: ACG clinical guideline: diagnosis and management of achalasia. *Am J Gastroenterol*, 2013; 108(8): 1238–49; quiz 1250
4. Maradey-Romero C, Gabbard S, Fass R: Treatment of esophageal motility disorders based on the Chicago classification. *Curr Treat Options Gastroenterol*, 2014; 12(4): 441–55
5. Kostic S, Johnsson E, Kjellin A et al: Health economic evaluation of therapeutic strategies in patients with idiopathic achalasia: results of a randomized trial comparing pneumatic dilatation with laparoscopic cardiomyotomy. *Surg Endosc*, 2007; 21(7): 1184–89
6. Boeckxstaens GE, Annese V, des Varannes SB et al: Pneumatic dilation versus laparoscopic Heller's myotomy for idiopathic achalasia. *N Engl J Med*, 2011; 364(19): 1807–16
7. Eckardt VF, Gockel I, Bernhard G: Pneumatic dilation for achalasia: late results of a prospective follow up investigation. *Gut*, 2004; 53(5): 629–33
8. Zerbib F, Thetiot V, Richy F et al: Repeated pneumatic dilations as long-term maintenance therapy for esophageal achalasia. *Am J Gastroenterol*, 2006; 101(4): 692–97
9. Tabola R, Grabowski K, Lewandowski A et al: Achalasia – balloon dilation or surgery? *Med Sci Monit*, 2013; 19: 1089–94
10. McCallum RW: Esophageal achalasia secondary to gastric carcinoma. Report of a case and a review of the literature. *Am J Gastroenterol*, 1979; 71(1): 24–29
11. Tracey JP, Traube M: Difficulties in the diagnosis of pseudoachalasia. *Am J Gastroenterol*, 1994; 89(11): 2014–18

Conclusions

Pseudoachalasia secondary to an esophageal malignancy should be suspected when dysphagia progresses despite technically well-performed pneumatic dilations, and endoscopic and imaging findings raise questions as to whether this is “classic” achalasia. EUS is a new diagnostic tool for staging the malignancy and obtaining diagnostic tissue.

Disclosure of financial arrangements

No financial disclosure for this article