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Case Report of a Primary Malignant Melanoma of the Esophagus: An Atypical Presentation of an Extremely Rare Disease

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Abstract

Primary Malignant Melanoma of the Esophagus (PMME) is an extremely rare disease with a dismal prognosis. Most of our current knowledge comes from single case reports and short case series. The purpose of this case report is to share our experience with a case of atypical presentation of this poorly known condition. We present a case of PMME in a 71-year-old patient displaying unique acute chest pain and a slight hindrance swallowing that had been evolving for six weeks. Endoscopy showed an achromic polypoid mass appended to the upper esophagus. The diagnosis of PMME was based on an immunohistochemical analysis of biopsies performed endoscopically after excluding the presence of esophageal metastasis from another primary melanoma. We assessed extension using endoscopic ultrasound and PET/CT and decided to surgically treat the patient without adjuvant or neoadjuvant therapy.

Introduction

Primary Malignant Melanoma of the Esophagus (PMME) is an extremely rare but highly aggressive neoplasm. It accounts for 0.1% to 0.2% of all primary esophageal malignancies [1,2]. The annual occurrence rate of this disease has been estimated at 0.03 per million people [3-5]. This tumor arises from melanocytes that are normally present in the basal layer of the esophageal epithelium in 4% of the normal population [4].

This condition was first described by Baur in 1906 and first histologically confirmed by Garfinkle and Cahan in 1952. By 2011, 337 cases had been published worldwide, mostly in single case reports [4].

PMME mainly affects men with a male-to-female ratio of 2:1 with a peak incidence in the 6th and 7th decades [1]. Patients with PMME most commonly present dysphagia (73%), retrosternal pain (24%) and weight loss (16%). A few patients may also display melena or hematemesis as first symptoms [4,6]. Mean duration of symptoms before diagnosis is short with an average of three months [1,7].

Endoscopically, the tumor is often in polypoid and pigmented form and occurs mostly (>90%) in the mid and lower third of the esophagus [8]. This is probably due to the greater concentration of melanocytes in these regions [2].

Literature reviews report average survival ranging from 10 to 13 months. In a series of 139 patients, Sabanathan et al. reported a 5-year survival rate of 4.2% [1], while a more recent literature review published in 2002 by Volpin et al. reported a more encouraging figure of 37% [2]. As mentioned above, the prognosis of PMME is dismal. Indeed, patients often present disease that is already at a metastatic stage [2]. The most common metastatic sites are the paraesophageal and supraclavicular lymph nodes, liver, lungs and brain [2,4,9-11].

Given the low number of cases published so far, PMME treatment has not yet been well established. The best outcomes have been observed with esophagectomy for patients with localized disease [12].

We present a case of a 71-year-old patient diagnosed with an atypical presentation of PMME and treated by total esophagectomy with extensive lymphadenectomy.

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Case Presentation

In December 2016, a 71-year-old female presented with slight hindrance in swallowing for 6 weeks without vomiting or weight loss. She also reported one episode of acute burning chest pain radiating to the jaw during this interval. In the relevant family history of the patient, we noted histories of lung cancer in the brother and father at age 60. The physical examination including dermatological evaluation was unremarkable.

All laboratory findings were normal. In this context, upper GI endoscopy was performed (Figure 1). A 1-centimeter polypoid mass was discovered in the upper esophagus located at 23 cm from the incisors. Accurate diagnosis was made histologically and immunohistochemically on the biopsy materials. Staining was positive for the S-100 protein, HMB-45 and Melan-A, indicating melanoma.

Further investigations were performed to distinguish between primary tumor and secondary metastatic sites. Careful cutaneous and ophthalmological examinations failed to identify any other suspected location.

Extension was assessed *via* Endoscopic Ultrasound (EUS) and PET/CT analysis. Endoscopic ultrasound showed a hypoechoic and regular polypoid lesion that did not infiltrate the muscularis (Figure 2). No lymphadenopathy was detected. The tumor was endoscopically classified uT1N0.

The esophageal lesion had increased metabolic activity on PET/CT images (Figure 3). The latter also showed a focus in the right parietal lobe of the brain with no substrate on non-injected CT. A brain CT with contrast injection excluded cerebral metastasis. No metastasis was found and the final diagnosis was PMME.

Following multidisciplinary consultation, the decision was made to surgically treat the melanoma. No surgical contraindications were found and the patient underwent esophagectomy three-field with extensive lymphadenectomy. The patient did not receive adjuvant or neoadjuvant therapy. The postoperative course was uneventful except for a cervical abscess resolved by surgical drainage.

Macroscopic analysis of the mass showed a whitish polypoid mass (1.9 cm × 1.3 cm × 1.3 cm) protruding in the esophageal lumen. Frozen sections showed healthy margins. Histological analysis revealed a widely amelanotic polypoid-shaped PMME infiltrating the muscularis mucosae with no evidence of deep invasion.

A total of 25 lymph nodes were analyzed and none was involved. The tumor was finally classified pT1a N0 M0 according to the TNM classification of malignant tumors 7th edition.

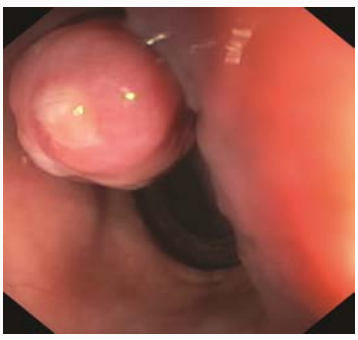

Figure 1: Polypoid mass of the upper esophagus.

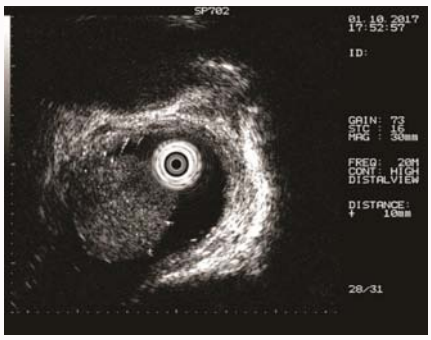

Figure 2: Endoscopic ultrasound showing a hypoechoic and regular polypoid lesion.

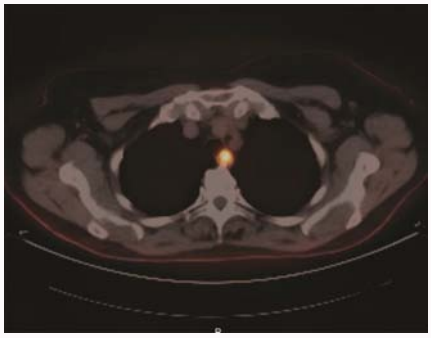

Figure 3: Metabolic activity of the esophageal lesion.

In July 2017, the patient was still disease-free on follow-up performed by a thorough physical examination and cervico-thoraco-abdominal CT-scan. The patient benefited from six endoscopic dilations following anastomotic stenosis. After four years of follow-up, the patient is doing well.

Discussion

We present a case of PMME in a 71-year-old female with symptoms of slight dysphagia and acute chest pain. The diagnosis of PMME was made by immunohistochemistry on biopsies performed endoscopically. Total esophagectomy with extensive lymph node dissection was performed. The patient is still in remission 11 months later.

This PMME differs from most presented cases for several reasons. After four years of follow-up, the patient is doing very well. First, despite the fact that the majority of cases presented are driven to seek medical care because of dysphagia, it should be noted that other symptoms are also encountered in this type of pathology such as acute chest pain in this case [13].

Moreover, the literature reports that the location of PMME is distributed as follows: Approximately 43% occur in the lower third, 29% in the middle third, 18% at the junction of the middle and lower thirds, and finally only 10% in the upper third. A mass in the proximal third of the esophagus does not exclude PMME and must be investigated in the same way as a mass in the lower two-thirds [1]. In addition, the majority of reported cases are pigmented forms whereas totally amelanotic forms represent only 10% to 25% of all PMME. The absence of visible pigmentation during endoscopy does not exclude PMME [4].

PMME symptoms most often encourage practitioners to perform

esophagoscopy as part of the work-up. In most cases, it will show a single tumor of polypoid form as in the present case [1,2,6]. It should be noted that other atypical forms of PMME exist, such as multiple lesions, ulcerative lesions, fungating lesions, circumferential lesions, irregular flat areas or diffuse esophageal thickening [4].

The diagnosis of this type of tumor is well established in the literature. A pigmented polyploid tumor may suggest the diagnosis. However, the final diagnosis is established by means of immunohistochemistry. PMME typically reacts to antibodies against S-100, HMB-45 and Melan-A proteins [10,13] while they are generally negative for epithelial markers, smooth muscle and lymphoid markers [4].

As used in our case for preoperative staging, endoscopic ultrasound is useful for assessing esophageal wall infiltration and possible periesophageal and mediastinal enlargement [9]. We subsequently performed a PET/CT to complete the extension assessment. According to several publications, this technique represents the first-line modality for preoperative staging of the tumor [5,8].

The literature suggests that the preferred treatment for PMME is total surgical resection and extensive lymph node dissection [2,5,9,13-15]. However, according to the latest publications, esophagectomy would be beneficial only for localized forms without lymphatic metastases [11,12]. Indeed, lymphatic involvement is associated with poor disease-free survival [5]. On the other hand, cases of localized PMME treated by esophagectomy with long-term survival have been described [4,5,9,12]. The place of chemotherapy, radiotherapy and immunotherapy is not well established [6,11,13,15]. According to some authors, these therapies could have a place in palliative situations [2,8,10]. Further investigation is needed to establish whether these therapies are useful in the treatment of PMME.

In conclusion, PMME is an extremely rare tumor with a dismal prognosis. Localized forms such as the present case can benefit from radical surgical treatment with the possibility of remission in the medium or long term.

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