Squamous Cell Carcinoma Arising in an Epiphrenic Diverticulum

Costa J1, De Michele S2, Pagan C3, Moccia R4 and Gorenstein L1*

1Department of Surgery, Columbia University Medical Center, USA
2Department of Pathology and Cell Biology, Columbia University Medical Center, USA
3Department of Medicine, Mount Sinai Medical Center, USA

Abstract

Esophageal cancer arising in an Epiphrenic Diverticulum (ED) is rare, whether or not associated with achalasia. The majority of ED develops in patients without a defined esophageal motor disorder, but they can also occur in patients with either achalasia or similar esophageal motility disorders, such as hypertensive LES syndrome (<10% of epiphrenic diverticula). To date there remains limited information in the literature addressing the development of cancer in an ED. This case report describes achalasia with an ED that presented with a massive GI bleeding, and was found unexpectedly to have squamous cell cancer arising within the ED.

Keywords: Epiphrenic diverticulum; Achalasia; Esophageal; Gastroesophageal junction

Introduction

Epiphrenic Esophageal Diverticula (ED) represents approximately 10% of esophageal diverticula [1-Plakett]. Cancer arising within an ED is extremely rare with a paucity of reported cases in the literature [2-Fu, 3-Choi, 4-Herbella]. Herein, we present a case report of squamous cell carcinoma arising in an ED, in a patient with a history of achalasia.

Case Presentation

A 66-year old woman with over ten years of dysphagia to both solids and liquids presented with an acute upper gastrointestinal bleed. She required two units of blood, but no intervention to stop the bleeding. The initial upper endoscopic (EGD) exam was limited due to large clots in the esophagus, and the initial diagnosis was felt to be bleeding in an incarcerated paraesophageal hiatal hernia. There was no active bleeding seen, but the mucosa appeared edematous and friable. A repeat EGD performed the next day, demonstrated a dilated serpiginous shaped esophagus with thickened mucosa, and a large ED arising at 35 cm. An adherent blood clot was present at the orifice of the diverticulum, and there was no active bleeding when the clot was removed. A smooth benign stricture or “bird beak” was noted at the Gastroesophageal Junction (GEJ) at 35 cm, easily allowing passage of the endoscope with gentle pressure into a normal appearing stomach. The mucosa in the diverticulum appeared inflamed and “cobblestoned,” but there were no irregularities or nodules to suggest malignancy.

No biopsy of the mucosa in the diverticulum was performed, as malignancy was not suspected. Biopsy of the gastric mucosa demonstrated focally active gastritis and was negative for H. pylori. Barium swallow showed a torturous and dilated esophagus and a large ED arising with smooth tapering of the LES consistent with achalasia (Figure 1). Esophageal manometry was performed, and was consistent with achalasia (Chicago classification type II). Based on manometry, endoscopy, and upper GI series, the presumed diagnosis was achalasia associated with a large epiphrenic diverticulum.

She underwent elective laparoscopic surgery with the intention to resect the diverticulum, and perform a long myotomy and fundoplication. Intraoperative EGD demonstrated a large ED, whose origin began three to four centimeters above the GEJ. The GEJ was hypertrophied and narrowed, but allowed the endoscope to pass, consistent with a diagnosis of achalasia. The esophagus was circumferentially mobilized within the hiatus to the level of the inferior pulmonary veins. Mobilization of the diverticulum within the mediastinum was challenging because of inflammation between the diverticulum, the aorta, and the right pleura. The posterior vagus nerve was intimately adherent to the junction of the diverticulum and the esophageal muscle. A laparoscopic anterior...
Barium study demonstrating achalasia and large epiphrenic diverticulum. The carcinoma showed a degree of squamous differentiation. Lymphovascular invasion and lymph nodes metastasis were present. The background stroma was fibrotic with hypertrophic nerves, and a prominent lymphoplasmacytic infiltrate (Figure 2A and 2B). The cells appeared pleomorphic with irregular and hyperchromatic nuclei. Nucleus to cytoplasm ratio is increased with numerous mitotic figures noted (Figure 2D). The histopathologic examination revealed a moderately differentiated squamous cell carcinoma with submucosal invasion. The margins were negative for malignancy, and the pathologic stage was T1bN0. The diverticulum is malignant. If malignant changes are identified, it is imperative to perform a limited posterior thoracotomy; the diverticulum was dissected from the origin of the diverticulum onto the stomach, resection of the diverticulum, and a partial fundoplication is the treatment of choice [3]. Epiphrenic diverticulum associated with achalasia should have a myotomy and partial fundoplication in conjunction with diverticulectomy.

Patients with achalasia are at increased risk of developing esophageal cancer. The lifetime prevalence of esophageal cancer in patients with achalasia is 3% [5]. Stasis in the dilated esophagus is felt to cause chronic inflammation resulting in mucosal cell genetic alterations, dysplastic degeneration and ultimately malignant transformation. Malignancy rarely occurs within an ED under 2 cm in size [6]. Esophageal cancer arising in achalasia is often diagnosed at an advanced stage, primarily because of delay in diagnosis; however the biologic behavior of cancers arising in achalasia may also be different than those occurring de novo. Malignancy developing within an ED may also be diagnosed at an advanced stage. Because of the absence of the muscularis propria, thereby tumors developing in an ED can easily extend into surrounding mediastinal structures, such as the aorta, pericardium, the lung, or left atrium [7].

If malignancy is identified preoperatively, esophagectomy should certainly be considered, especially if the mucosa at the base of the diverticulum is malignant. Esophagectomy, with either an intrathoracic or cervical anastomosis would reduce the risk of a local recurrence, by insuring an adequate resection margin. Esophagectomy also provides good functional results for patients with long standing achalasia, especially when there is significant general, esophageal cancers confined to the mucosa (Tis or T1a) esophageal dilation and tortuosity (i.e. sigmoid esophagus). In can be cured by endoscopic resection, thereby avoiding the necessity of an esophagectomy. As evidenced by this case, if the malignant mucosal disease is confined to the ED and does not involve the base of the diverticulum, then diverticulectomy alone can be curative [5]. Because the resection margins were negative, no further treatment was considered, and she has been followed with endoscopic and CT scan surveillance. Had malignancy been identified at the surgical resection margin, then further treatment, ideally esophagectomy would be recommended.

In summary, patients with achalasia found to have an ED, and many years of symptoms, should have a thorough endoscopic evaluation of the ED, including biopsies to exclude the possibility of malignancy. If malignant changes are identified, it is imperative to
perform thorough staging, including PET/CT, and EUS, to determine both local extent, and exclude distant spread, when determining surgical options.

References


