Research Article

Amelanotic Melanoma of The Rectum Presenting As A Polypoid Lesion: A Case Report

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Abstract

Background: Primary anorectal melanoma is a rare and aggressive malignancy, accounting for a small fraction of all melanomas and often posing a diagnostic challenge when amelanotic, as it can mimic benign or epithelial lesions both endoscopically and histologically. **Case Presentation:** We report the case of a 75-year-old man who presented with rectal bleeding. Colonoscopy revealed a polypoid mass with ill-defined, ulcerated borders; the pedicle was macroscopically non-infiltrated. The lesion was removed by submucosal (endoscopic) polypectomy. Histopathological examination showed a poorly differentiated malignant neoplasm without visible melanin pigment. Immunohistochemistry demonstrated positivity for SOX10, vimentin, HMB-45, and CD10, with a high Ki-67 proliferation index, and negativity for pancytokeratin and CD45, confirming the diagnosis of amelanotic melanoma. Staging CT showed no distant lesions, and the patient underwent wide surgical excision of the rectum, with no metastatic involvement of the regional lymph nodes. The patient subsequently traveled abroad for further postoperative treatment, limiting the availability of long-term follow-up data.**Conclusion:** This case underscores the importance of maintaining a high index of suspicion for melanoma in anorectal polypoid lesions, particularly when routine histology is inconclusive, and highlights the essential role of a targeted immunohistochemical panel in reaching the correct diagnosis.

Keywords— Amelanotic Melanoma, Anorectal Melanoma, Rectal Polyp, SOX1 0, HMB-45, Polypectomy.

I. INTRODUCTION

Anorectal melanoma is a rare malignancy, representing approximately 0.4–1 .6% of all malignant melanomas and roughly 1 % of anal canal cancers, arising from melanocytes normally present at the anorectal transition zone [1 –4]. Unlike cutaneous melanoma, anorectal melanoma frequently lacks classic pigmentation — the so-called "amelanotic" variant — which substantially complicates both clinical and pathological diagnosis and is a recognized cause of misdiagnosis as a benign polyp, adenocarcinoma, or other epithelial lesion [5–7]. Patients often present late, most commonly with rectal bleeding, and the disease carries a poor prognosis, with reported 5-year survival rates of only 6–22% even after radical surgery [2,3]. We describe a case of primary amelanotic melanoma of the rectum, initially resected as a presumed benign polyp, with emphasis on the immunohistochemical workup that established the diagnosis.

II. CASE PRESENTATION

A 75-year-old man presented with rectal bleeding. Past medical history was notable for GI pathology and the physical examination did not find anything specific. Colonoscopy identified a polypoid mass with ill-defined, ulcerated borders. The pedicle was macroscopically non-infiltrated, and the lesion showed no evident pigmentation, an appearance without the features typically expected of a melanocytic lesion. Given its endoscopic appearance, it was managed as a routine polyp and removed by submucosal endoscopic polypectomy. The margins were clear from the lesion.

III. PATHOLOGICAL FINDINGS

1) Macroscopy

The specimen consisted of a polypoid fragment measuring 3cm, the surface was disordered, ulcerated and cross-sectional area was white with hemorrhage and has a crumbly consistency.

2) Microscopy

Histological sections showed a submucosal proliferation of atypical epitheloid cells with diffuse architecture. The cells are large with eosinophilic cytoplasm with vesicular nuclei, prominent eosinophilic nucleoli, and frequent mitotic figures and atypical one. No intracytoplasmic melanin pigment was identified, consistent with an amelanotic phenotype. The lesion infiltrate and ulcerated the rectal mucosea and the lesion was not spread through the pedicle. We did not find vascular infiltration and the submucosal surgical margin was free from the tumor. [Figure 1]

3) Immunohistochemistry

The lesion did not present reactivity for Pancytokeratin (AE1/AE3) and CD45. On the S100 [Figure2] stain it was strongly positive, Vimentin stain was strongly positive, and it present a high reactivity for SOX10 and HMB45. This immunoprofile excludes an epithelial origin (pancytokeratin negative), excludes lymphoma (CD45 negative), and confirms melanocytic differentiation (SOX10 [Figure 3] and HMB-45 [Figure4] positivity), with a high proliferative index reflecting an aggressive biological behavior. The combined findings were

diagnostic of amelanotic malignant melanoma.

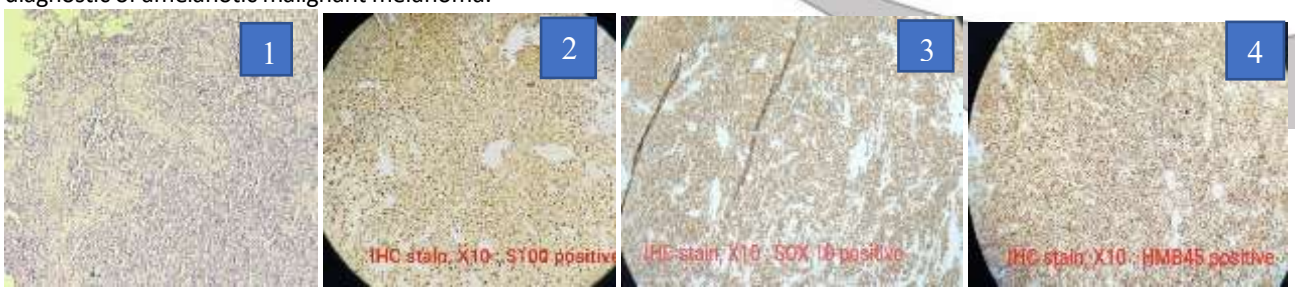

Figure: 1- HE stain x4: diffuse proliferation of large non pigmented atypical cells, ulcerating the mucosae. **2**- IHC stain, S100x4 – strongly positive ; **3**- IHC stain, SOC10x4- strongly positive ; **4**- IHC stain, HMB45x4 – strongly positive

IV. Staging, Treatment, and Follow-up

Staging computed tomography (CT) did not identify any distant lesions. Following multidisciplinary discussion, the patient underwent wide surgical excision of the rectum. Histopathological examination of the resection specimen showed no metastatic involvement of the regional lymph nodes. The patient subsequently traveled abroad to continue postoperative/adjuvant treatment; further clinical details and follow-up outcome were therefore not available to the reporting team.

V. Discussion

Anorectal melanoma is an aggressive malignancy with a poor prognosis, with reported 5-year survival rates typically below 22%, largely due to early lymphatic and hematogenous dissemination and frequent delayed diagnosis [2,3]. The amelanotic variant is particularly prone to misdiagnosis, as it can mimic adenocarcinoma, lymphoma, gastrointestinal stromal tumor, or a benign polyp both grossly and on routine hematoxylin and eosin staining — a pattern well documented across amelanotic melanomas at other anatomical sites [5–7]. Immunohistochemistry is essential in this differential. SOX1 0 and HMB-45 are sensitive and relatively specific markers of melanocytic differentiation, including in spindle-cell and poorly pigmented variants [8–1 0]; vimentin supports a mesenchymal/non-epithelial phenotype; and the combination of pancytokeratin negativity (excluding carcinoma) with CD45 negativity (excluding lymphoma) narrows the differential decisively toward melanoma. CD1 0 positivity, while less specific on its own, has also been reported to correlate with tumor progression and unfavorable prognosis in malignant melanoma, supporting its inclusion in the diagnostic panel [1 1 , [1 2]. A high Ki-67 index reflects the typically high proliferative activity of anorectal melanoma and correlates with its aggressive clinical course.

This case highlights two important lessons: first, that anorectal polypoid lesions — even those without visible pigmentation — should be regarded with suspicion when histology is atypical, and second, that a broad immunohistochemical panel is indispensable when the differential includes melanoma, carcinoma, and lymphoma, given the divergent management implications of each.

A limitation of this report is the incomplete long-term follow-up, as the patient relocated abroad to continue postoperative care; this precluded documentation of adjuvant treatment details and survival outcome beyond the immediate postoperative period. This is not uncommon in case reports of patients who seek care across borders and should be transparently acknowledged rather than obscured.

VI. Conclusions

Amelanotic anorectal melanoma is a rare but aggressive entity that can be readily mistaken for a benign polyp on endoscopy. This case reinforces the value of routine histopathological and immunohistochemical evaluation of all resected anorectal polyps, and illustrates a diagnostic immunoprofile (SOX1 0+, HMB-45+, vimentin+, CD1 0+, high Ki-67, pancytokeratin–, CD45–) that should prompt urgent recognition and staging given the aggressive natural history of this disease.

Acknowledgement

Nothing to declare.

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