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(CASE REPORT)

Primary rectal melanoma with CD117 expression: A case report and review of the literature

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Abstract

Primary rectal melanoma is a rare and aggressive malignancy, often diagnosed at an advanced stage. We report a case of an 84-year-old woman presenting with rectal bleeding, found to have a lower rectal tumor extending to the anal canal with nodal involvement on imaging. Histological examination revealed an amelanotic malignant proliferation composed of epithelioid and spindle cells. Immunohistochemistry showed diffuse SOX10 and S100 expression, with focal HMB45 and Melan-A positivity. Notably, tumor cells expressed CD117, raising the differential diagnosis of gastrointestinal stromal tumor, which was excluded by negative DOG1 and CD34 staining. This case highlights a major diagnostic pitfall and underscores the importance of a comprehensive immunohistochemical panel in undifferentiated anorectal tumors.

Keywords: Rectal melanoma; Amelanotic melanoma; CD117; Diagnostic pitfall; Immunohistochemistry

1. Introduction

Primary rectal melanoma is a rare malignancy representing less than 1% of anorectal tumors and is associated with poor prognosis due to delayed diagnosis and early dissemination. Amelanotic variants are particularly challenging, often mimicking other malignancies. We report a case illustrating a significant diagnostic pitfall related to CD117 expression.

2. Case report

An 84-year-old woman presented with terminal rectal bleeding evolving over two months.

Rectosigmoidoscopy revealed an ulcerated lesion of the lower rectum with hypertrophic, blackish margins. Computed tomography demonstrated circumferential wall thickening involving the lower rectum and anal margin. Pelvic magnetic resonance imaging (MRI) showed a tumor of the lower rectum extending to the mid-rectum and anal canal, with intermediate signal intensity on T2-weighted images and moderate enhancement after contrast administration (Figure 1-2). The lesion was staged as T4bN2bMx. Positron emission tomography revealed intense hypermetabolic activity of the anal canal tumor associated with bilateral pararectal hypermetabolic lymph nodes.

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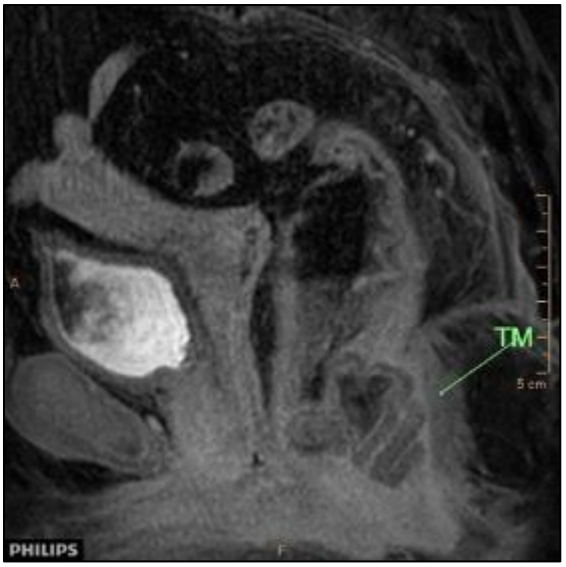

Figure 1 Pelvic MRI, T2-weighted sagittal image. Circumferential wall thickening of the distal rectum extending to the anal canal

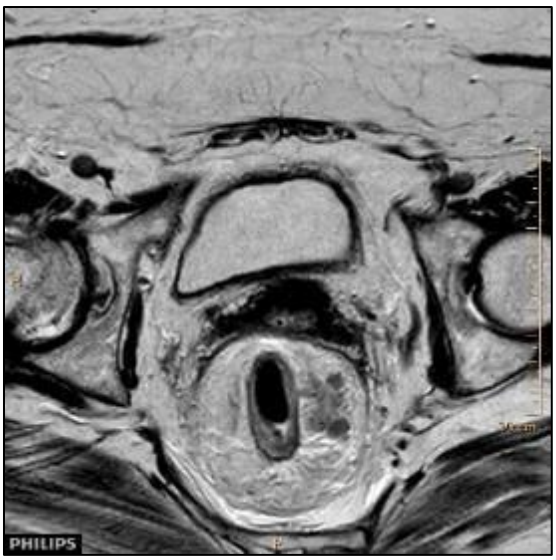

Figure 2 Pelvic MRI, T2-weighted axial image. Irregular circumferential thickening of the distal rectum with mesorectal fat infiltration and multiple enlarged mesorectal lymph nodes

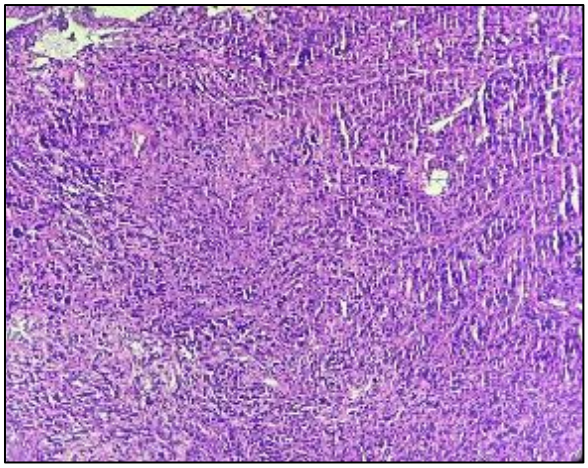

Figure 3 Malignant spindle and epithelioid cell proliferation arranged in sheets and fascicles (H&E, ×100)

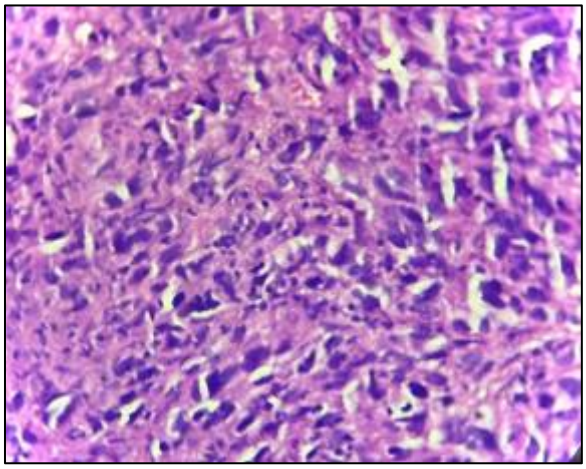

Figure 4 Marked cytological atypia with pleomorphic nuclei and prominent nucleoli (H&E, ×400)

Histological examination of the biopsy specimens revealed an infiltrative malignant neoplasm involving the rectal mucosa, composed of sheets and fascicles of atypical epithelioid and spindle cells (Figure 3). Tumor cells exhibited marked nuclear pleomorphism with coarse chromatin and prominent nucleoli, along with a high mitotic index (Figure 4). No melanin pigment was identified, supporting an amelanotic variant.

Immunohistochemical analysis demonstrated diffuse nuclear positivity for SOX10 (Figure 5) and strong nuclear and cytoplasmic expression of S100 protein. HMB45(Figure 6) and Melan-A showed focal cytoplasmic expression. Tumor cells also exhibited membranous and cytoplasmic staining for CD117 (Figure 7), raising the differential diagnosis of gastrointestinal stromal tumor. However, DOG1 (Figure 8) and CD34 were negative, excluding this possibility.

Based on the morphological and immunohistochemical findings, a diagnosis of primary amelanotic rectal melanoma was established.

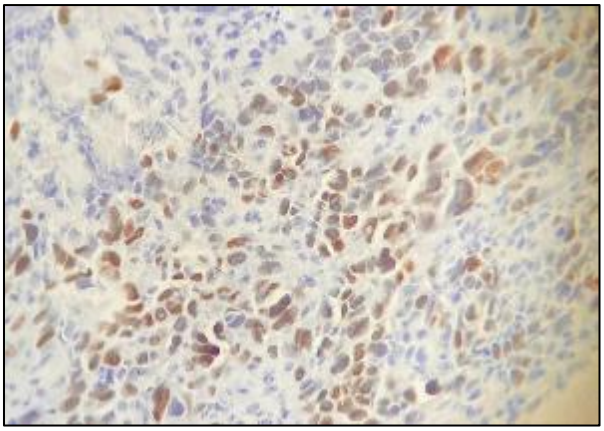

Figure 5 Diffuse nuclear expression of SOX10 (×100)

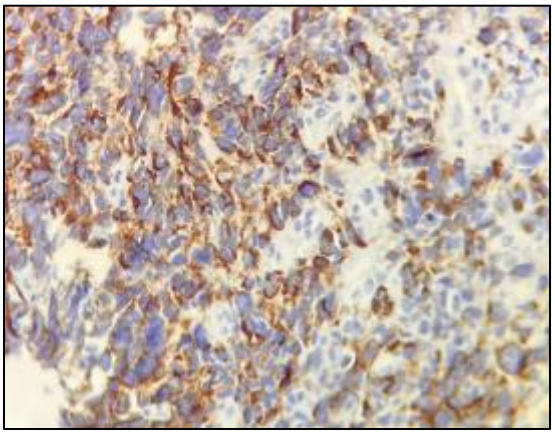

Figure 6 Cytoplasmic expression of HMB45 (×100)

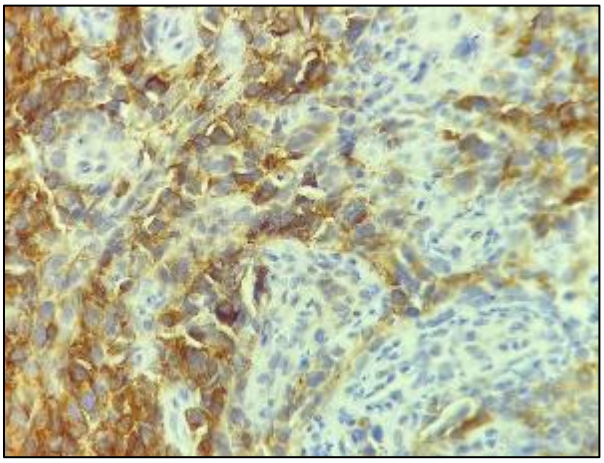

Figure 7 Membranous and cytoplasmic expression of CD117 (×100)

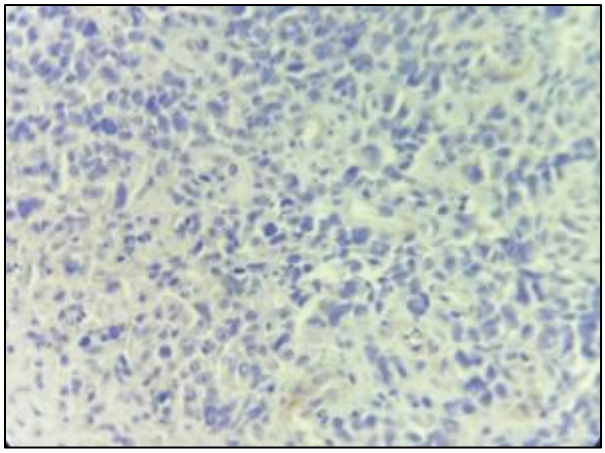

Figure 8 Absence of DOG1 expression in tumor cells (×100)

3. Discussion

Primary rectal melanoma is an uncommon and aggressive tumor, often diagnosed at an advanced stage, with poor survival outcomes [1–4]. Clinical presentation is non-specific, most commonly including rectal bleeding, which frequently delays diagnosis [2,8].

Histologically, these tumors exhibit marked morphological heterogeneity, including epithelioid and spindle cell patterns. Amelanotic variants, as observed in this case, represent a diagnostic challenge, as they may mimic poorly differentiated carcinoma, sarcoma, or lymphoma. In this context, immunohistochemistry is essential. SOX10 and S100 are highly sensitive markers, whereas HMB45 and Melan-A are more specific but may show focal expression [1,5].

A key feature in this case is the diffuse expression of CD117 (c-KIT), which represents a well-recognized diagnostic pitfall. CD117 positivity is more frequently observed in mucosal melanomas than in cutaneous forms and may lead to misdiagnosis as gastrointestinal stromal tumor, particularly in anorectal locations [6,9]. The absence of DOG1 and CD34 expression, combined with melanocytic marker positivity, is crucial for establishing the correct diagnosis.

At the molecular level, mucosal melanomas differ from cutaneous melanomas by a higher prevalence of KIT mutations and a lower frequency of BRAF alterations [4,9]. These molecular features have therapeutic implications, as some patients may benefit from targeted therapy with tyrosine kinase inhibitors such as imatinib [7,9]. However, CD117 expression alone does not predict the presence of KIT mutations, and molecular testing is required for therapeutic decision-making.

Surgical resection remains the mainstay of treatment, although the optimal approach remains debated, as prognosis appears more closely related to tumor biology than to surgical extent [1–3].

In Conclusion, this case highlights the diagnostic difficulty of amelanotic rectal melanoma and underscores CD117 expression as a potential diagnostic pitfall. A comprehensive immunohistochemical approach is essential to avoid misdiagnosis and guide appropriate management

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Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of Ethical Approval

According to institutional policy, ethical approval was not required for this type of study.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

References

[1] Falch C, Stojadinovic A, Hann-von-Weyhern C, et al. Anorectal malignant melanoma: extensive 45-year review and proposal for a novel staging classification. J Am Coll Surg. 2013;217(2):324–335.

[2] Iddings DM, Fleisig AJ, Chen SL, et al. Practice patterns and outcomes for anorectal melanoma in the USA. Ann Surg Oncol. 2010;17(2):321–326.

[3] Postow MA, Hamid O, Carvajal RD. Mucosal melanoma: pathogenesis, clinical behavior, and management. Curr Oncol Rep. 2012;14(5):441–448.

[4] Curtin JA, Fridlyand J, Kageshita T, et al. Distinct sets of genetic alterations in melanoma. N Engl J Med. 2005;353(20):2135–2147.

[5] Agaimy A, Wünsch PH, Hofstaedter F, et al. KIT expression in melanomas: a potential pitfall. Am J Surg Pathol. 2009;33(3):421–427.

[6] Miettinen M, Lasota J. KIT (CD117) expression in melanomas: diagnostic relevance. Arch Pathol Lab Med. 2005;129(6):705–709.

[7] Hodi FS, Corless CL, Giobbie-Hurder A, et al. Imatinib in melanoma with KIT mutations. J Clin Oncol. 2013;31(26):3182–3190.

[8] Tirumani SH, Tirumani H, Jagannathan JP, et al. Imaging features of anorectal melanoma. AJR Am J Roentgenol. 2013;201(4):W635–W642.

[9] Yeh JJ, Shia J, Hwu WJ, et al. The role of KIT in mucosal melanoma. Clin Cancer Res. 2005;11(20):7024–7030.

[10] Bello DM, Smyth E, Perez D, et al. Mucosal melanoma: epidemiology and outcomes. Ann Surg Oncol. 2013;20(11):3617–3624.