Case Report

A Case Report: Primary Malignant Melanoma of the Jejunum

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Abstract

Background: Malignant melanoma represents only less than 3% of all malignant tumors in the gastrointestinal (GI) tract. Differentiating primary melanomas from metastatic melanoma can be difficult.

Case Presentation: We report the case of a 60-year-old woman who presented with signs of intestinal obstruction. An USG abdomen and KUB shows intussusception small bowel, and CT imaging revealed a mass in the proximal small intestine. Emergency laparotomy was performed, which identified an intraluminal tumor in the proximal jejunum. This tumor was surgically resected, followed by end-to-end anastomosis. Histopathological and immunohistochemical analyses confirmed malignant melanoma, with tumor cells testing positive for S-100, HMB-45, and Melan-A. A comprehensive postoperative evaluation—including whole-body PET-CT, dermatological, ophthalmological, and nodal assessments—showed no evidence of a primary cutaneous, mucosal, ocular, or nodal lesion, nor any additional metastases. These findings supported the diagnosis of primary jejunal melanoma, an exceedingly rare condition.

Outcome: The patient recovered without complications and was referred to medical oncology. She began adjuvant immunotherapy with nivolumab. At the 6-month follow-up, she remained clinically asymptomatic, with no evidence of recurrence observed in surveillance imaging.

Conclusion: In rare but significant instances of small intestinal obstruction, primary malignant melanoma of the small intestine is taken into consideration. The best results can only be obtained with early surgical intervention, a precise histological diagnosis, and prompt immunotherapy treatment.

Keywords: Malignant melanoma; Primary intestinal melanoma; Jejunum; Small bowel intussusception; Gastrointestinal melanoma; Metastatic melanoma; Immunohistochemistry (IHC); S100; HMB-45; Melan-A; Rare tumor; Histopathology.

Introduction

Melanoma develops from melanocytes, which are primarily present in the skin, eye choroid, meninges, and anal margin. Malignant melanoma accounts for less than 3% of all malignant tumors in the gastrointestinal tract. The bulk of these cancers are metastatic and originate from other primary lesions. Most malignant melanomas in the small intestine are metastases from cutaneous lesions, although they can also appear as primary tumors in the gastrointestinal (GI) tract. It is important to highlight that distinguishing between primary and metastatic melanomas can be difficult.

Case Report

We present a case of a 60-year-old female admitted to Sir T Hospital in May 2023 with clear signs of intestinal obstruction and an unremarkable past medical history.

Investigations: An USG abdomen and KUB shows intussusception small bowel, and CT imaging revealed a mass in the proximal small intestine. Given the urgency, an exploratory laparotomy was performed, revealing an intraluminal tumor in the proximal jejunum, which was resected, followed by end-to-end anastomosis.

Methodology: An 18 cm segment of the small bowel was received for examination. The proximal portion was dilated and invaginating into the distal segment, which appeared normal externally but was contracted and reddish internally. The tumor measured 5.5 cm × 3.5 cm × 1.2 cm and was positioned strategically within the bowel.

Tumor Characteristics: The tumor was exophytic, polypoidal, and blackish in appearance, with infiltration through the muscle layer and serosa into mesenteric fat. The remaining mucosa was unremarkable.

Lymph Node Findings: Four lymph nodes were examined, with the largest measuring 1 cm × 1 cm. Tissue samples were meticulously processed and stained for microscopic examination.

Microscopy: Sections show an infiltrating tumor composed of nests and lobules of epithelioid to spindle-shaped cells having round to oval nuclei, prominent nucleoli, and abundant eosinophilic cytoplasm. There is extensive melanin pigmentation and marked nuclear pleomorphism, with approximately 8–9 mitoses/10 HPF (3.6 mitoses/mm²).

Tumor extension–Through the serosa into mesenteric fat. Lymphovascular invasion–Present. Lymph node–4/4 involved. Perinodal extension–Present.

Postoperative Course and Management: Whole-body PET-CT scan: No evidence of cutaneous, mucosal, ocular, or nodal primary lesion; no additional metastases. Full dermatologic examination: Negative for suspicious pigmented lesions. Comprehensive ophthalmologic examination: No evidence of ocular melanoma. The patient had an uneventful recovery. Given the histologically confirmed primary malignant melanoma of the jejunum and intermediate mitotic activity, the patient was referred to medical oncology. Adjuvant therapy: Initiated on nivolumab. At 6-month follow-up, the patient remains clinically disease-free, with no recurrence on surveillance imaging.

Impression: Tumor Site: Jejunum, Histologic Type: Malignant Melanoma, Immunohistochemistry (IHC): S100: Positive, HMB-45: Positive, Melan-A: Positive, Ki-67: 20% (moderate proliferative index).

Table 1: Immunohistochemistry Results

Marker	Clone / Source	Epitope Retrieval	Detection Method	Result
S100 (Anti S100-B)	Clone 15E2E2; BioGenex	EZ-AR2 Elegance buffer	Super Sensitive Polymer-HRP/DAB	Strong nuclear & cytoplasmic
HMB-45	Clone HMB-45; BioGenex	EZ-AR2 buffer	Super Sensitive Polymer-HRP/DAB	Strong cytoplasmic
Melan-A	Clone A103; BioGenex	EZ-AR2 buffer	Polymer-HRP with DAB	Strong cytoplasmic
Ki-67	Clone MIB-1; Dako	EZ-AR1 buffer	Polymer-HRP with DAB	∼20% nuclear positivity

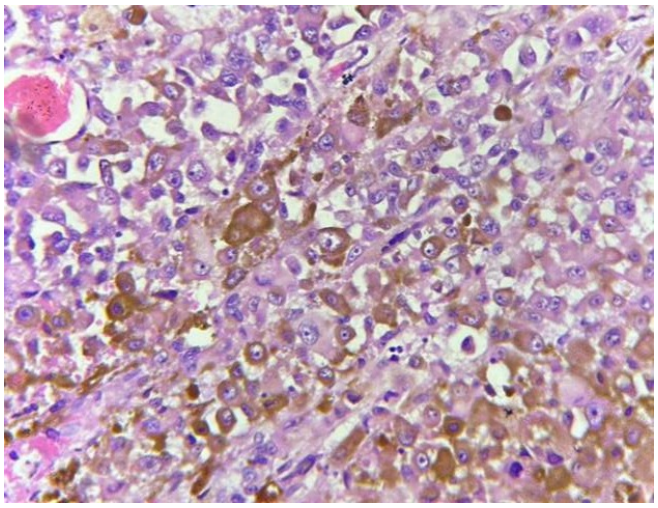

Figure 1: Malignant cell proliferation with melanin pigmentations (40x view).

Discussion

Primary malignant melanoma of the small intestine is a rare and diagnostically challenging entity. It typically presents with nonspecific symptoms such as abdominal pain, gastrointestinal bleeding, anemia, or intestinal obstruction, often due to complications like intussusception [3, 4]. While the small bowel is a common site for metastatic melanoma, true primary melanomas in this location are extremely uncommon [1, 2]. Theories regarding their origin include the presence of ectopic melanocytes derived from neural crest cells that migrate during embryogenesis [1, 2]. Diagnosis relies heavily

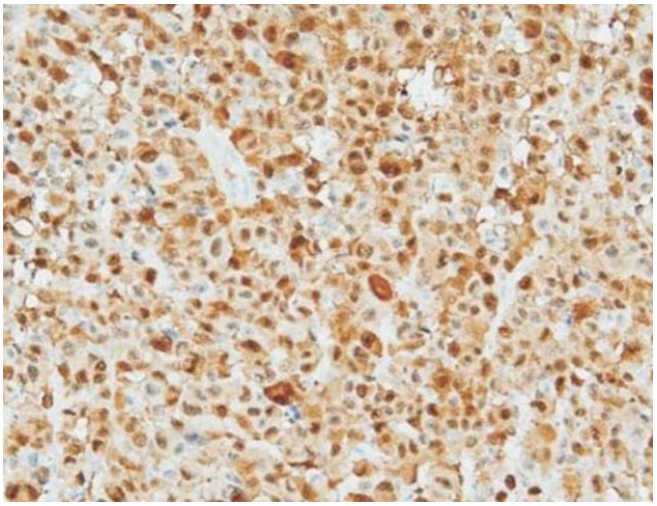

Figure 2: Tumor cells are positive in IHC staining for S100.

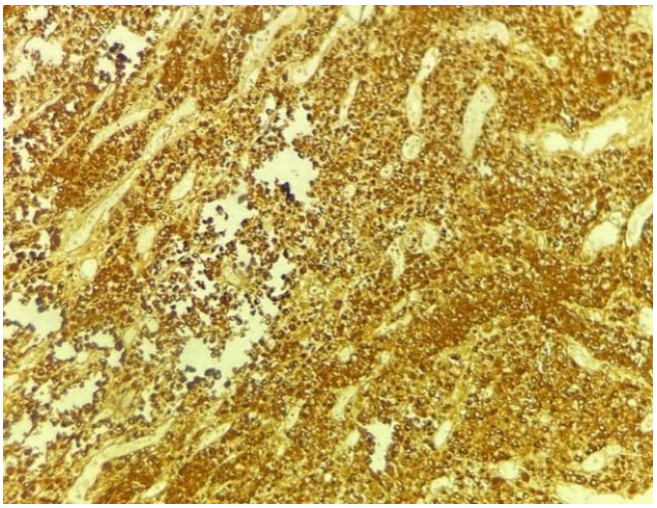

Figure 3: Tumor cells are positive in IHC staining for HMB 45.

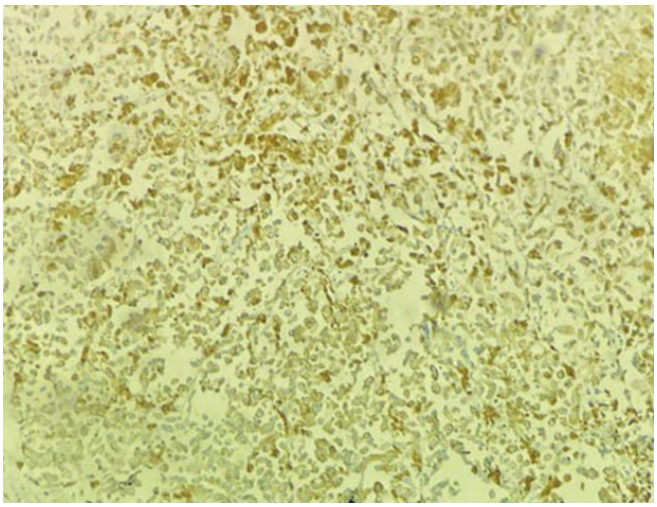

Figure 4: Tumor cells are positive in IHC staining for Melan A.

on histopathological evaluation and immunohistochemical staining for melanocytic markers such as S-100, HMB-45, and Melan-A [5, 6]. However, establishing a lesion as primary rather than metastatic requires exclusion of a primary cutaneous, ocular, or mucosal site through detailed clinical evaluation and imaging, often supported by PET/CT [2, 6]. Complete surgical resection remains the cornerstone of treatment, with adjuvant immunotherapy increasingly considered, though prognosis remains guarded due to the tumor's aggressive behavior [1, 5, 6].

A critical aspect of diagnosing primary small intestinal melanoma involves differentiating it from other mimicking tumors, particularly clear cell sarcoma (CCS). Both tumors share overlapping immunohistochemical profiles, including expression of S-100 and melanocytic markers, which can lead to diagnostic confusion [6]. However, CCS often affects younger patients and typically arises in the soft tissues of extremities rather than within the gastrointestinal tract [6]. Unlike melanoma, CCS lacks junctional activity and melanin pigmentation, and it is characterized by a specific chromosomal translocation involving the EWSR1 gene (most commonly EWSR1-ATF1), detectable through molecular testing [6]. Additionally, careful exclusion of cutaneous melanoma is essential, as intestinal metastases may occur from a regressed or undetected primary skin lesion [2, 7]. Thus, a combination of thorough clinical workup, histopathology, immunohistochemistry, and in selected cases, molecular analysis, is required to establish a confident diagnosis of primary intestinal melanoma [1, 6].

Conclusion

The gross pathological features, microscopic evaluation, and immunohistochemical (IHC) analysis are consistent with a diagnosis of malignant melanoma. Primary malignant melanoma of the small intestine, particularly involving the jejunum, is an extremely rare neoplasm. Favorable clinical outcomes depend on early surgical management, accurate histopathological diagnosis, and timely initiation of immunotherapy.

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Informed Consent Written informed consent was obtained from the patient for the publication and presentation of this case, including all relevant clinical data and images.

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Conflicts of Interest The authors declare no conflicts of interest related to this work.

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