KANSAS JOURNAL of MEDICINE

Case Report

Disseminated Melanoma with Extensive Gastrointestinal Tract Involvement: Incidental Detection of Metastases on Upper Endoscopy

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Received Mar. 8, 2025; Accepted for publication Jul. 3, 2025; Published online Aug. 15, 2025
Kans J Med 2025 Jul-Aug; 18:92-94. https://doi.org/10.17161/kjm.vol18.23693

INTRODUCTION

Malignant melanoma (MM) is a neoplasm arising from melanocytes, which are primarily located in the skin but also can be found in the uvea, oral cavity, nasopharynx, anus, urinary tract, and vagina.[1,2] MM accounts for approximately 2% of all cancers and is the most common carcinoma to metastasize to the gastrointestinal (GI) tract, with symptomatic GI involvement reported in 1-5% of patients.[2,3] The small intestine is the most frequently affected site (51-71%), followed by the stomach (27%) and colon (22%); esophageal involvement is rare (5%).[2]

Symptoms typically are non-specific and can mimic those of other GI metastases. Common presentations include abdominal pain, fatigue, constipation, melena, hematochezia, dysphagia, small bowel obstruction, and bowel perforation.[4]

CASE REPORT

A 62-year-old male with no known medical history presented with dysphagia, poor oral intake, unquantified weight loss, jaundice, and dark urine over several weeks. Laboratory evaluation revealed a cholestatic pattern of liver injury, with a total bilirubin of 6.3 mg/dL, alkaline phosphatase of 1465 U/L, and aspartate aminotransferase/alanine aminotransferase levels of 215/164 U/L. A computed tomography (CT) scan of the abdomen and pelvis showed numerous hypoattenuating lesions throughout the liver, consistent with metastatic disease.

To evaluate the patient's progressive dysphagia, an esophagogastroduodenoscopy (EGD) was performed. It revealed a single melanotic plaque in the upper third of the esophagus, along with melanotic lesions in the stomach and duodenum (Figures 1 and 2). Histopathological analysis showed nests of melanocytes with atypical mitotic figures (Figure 3). Immunohistochemical staining was positive for S100, SOX-10, and Mart-1, and negative for pancytokeratin, findings consistent with metastatic melanoma (Figures 4 and 5).

Since no other significant intraluminal lesions were found aside from the esophageal plaque, further imaging was performed. CT revealed a mildly enhancing mass in the left tonsil and multiple enlarged cervical lymph nodes.

A subsequent dermatologic examination identified a small, black, ulcerated lesion on the patient's back, which was confirmed to be the primary cutaneous melanoma. Treatment options were discussed, but due to his frailty, the patient declined further intervention.

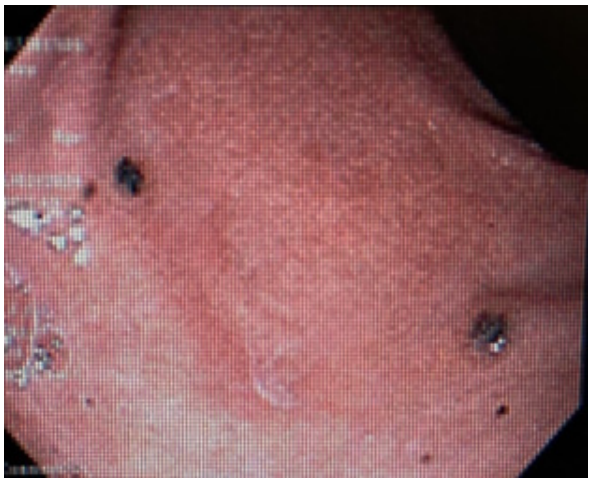

Figure 1. Melanotic lesions in the stomach found during EGD.

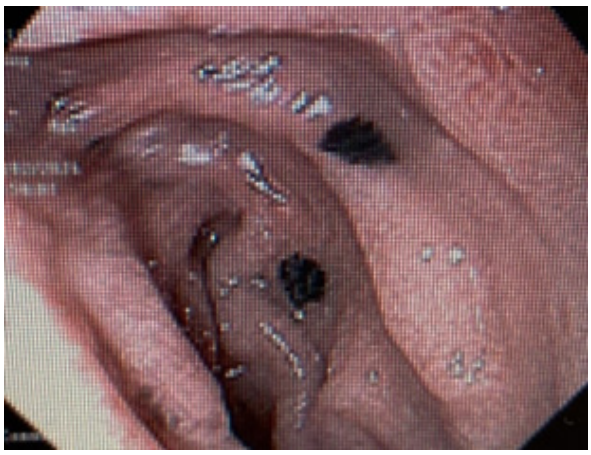

Figure 2. Melanotic lesions in the duodenum found during EGD.

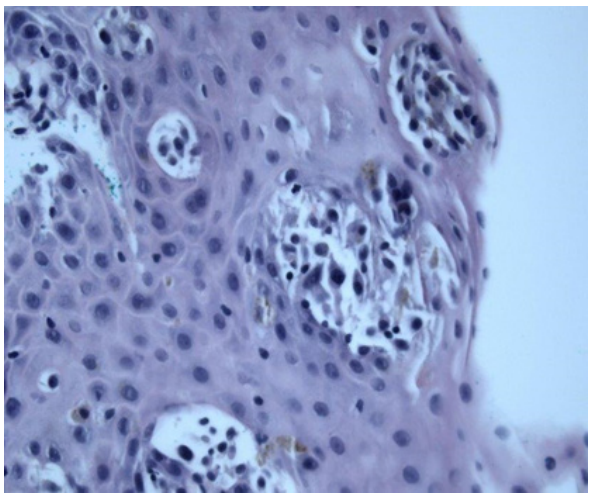

Figure 3. Histopathological analysis showed nests of melanocytes with atypical mitosis in the esophagus.

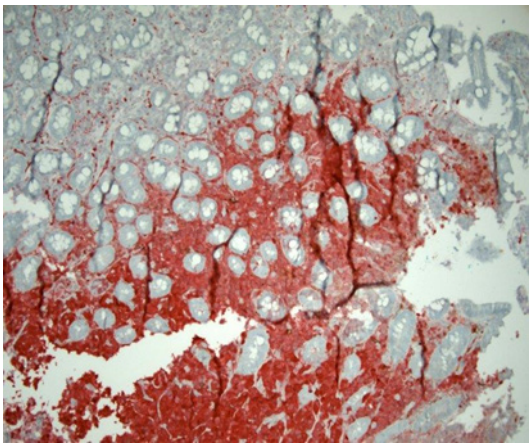

Figure 4. Immunohistochemical staining showed positivity for S100 in the duodenum.

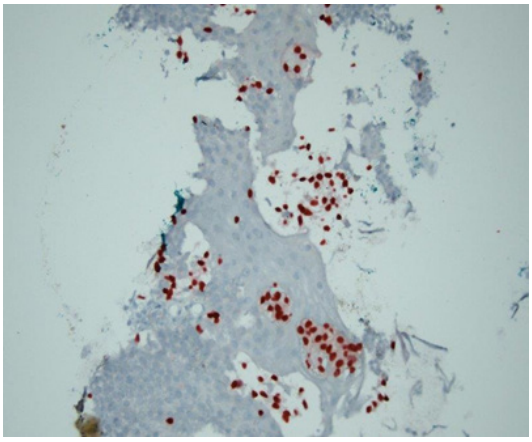

Figure 5. Immunohistochemical staining showed positivity for SOX-10 in the esophagus.

DISCUSSION

MM is a highly aggressive neoplasm with a high propensity for metastasis, which remains the leading cause of morbidity and mortality in affected patients. Visceral dissemination, particularly to the GI tract, is not uncommon but often underrecognized due to its non-specific presentation. Although only 1-5% of MM patients exhibit symptomatic GI involvement, autopsy series suggest a prevalence as high as 60%, underscoring the importance of maintaining a high index of suspicion in patients with GI symptoms and a history of MM.[2,5,6]

Among GI sites, the small intestine, particularly the jejunum and ileum, is most frequently involved. MM accounts for approximately 50-60% of all secondary malignancies to the small bowel.[3] The predilection for small bowel involvement may be mediated by high expression of the chemokine receptor CCR9 (C-C chemokine receptor 9) on melanoma cells, which facilitates migration to the small intestine via its ligand CCL25 (C-C motif chemokine ligand 25).[2,3,5] Other commonly affected GI organs include the stomach, duodenum, and colon, while esophageal involvement remains rare (~5%).[2,6] Liver metastases are among the most common visceral sites, seen in up to 77% of cases in autopsy studies, and particularly are common in uveal melanoma.[7,8]

Diagnosis of GI metastases from MM remains challenging. Symptoms often are vague and may include abdominal pain, melena, anemia, obstruction, or perforation. Imaging modalities such as CT and positron emission tomography (PET) are crucial for initial evaluation. However, standard CT has limited sensitivity (60-70%) for GI involvement.[2,4,9] CT enteroclysis may improve detection, and PET offers enhanced sensitivity and specificity. Endoscopic techniques, including EGD, colonoscopy, and capsule endoscopy, are invaluable for direct visualization and biopsy. Capsule endoscopy is particularly useful for detecting small bowel lesions but lacks biopsy capability, often necessitating balloon-assisted enteroscopy.[2,3,9]

Histopathologic analysis with immunohistochemistry remains the gold standard for diagnosis. Melanoma metastases may appear as melanotic or amelanotic, and may assume polypoid, infiltrative, cavitary, or exophytic forms.[3,6] In this case, the presence of mucosal lesions in the esophagus, stomach, and duodenum, confirmed by biopsy and immunostaining (positive for S100, SOX-10, and Mart-1; negative for pancytokeratin), established the diagnosis of metastatic melanoma.

This patient's case is notable for the timing of GI involvement, which preceded identification of the primary cutaneous lesion. Although uncommon, MM can present with GI metastasis before the primary lesion is recognized or even in the absence of an identifiable primary. This raises the diagnostic consideration of primary GI melanoma, a rare and controversial entity. Because melanocytes are not normally found in the mucosa of the stomach, small intestine, or colon, several theories have been proposed to explain primary GI melanoma, including origin from ectopic neural crest cells, Schwann cells, or APUD (Amine Precursor Uptake and Decarboxylation) Icells. However, most experts favor the theory that such lesions represent metastases from regressed or undiagnosed primary sites.[1-3,10]

In rare cases, melanoma may arise in the esophagus, accounting for 0.1-0.5% of primary esophageal tumors.[11] These are believed to originate from melanocytes present in the esophageal mucosa, potentially associated with esophageal melanocytosis.[12] Diagnostic criteria for primary GI melanoma include absence of cutaneous or mucosal lesions elsewhere, presence of a solitary tumor, and histological evidence of in situ melanoma or intramucosal melanocytes in adjacent tissue.[1,3] In our patient, the identification of a cutaneous lesion on the back, along with widespread visceral involvement, strongly supported metastatic disease from a cutaneous primary.

Therapeutic options for metastatic MM have evolved substantially with the introduction of immune checkpoint inhibitors (e.g., pembrolizumab) and targeted therapies (e.g., BRAF and MEK inhibitors). Surgical resection may provide palliative benefit or improved survival in cases of isolated GI metastases.[2,6,9] Despite these advancements, GI metastases often are associated with poor prognosis. In this case, the patient's frailty and advanced disease precluded further intervention, and he elected to pursue hospice care.

CONCLUSIONS

MM can metastasize to the GI tract, most commonly affecting the small intestine, but also the colon, stomach, and esophagus. Diagnosis relies on clinical suspicion, imaging, and biopsy for histopathologic confirmation. Although rare, primary GI melanoma remains a consideration and warrants further study to clarify its

pathogenesis. This case reinforces the importance of early recognition and a multidisciplinary approach in managing GI manifestations of MM.

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Keywords: Melanoma, melanocytes, Esophageal Metastasis, Esophageal Neoplasms, Duodenum, Stomach

Presentation: This work was previously published as an abstract and presented as a poster at the American College of Gastroenterology (ACG) Annual Meeting in October 2024, Philadelphia, PA.