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Primary Intestinal Melanoma: An Exceptional Case Report

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Abstract **Case Report**

Primary melanoma of the small bowel is an exceptionally rare and aggressive entity, often difficult to distinguish from metastatic disease and frequently diagnosed at an advanced stage. A 17-year-old patient presented with acute diffuse abdominal pain, fever, and vomiting. On admission, he was hemodynamically unstable with signs of generalized peritonitis. Laboratory tests showed leukocytosis, elevated inflammatory markers, and severe anemia. Abdominal CT revealed pneumoperitoneum, abundant peritoneal fluid, and a small bowel mass. Emergency surgery identified a perforated 10 cm small bowel tumor located 60 cm distal to the first jejunal loop, with generalized peritonitis. Segmental resection with primary anastomosis was performed. Histopathological and immunohistochemical analysis confirmed malignant melanoma. Despite immunotherapy, the patient developed diffuse peritoneal carcinomatosis and died six months later. Primary small bowel melanoma is a rare condition that may present with acute complications such as perforation. Prognosis remains poor despite surgical and systemic treatment.

Keywords: Intestinal melanoma, primary, surgery, case report.

INTRODUCTION

Primary melanoma of the small bowel is an exceptionally rare and controversial entity, as the gastrointestinal tract is more commonly involved by metastatic spread from cutaneous melanoma [1]. Distinguishing primary from secondary lesions remains challenging, and its pathogenesis is not fully understood. Clinical presentation is often non-specific or revealed by acute complications such as obstruction or perforation, leading to delayed diagnosis. Confirmation relies on histopathological and immunohistochemical analysis, and prognosis remains poor due to the aggressive nature of the disease [1, 2]. We report a case of primary small bowel melanoma presenting with generalized peritonitis secondary to tumor perforation.

CASE PRESENTATION

A 17-year-old patient with no significant past medical history presented to the emergency department with a three-day history of diffuse abdominal pain associated with fever and vomiting. On admission, the patient was hemodynamically unstable, with tachycardia at 120 beats per minute, blood pressure of 80/50 mmHg, and a temperature of 38.5°C. Physical examination revealed generalized abdominal guarding without other notable findings. Laboratory investigations demonstrated leukocytosis (19,000/mm³), elevated C-reactive protein (285 mg/L), and severe anemia with a hemoglobin level of 6 g/dL. Contrast-enhanced abdominal computed tomography revealed abundant intraperitoneal fluid, pneumoperitoneum, and a mass arising from the small intestine, consistent with a perforated small bowel lesion.

Following hemodynamic stabilization and blood transfusion, the patient was transferred to the operating room for emergency surgery. Intraoperative exploration revealed a diffuse blackish discoloration of the peritoneal cavity associated with generalized peritonitis. A perforated small bowel mass measuring approximately 10 cm in diameter was identified 60 cm distal to the first jejunal loop. Extensive peritoneal lavage was performed, followed by segmental small bowel resection with primary anastomosis.

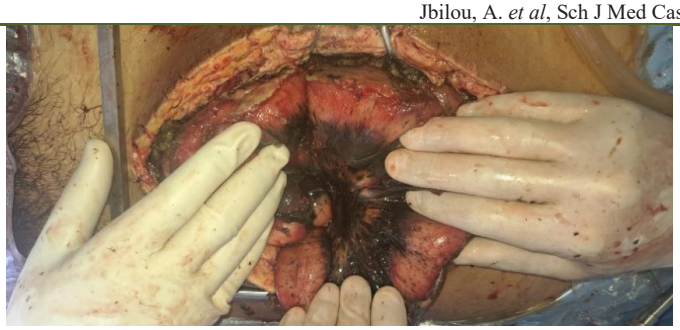

Figure 1: Intra operative image showing peritonitis with blackish discoloration

Figure 2: Intra operative image showing a perforated intestinal tumor

Histopathological and immunohistochemical examination of the resected specimen confirmed the diagnosis of malignant melanoma [figure 3]. The immediate postoperative course was uneventful. The patient subsequently received immunotherapy; however, follow-up computed tomography demonstrated diffuse peritoneal carcinomatosis. The patient died six months after surgery.

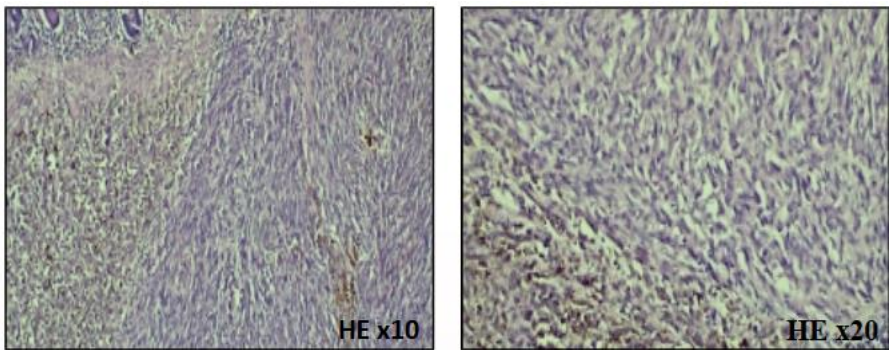

Figure 3: Histological sections (H&E, ×10 and ×20) demonstrate a malignant spindle cell neoplasm characterized by hyperchromatic nuclei, eosinophilic cytoplasm, and diffuse melanin pigment deposition

DISCUSSION

Primary intestinal melanoma is an exceptionally rare malignancy, representing a small subset of mucosal melanomas, which themselves account for less than 2% of all melanoma cases. Recent population-based analyses estimate its incidence at approximately 0.5–0.6 per million individuals, underscoring its rarity and the consequent diagnostic challenges [1,2]. The gastrointestinal tract is more commonly affected by metastatic spread from cutaneous melanoma than by primary disease, making the distinction between primary and secondary lesions critical yet often difficult.

The pathogenesis of primary intestinal melanoma remains controversial. Contemporary hypotheses suggest aberrant migration of neural crest-derived melanocytes during embryogenesis or differentiation of pluripotent stem cells within the intestinal mucosa [3]. Despite ongoing debate, diagnostic criteria continue to rely on clinicopathological correlation, including the absence of a known primary cutaneous lesion, lack of other metastatic sites at diagnosis, and the presence of a solitary intestinal tumor [4]. In the present case, these criteria were satisfied, supporting the diagnosis of a primary lesion.

Clinically, primary intestinal melanoma presents with nonspecific symptoms such as abdominal pain, gastrointestinal bleeding, anemia, or bowel obstruction. These vague manifestations often result in delayed diagnosis, as highlighted in recent case series and reviews [5,6]. Advances in imaging, particularly the use of contrast-enhanced computed tomography and positron emission tomography (PET-CT), have improved the detection and staging of gastrointestinal melanomas and are essential for excluding an extraintestinal primary site [7].

Histopathological examination remains the cornerstone of diagnosis. Tumors typically demonstrate atypical melanocytic proliferation, with or without melanin pigmentation. Immunohistochemical staining is indispensable, with markers such as S100, HMB-45, and Melan-A consistently reported as highly sensitive for melanocytic differentiation [3,8]. These tools are particularly valuable in amelanotic variants, where morphology alone may be misleading.

Surgical resection continues to be the mainstay of treatment for localized disease and is associated with improved symptom control and potential survival benefit [6]. However, even with complete resection, the prognosis remains poor due to the aggressive nature of the disease and the high risk of early dissemination. Recent data suggest a 3-year survival rate of approximately 20–30%, reflecting the unfavorable prognosis compared to cutaneous melanoma [2].

The therapeutic landscape of melanoma has evolved significantly with the advent of immune checkpoint inhibitors. Agents such as Nivolumab and Pembrolizumab have demonstrated substantial survival benefits in advanced melanoma. Although most evidence derives from cutaneous melanoma, recent studies suggest that mucosal melanomas, including gastrointestinal forms, may also benefit from these therapies, albeit with generally lower response rates [9,10]. Ongoing research into the molecular characteristics of mucosal melanoma has revealed distinct genetic profiles compared to cutaneous melanoma, which may partly explain differences in therapeutic response and highlights the need for tailored treatment strategies [10].

Distinguishing between primary and metastatic melanoma remains difficult. According to Sachs *et al.,*[4], a lesion may be considered primary if it is solitary, if no distant metastases are present other than locoregional lymph nodes, and if the patient remains disease-free for at least 12 months after diagnosis. These criteria reflect the fact that most patients with metastatic melanoma die within a year, that median survival after metastasectomy is about 15 months, and that metastatic disease usually involves multiple lesions.

CONCLUSION

Overall, primary intestinal melanoma remains a rare and aggressive neoplasm with significant diagnostic and therapeutic challenges. Early recognition, accurate differentiation from metastatic disease, and complete surgical resection are crucial for improving outcomes. The integration of modern imaging techniques and immunotherapy offers promising avenues for management, but further studies are needed to establish standardized treatment protocols and to better understand the biology of this uncommon malignancy.

Patient perspective: The procedure of surgery was explained to the patient with all advantages and possible complications. And it was agreed.

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Author's contribution:
Aymane Jbilou: Writing, review and editing of the manuscript.
Issam Yazough, Rajae Alkouh, Omar Bennour, Rabie soultana: Contributed for diagnose and treatment of the patient.
Younes Aggouri, Said Aitlaalim: Review, Supervision of treatement.

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