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Case Report

An unusual presentation of primary oesophagal melanoma in an elderly male: a case report

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ABSTRACT

Primary malignant melanoma of the oesophagus (PMME) is a very rare and highly aggressive neoplasm, representing <0.2% of all oesophageal carcinomas. Malignant melanoma usually occurs in the skin but can sometimes arise from other non-cutaneous locations, such as mucosa, uvea, upper respiratory tract, gastrointestinal tract, and genitourinary tract. In particular, primary oesophageal melanoma is an extremely rare condition that is associated with a bad prognosis because of the aggressive nature of this tumour and late diagnosis. We present a rare case of an 85-year-old patient who had complaints of per rectal bleeding, vomiting, and shortness of breath. Physical and endoscopic examination found an oesophageal tumour. Histologic examination showed changes consistent with a malignant melanoma. PMME was diagnosed on the basis of the presence of typical histomorphological and immunohistological changes. Being extremely rare, PMME is a difficult disease for diagnosis and should be included in the differential diagnoses of melanomas of the oesophagus.

Keywords: Primary oesophagal melanoma, Gastrointestinal bleeding, Oesophagal mass, Elderly patient, Thoracic

INTRODUCTION

Primary malignant melanoma of the oesophagus (PMME) is an extremely rare and aggressive neoplasm, first described by Raven in 1906.[1,2] It accounts for less than 0.2% of all oesophageal malignancies and typically affects elderly males, most often in the sixth to eighth decades of life.[1,3,4] The tumour arises from melanocytes present in the basal layer of oesophageal squamous epithelium and usually involves the lower and middle third of the oesophagus.[3] Clinical symptoms are often nonspecific, with dysphagia, weight loss, and chest discomfort being common. This report describes a rare presentation of oesophageal melanoma in an elderly male, where gastrointestinal bleeding and systemic symptoms were the predominant features.

CASE REPORT

An 85-year-old male presented to the gastroenterology department with complaints of per rectal bleeding, repeated episodes of vomiting, and progressive breathlessness over last 15 days. There was history of weight loss, black stools, blood in vomitus. On physical examination, there were no pigmented skin lesions or lymph node swellings. Abdominal examination revealed colicky pain in the abdomen.

Laboratory investigations demonstrated normocytic normochromic anaemia (haemoglobin: 10.8 g/dl) and elevated inflammatory markers. Liver and renal profiles were also mildly deranged. Ultrasound of the abdomen revealed inflammatory changes in the distal ileum and ascending colon, with moderate ascites and evidence of

cystitis. Given the respiratory complaints and vomiting, a contrast-enhanced computed tomography (CECT) of the thorax was performed, which revealed asymmetric mural thickening of the proximal esophagus, with an enhancing soft tissue mass completely occupying the esophageal lumen, leading to significant narrowing and luminal obstruction (Figure 1A). The appearance raised suspicion of a malignant esophageal mass. Given the patient's age and unusual presentation, a primary esophageal melanoma was included in the differential diagnosis.

Endoscopy and biopsy were recommended for histopathological confirmation. On endoscopy, a polypoidal, nodular black pigmented mass was seen projecting in the esophageal lumen (Figure 1B). On histopathological examination, infiltrative tumor arranged in nests and sheets were seen. The tumor cells were round to spindle shaped displaying moderate to marked nuclear pleomorphism, high nucleocytoplasmic ratio, nuclear hyperchromasia, prominent nucleoli and pigmented cytoplasm. There was irregular distribution of melanin pigment. Mitosis was frequent. Areas of necrosis and inflammatory infiltrates were seen and no esophageal lining epithelium was seen (Figure 2 A and B).

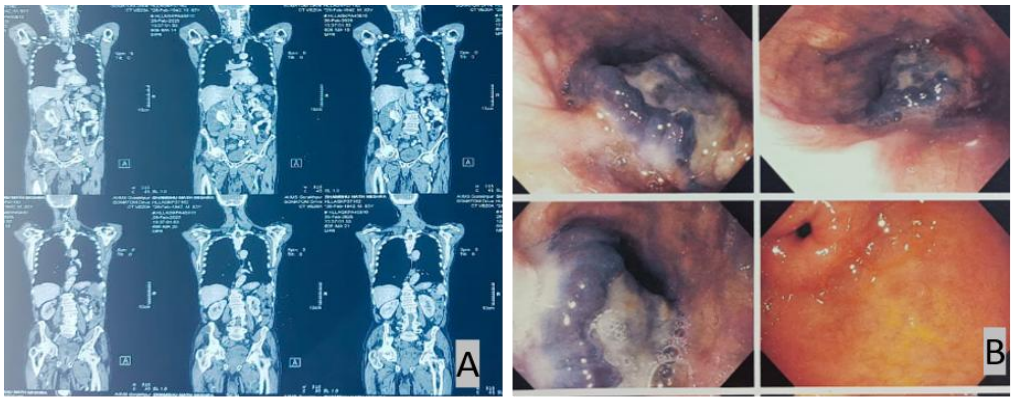

Figure 1: (A) CECT showing enhancing soft tissue mass lesion occupying esophageal lumen causing significant narrowing, (B) Endoscopy revealed large eccentric Polypoidal, nodular black pigmented mass just below upper esophangeal sphincter.

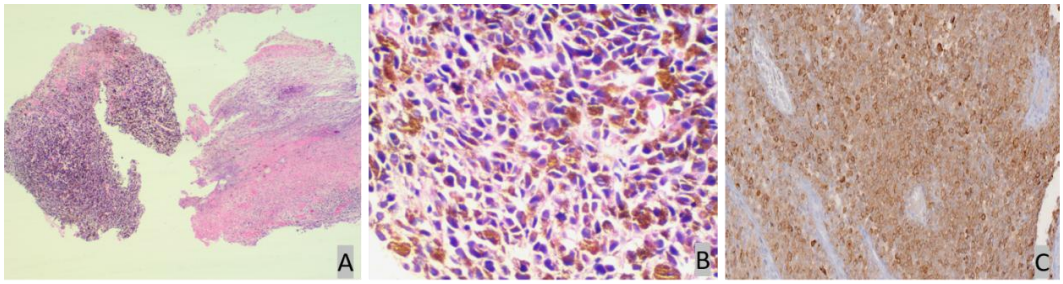

Figure 2: (A and B) low power and high power of infiltrative tumor arranged in nests and sheets along with irregular distribution of melanin pigment (H&E), (C) Immunohistochemistry revealed positivity for HMB45 marker.

Immunohistochemistry was positive for HMB-45 (Figure 2 c) favouring esophageal melanoma. In light of the age of the patient, their condition, and the aggressiveness of the disease, a palliative management strategy was employed. Palliative care was provided to the patient and included efforts to relieve symptoms and improve the quality of life of the patient. Efforts were geared towards relieving the patient of symptoms as opposed to curative efforts. It was recommended that the patient should make follow-ups at the oncology center regarding their response to the management and any progression of the disease.

DISCUSSION

PMME is a rare entity with fewer than 400 cases reported globally.[1,4] It comprises approximately 0.1% to 0.2% of all primary esophageal cancers.1 The origin of the tumor is believed to be melanocytes located in the basal layer of the squamous epithelium, whose presence in the esophagus was first confirmed histologically in the 1960s.[5] The mechanism of malignant transformation is not completely understood but is thought to involve dysregulation of melanin-producing pathways and genetic mutations commonly seen in cutaneous melanoma.[6] It is

often misdiagnosed due to its resemblance to more common tumors like squamous cell carcinoma or adenocarcinoma. Melanocytes, although sparsely distributed, are present in the esophageal epithelium and can give rise to malignant melanoma.[5] The tumor most commonly affects men in their 60s–80s and frequently involves the middle to lower third of the esophagus.[1,4] Dysphagia is the most common symptom, followed by weight loss, retrosternal pain, odynophagia, and melena.[5] Hematemesis and systemic symptoms, such as those seen in our patient, are uncommon but may indicate advanced disease with necrosis, ulceration, or secondary gastrointestinal involvement.[6] In rare cases, patients may present with upper GI obstruction or atypical symptoms such as vomiting and dyspnea, as seen in our case. The presentation of per rectal bleeding is even rarer and may indicate synchronous colonic pathology or bleeding secondary to altered gastrointestinal motility and mucosal compromise. Radiologic findings often show a lobulated or polypoidal mass with eccentric thickening and luminal obstruction. Endoscopic evaluation and biopsy remain the cornerstone for diagnosis. Immunohistochemistry plays a crucial role in distinguishing melanoma from other malignancies, with S-100, HMB-45, and Melan-A being the key markers.[7,8] Differential diagnoses include metastatic melanoma to the esophagus (more common than PMME), pigmented neuroendocrine tumors, gastrointestinal stromal tumors (GISTs), and spindle cell carcinomas. Exclusion of a primary lesion elsewhere, especially the skin, ocular region, and anus, is essential. A thorough dermatologic and ophthalmologic exam, along with PET-CT when available, aids in excluding other primary sites.[6]

PMME is highly aggressive, with a tendency for early lymphatic and hematogenous spread, most commonly to the liver, lungs, and regional lymph nodes. Approximately 40%–50% of cases have metastasis at the time of diagnosis.[9] The median survival is dismal, usually less than 12–15 months, despite treatment. Surgical resection remains the cornerstone of therapy in localized disease and offers the best chance of prolonged survival.[10] Esophagectomy with wide margins and lymphadenectomy is typically recommended. However, due to late presentation and comorbidities, many patients are not surgical candidates. In such cases, palliative radiotherapy and endoscopic stenting for obstruction relief may be employed.[10] Systemic therapy is an evolving area in PMME. Given the poor prognosis, early diagnosis is crucial. PMME should be suspected in elderly patients presenting with obstructive esophageal symptoms, especially when a pigmented lesion is seen on endoscopy. A multidisciplinary approach involving gastroenterologists, pathologists, oncologists, and surgeons is vital for diagnosis and management.

CONCLUSION

This case illustrates the diagnostic complexity of primary esophageal melanoma, particularly when presenting with nonspecific systemic and gastrointestinal symptoms. While rare, clinicians must consider such diagnoses in elderly patients with obstructive esophageal masses and atypical GI symptoms. Early endoscopic evaluation and histopathology are crucial for timely diagnosis and management.

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