Published in final edited form as:

Ann Thorac Surg. 2012 January ; 93(1): e11–e12. doi:10.1016/j.athoracsur.2011.07.064.

A Diagnostic Consideration for All Ages: Pseudoachalasia in a 22-year-old

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Abstract

Pseudoachalasia is a rare clinical diagnosis with diverse manifestations. We present the case of a 22-year-old male with esophageal adenocarcinoma who was initially diagnosed with achalasia. This unfortunate presentation reinforces the importance of a careful preoperative workup for dysphagia irrespective of age.

Case Report

A 22-year-old male was referred to our institution with the presumptive diagnosis of achalasia. His history was significant for a thirty-pound weight loss over a three-month period with progressive dysphagia to solids. A previously healthy and active individual, he noted no abnormalities in growth or development with no medical or surgical history. Physical examination revealed an absence of palpable adenopathy, abdominal tenderness, or mass effect. Barium swallow demonstrated a bird-beaked impairment at the lower esophageal sphincter, consistent with the diagnosis of achalasia (figure 1). Esophageal manometry confirmed decreased esophageal peristalsis with failure of the lower esophageal sphincter to relax. An esophagogastroduodenoscopy (EGD) and dilation was performed with minimal symptom relief. He was then referred to our institution for surgical intervention.

The patient was counseled regarding the diagnosis of achalasia and scheduled for repeat EGD and laparoscopic Heller myotomy. In the operating room, EGD revealed significant thickening in the distal esophagus with a tight, irregular stricture. We could not pass the scope into the stomach, took multiple biopsies of the stricture, and the myotomy was cancelled. The final pathology was consistent with adenocarcinoma. Staging of his esophageal adenocarcinoma included endoscopic ultrasound, demonstrating a T3N0 tumor, and positron emission tomography (PET) that revealed no evidence of metastases. He was treated with neoadjuvant chemotherapy (Cisplatin and 5-FU) and radiation therapy (50.4Gy, 28 fractions). An Ivor-Lewis esophagectomy was performed six weeks after he completed his neoadjuvant therapy (figure 2.). The patient recovered without postoperative complications and his pathology showed persistent T3N0 disease with significant treatment effect. Unfortunately, two liver metastases were detected on routine surveillance and he died eleven months after his esophagectomy.

Discussion

Pseudoachalasia is a defined spectrum of disease that secondarily impairs distal esophageal motility and imperfectly mimics primary achalasia¹. This diagnosis demands committed clinical, radiographic, manometric, and endoscopic assessment to achieve distinction from primary achalasia. Pseudoachalasia, originally described by Ogilvie in 1947, is the result of a primary tumor at the esophagogastric junction in 70% of patients^{2–4}. Adenocarcinoma of the distal esophagus in the setting of Barrett's esophagus is the predominate presentation; however, many intrinsic and extrinsic conditions resulting in lower esophageal sphincter impairment have been described³. Pseudoachalasia accounts for only 2.4–4% of patients with an achalasia-like syndrome^{2,3}. The classic symptom presentation of short-term interval weight loss, progressive dysphagia, and chest pain are secondary to impaired esophageal emptying at the distal esophagus. Demographic study has demonstrated no significant gender predisposition with a median age of 61 years³. Our experience with this unfortunate 22-year-old man is extremely rare and highlights the need for committed preoperative and intraoperative assessment for all distal esophageal pathologies irrespective of age.

Careful preoperative assessment is imperative prior to surgical intervention. Barium swallow findings often mimic the classic primary achalasia findings of a bird-beaked distal esophagus, as demonstrated in our case presentation. Amyl nitrate, a pharmacologic inhalant and smooth muscle relaxant, induces a measurable increase of 2mm or more in sphincter diameter for patients with primary achalasia. Patients with pseudoachalasia exhibit no response⁵. Conventional esophageal manometry, while routine for our esophageal myotomy candidates, often provides no differentiation between pseudoachalasia and primary achalasia¹. Endoscopic findings supporting consideration of pseudoachalasia are mucosal ulceration or nodularity, reduced compliance of the esophagogastric junction, or an inability to pass the endoscope into the stomach¹. Endoscopic biopsy remains the most definitive diagnostic tool for the diagnosis of pseudoachalasia occurring secondary to distal esophageal or esophagogastric junction carcinoma. The hallmark pathologic characteristics of this condition are an invasion and disruption of the myenteric plexus or a paraneoplastic autoimmune-mediated depletion of myenteric ganglion cells³.

This unfortunate 22-year-old male with esophageal adenocarcinoma and the presumptive diagnosis of achalasia imparts an appreciation for the diverse clinical manifestations of pseudoachalasia. Irrespective of age, a lack of commitment to definitive preoperative assessment may result in inappropriate surgical treatment or progression of malignancy. Intraoperative endoscopy and laparoscopic evaluation of the distal esophagus and proximal stomach are imperative for confirmatory diagnosis at the time of potential myotomy.

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Figure 1. Barium esophagogram demonstrating a bird-beaked appearance of the distal esophagus with marked dysmotility.

Figure 2. Surgical specimen following neoadjuvant therapy demonstrating marked fibrosis at the gastroesophageal junction. Pathology revealed negative margins and a T3N0 esophageal adenocarcinoma.