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JCIMCR OPEN ACCESS Journal of Clinical Images and Medical Case Reports

ISSN 2766-7820

Case Report

Open Access, Volume 6

Primary malignant melanoma of the colon: A rare and challenging diagnosis

Tafaj Irgen[1]*; Allko O[1]; Shehi D[2]

[1]University Hospital Center "Mother Teresa", Department of Gastroenterology/Hepatology, Tirana, Albania.

[2]University Hospital Center "Mother Teresa", 1st Department of General Surgery, Tirana, Albania.

***Corresponding Author: Tafaj Irgen**
University Hospital Center "Mother Teresa",
Department of Gastroenterology/Hepatology,
Tirana, Albania.
Email: irgentafaj@yahoo.com

Received: Jun 04, 2025
Accepted: Jul 04, 2025
Published: Jul 11, 2025
Archived: www.jcimcr.org

DOI: www.doi.org/10.52768/2766-7820/3680

Abstract

Primary malignant melanoma of the colon is an exceedingly rare clinical entity, accounting for less than 1% of all gastrointestinal melanomas. Its diagnosis is challenging due to nonspecific symptoms and the need to exclude more common metastatic disease. We report the case of a 72-year-old male who presented with rectorrhagia, fatigue, and severe anemia. Colonoscopy revealed a large polypoid, ulcerated lesion in the ascending colon. Imaging showed colo-colic intussusception without evidence of metastasis. Histopathological examination confirmed malignant melanoma, with immunohistochemistry positive for S-100 and PRAME and a high Ki-67 index (50%). A thorough clinical and ophthalmologic evaluation ruled out any other primary site. Persistent gastrointestinal bleeding necessitated a semi-urgent right hemicolectomy, which confirmed the diagnosis. The postoperative course was uneventful, and the patient was referred to oncology for further management. Primary colonic melanoma is a rare but important differential diagnosis for large bowel tumors, particularly in the absence of a known primary site. Accurate diagnosis requires histological confirmation and exclusion of metastatic disease. Surgical resection remains the cornerstone of treatment, with adjuvant therapies under consideration in selected cases.

Introduction

Melanoma is a malignant tumor originating from melanocytes, primarily affecting the skin and rarely mucosal membranes, eyes, and less commonly, internal organs, with less or none at all sun exposure [1,2]. Colonic melanoma is extremely rare. Most melanomas of the colon are metastatic, arising from a cutaneous primary site [3]. Primary Colonic Melanoma (PCM) represents a diagnostic and therapeutic challenge due to its rarity, nonspecific clinical presentation, and histological similarities with other colonic tumors. In the evaluation to exclude a primary melanoma at another site, the diagnostic approach should involve a thorough detailed medical history and a comprehensive physical examination, with particular attention to dermatologic, ophthalmologic, and genitourinary assessments. Primary gastrointestinal (GI) melanomas are rare, accounting for approximately 2% of all mucosal melanomas [1]. Within the spectrum of primary GI melanomas, lesions are more commonly located in mucosal regions that naturally contain melanocytes. Reported sites include the pharynx (32.8%), anal canal (31.4%), rectum (22.2%), esophagus (5.9%), stomach (2.7%), small intestine (2.3%), gallbladder (1.4%), and colon (0.9%) (4). Primary melanoma of the colon is an exceptionally rare entity, with only 44 cases documented in the literature till now [5-9].

Case presentation

A 72-year-old male presented to the Emergency Department with massive rectorrhagia, fatigue, generalized weakness, and severe anemia. He had been experiencing these symptoms for

the past three months, with the onset of rectorrhagia occurring during the last ten days. The patient had been taking oral iron supplements for anemia. He had a medical history of atrial fibrillation and was receiving anticoagulant therapy. There was no prior history of malignancy reported in his personal or family medical history. The patient indicated that his symptoms had worsened in the recent days. He was admitted to the Department of Gastroenterology, and after hemodynamic stabilization and multiple blood transfusions, a colonoscopy was performed. The procedure revealed a large subocclusive polypoid lesion with ulcerated and necrotic areas in the ascending colon. A biopsy specimen was obtained and referred to the Pathology Department for further evaluation. Tumor markers, including cancer antigen 19-9 (CA 19-9), Carcinoembryonic Antigen (CEA), and alpha-fetoprotein (AFP), were evaluated and all of them resulted within normal limits. After that, a contrast-enhanced CT scan was performed, revealing a "pseudokidney" sign in the supraumbilical region near the abdominal wall (Figure 1), suggestive of a colo-colic intussusception (Figure 2). No additional suspicious lesions indicative of malignancy was identified. Furthermore, no evidence of a primary tumor was found on physical examination or total body CT imaging.

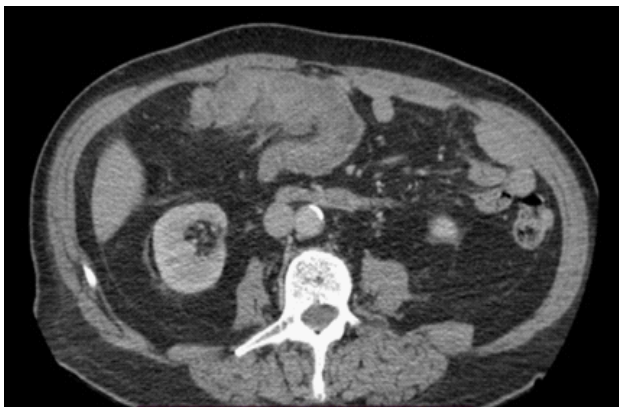

Figure 1: Axial CT scan with IV contrast showing "pseudokidney" sign.

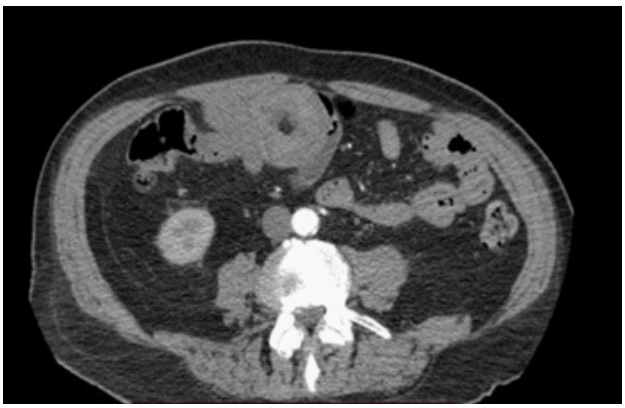

Figure 2: Axial CT scan with IV contrast showing an image in favor of colo-colic intussusception.

The pathology department confirmed the diagnosis of colonic melanoma. Histopathological analysis revealed a tumor composed of epithelioid cells exhibiting marked atypia and arranged in a syncytial growth pattern. The neoplastic cells demonstrated high nucleocytoplasmic ratios, with 2-3 prominent nucleoli, and variable morphology ranging from polygonal to spindle-shaped in certain areas. Immunohistochemical (IHC) staining showed the following profile: CK AE1/AE3 --- (negative), CD56--- (negative), S-100 (strongly positive +++), Actin--- (negative), CD117-- (negative), PRAME (strongly positive +++), Ki67 (proliferation index 50%), and Melan A--- (negative). These findings strongly support the diagnosis of a colonic melanoma. A BRAF mutation analysis was also performed and had a negative result. The cutaneous and mucosal inspection did not find any other lesion compatible with primary melanoma. No previously excised skin melanoma was reported from the patient. After the patient was examined by an ophthalmologist and ocular melanoma was ruled out, the diagnosis of primary ascending malignant melanoma of the colon, was confirmed. During hospitalization, the patient experienced ongoing uncontrolled gastrointestinal bleeding for several days, and his anemia remained unresponsive despite multiple blood transfusions. The multidisciplinary team consultation decided an emergency surgical treatment. The patient underwent semi-urgent right hemicolectomy (Figure 3) with an uneventful postoperative period.

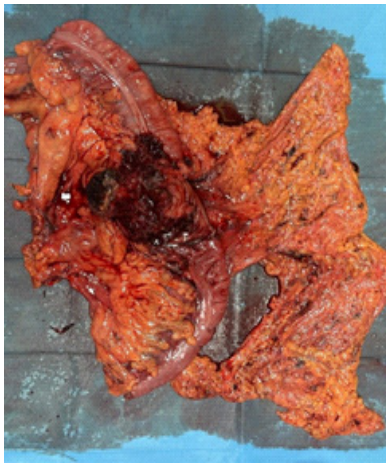

Figure 3: Right hemicolectomy.

The operative specimen, reconfirmed the diagnosis of colonic melanoma. The tumor consists of epithelioid cells with pronounced atypia that grow in a syncytial manner and infiltrate the intestine from mucosa to peri-intestinal adipose tissue. The cells have increased nucleoplasm ratios, with 2-3 evident nucleoli and the morphology varies from polygonal to spinal in some areas. Post-discharge the patient has been recovering well at home and he was referred to the Oncological Department for further evaluation and management.

Discussion/conclusion

Primary gastrointestinal melanoma most commonly presents with abdominal pain and weight loss, which contrasts with the typical presentation of metastatic GI melanoma, where symptoms such as gastrointestinal bleeding, bowel obstruction, and abdominal pain are more frequently observed [2,5,9,10]. The average age at diagnosis is approximately 60 years, with no significant gender tendency. According to Ozdemir, mucosal melanomas are defined by the following criteria: [1] the presence of a solitary lesion within the affected tissue, [2] absence of a prior ocular melanoma, [3] no history of cutaneous melanoma, [4] histopathologic and clinical features consistent with a primary tumor, and [5] absence of other melanoma lesions at the time of surgical intervention. In our case, the patient had no prior history of melanoma, presented with a solitary lesion

presenting with histological characteristics typical of a primary melanoma, and no additional melanomas were identified during surgery. These findings demonstrate the importance of a multidisciplinary approach in establishing the diagnosis and guiding the management of primary colonic melanoma. The occurrence of primary melanoma in the colon is extremely rare, and its pathogenesis remains poorly understood. Several hypotheses have been proposed to reveal its origin. One such theory—the ectodermal differentiation theory-suggests that neural crest cells may abnormally migrate to the colon during embryologic development. These cells, capable of melanin production, may later undergo malignant transformation, resulting in melanoma. However, this theory has only been validated in vitro [5]. Another hypothesis posits that melanocytes from the anorectal region or undifferentiated progenitor cells may migrate to the colon, subsequently giving rise to melanocytic neoplasms [5]. In this context, histopathological evaluation plays a critical role in distinguishing primary from metastatic melanoma. Primary melanomas typically demonstrate pagetoid spread of melanocytes within the superficial epithelium, atypical or malignant melanocytes in the basal layer, and in-situ histological changes. Conversely, metastatic melanoma is characterized by dermal lymphocytic infiltration with melanophages, absence of atypical melanocytes, evidence of vasculogenesis, and fibrotic stromal changes. Histologically, the diagnosis of primary colonic melanoma is usually best supported by the results of specific immunohistochemistry staining with positivity for S-100 being highly sensitive (90%) [2,9]. Currently there is no standard of care for treatment. Surgical resection with wide margins of primary colonic melanoma that has not metastasized has been suggested as a viable treatment option, and the treatment that we decided upon [11]. Following surgery, the patients must be referred to the Department of Oncology for further evaluation and consideration of targeted immunotherapy. Referral to medical oncology is particularly prudent in cases of mucosal melanoma, as PD-1 inhibitors such as pembrolizumab have demonstrated clinical efficacy, achieving response rates of approximately 23% and contributing to prolonged progression-free survival [12].

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