Submucosal carcinoma of the gastroesophageal junction diagnosed after peroral endoscopic myotomy

Gastroesophageal junction (GEJ) malignancies are found in 4.7% of patients who fulfill the manometric criteria for achalasia [1]. Such malignancies may manifest as pseudoachalasia because of submucosal infiltration and secondary impairment of the inhibitory neurons of the esophageal myenteric plexus, therefore mimicking the manometric pattern of achalasia [2]. Despite endoscopic biopsies, false-negative rates of 25% may mask this cause of pseudoachalasia [3].

A 62 year old man with no history of Barrett’s esophagus presented with a 4-month history of dysphagia, vomiting, and weight loss. His initial esophagogastroduodenoscopy (EGD) showed a dilated esophagus and tight GEJ; multiple biopsies were negative. A computed tomography (CT) scan of the thorax and abdomen showed a bulky GEJ with no definite mass lesion. Endoscopic ultrasound (EUS) also showed a circumferential thickening in the area of the GEJ, but no masses suggestive of malignancy or suspicious lymph nodes were seen (Fig. 1). High resolution manometry suggested type II achalasia.

As malignancy had been excluded as a cause of the tight GEJ, the patient underwent peroral endoscopic myotomy (POEM). During creation of the submucosal tunnel, thickening and fibrosis of the muscularis propria was encountered near the GEJ (Fig. 2); however, intraoperative frozen sections were normal. The remainder of the POEM proceeded uneventfully, the myotomy being performed with a Triangle Tip knife (Olympus) and the tunnel entry being closed with clips (Video1). Multiple biopsies of the mucosa and muscularis propria showed cytologic atypia of unknown significance but were negative for malignancy.

The patient was monitored closely in the clinic and his symptoms recurred within a month. Another EGD showed a GEJ stricture 38 cm from the incisors. Biopsies were again indeterminate. A repeat CT scan confirmed a 3.6-cm concentric mural thickening at the GEJ. Surgery was therefore advised. Intraoperatively, a localized tumor was found at the GEJ. A total gastrectomy with D2 lymphadenectomy was performed. Final histology revealed a moderately differentiated submucosal adenocarcinoma of the GEJ with no involvement of the mucosa (Fig. 3). After receiving chemotherapy, the patient remained free of symptoms and disease 1 year postoperatively.

Fig. 1 Endoscopic ultrasound (EUS) in a 62 year old man with type II achalasia showing a circumferential thickening at the area of the gastroesophageal junction (GEJ) but no mass lesions.

Fig. 2 Image taken during peroral endoscopic myotomy (POEM) showing the scarred and infiltrating tissue that was found during submucosal dissection at the level of the abnormally thickened and indurated gastroesophageal junction (GEJ).

Fig. 3 Final histology following total gastrectomy showing normal looking mucosa on the right of the image and a submucosal tumor not infiltrating beyond the muscularis mucosa on the left of the image (magnification × 100).
This case highlights the rare occurrence of a submucosal GEJ adenocarcinoma that was not identified on either pre-POEM endoscopy with mucosal biopsies or on EUS in a patient with symptoms and signs suggestive of achalasia. This may have been due to a submucosal tumor and sampling error. After performing POEM, clinicians should maintain a degree of suspicion for malignancy if a patient’s symptoms fail to resolve.

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