World Journal of Surgical Oncology

CASE REPORT

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A rare case of rectal malignant melanoma with long-term survival: case report and literature review

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Abstract

Background Although cutaneous melanoma is relatively common, rectal malignant melanoma is extremely rare. Due to its rarity, rectal malignant melanoma is often not considered in the initial differential diagnosis. In such clinical scenarios, avoiding misdiagnosis, achieving early detection, and providing appropriate treatment remain major challenges and require particular attention. This case report presents a detailed account of the diagnostic and therapeutic process in a patient with primary rectal malignant melanoma. By sharing this case, we aim to provide clinicians with practical experience and valuable insights, thereby enhancing awareness and understanding of this rare condition within the medical community.

Case presentation A 56-year-old female presented to the hospital with a 10-day history of a perianal mass and hematochezia. Physical examination revealed a soft, flat abdomen with no palpable masses and normal bowel sounds. Imaging findings showed localized thickening of the distal rectum, diffuse mild thickening of the mid-to-upper rectal wall, and multiple small lymph nodes in the sigmoid mesocolon.Postoperative histopathological analysis, supported by immunohistochemical staining, confirmed the diagnosis of malignant melanoma. The patient underwent laparoscopic Miles surgery, followed by adjuvant abdominopelvic radiotherapy (GTVtb 50 Gy, CTV45 Gy) and targeted therapy with tislelizumab. She had an uneventful recovery and remained free of disease progression during 48 months of follow-up.

Conclusions This case of rectal malignant melanoma, incidentally diagnosed during hemorrhoidectomy, was successfully treated with laparoscopic Miles' procedure and targeted immunotherapy, resulting in long-term disease-free survival, thereby raising clinical awareness and providing valuable insights for managing similar cases.

Keywords Rectal malignant melanoma, Laparoscopic Miles procedure, Tislelizumab, Case report

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Background

Rectal malignant melanoma is a rare but highly aggressive tumor arising from neural crest-derived melanocytes of the ectoderm. Although melanoma most commonly affects the skin, primary rectal malignant melanoma is exceedingly uncommon, accounting for only 0.4–1.6% of all melanoma cases and less than 0.1% of all rectal malignancies [1, 2]. The disease shows a slightly higher incidence in females than in males and is associated with an extremely poor prognosis, with a five-year survival rate of less than 20% [3].

Due to its insidious onset and nonspecific clinical presentation, the diagnosis of rectal malignant melanoma is often challenging and may be made incidentally during routine procedures such as hemorrhoidectomy. This article presents the diagnostic and therapeutic course of a case of primary rectal malignant melanoma, accompanied by a review of relevant national and international literature. We aim to provide valuable insights and practical experience for clinicians to enhance awareness and understanding of this rare condition.

Case presentation

A 56-year-old female presented with a 10-day history of rectal bleeding and a tender anal mass. After surgical contraindications were ruled out, she underwent a circular hemorrhoidectomy. Postoperative histopathological examination revealed malignant melanoma of the rectum. She was subsequently referred to our outpatient clinic with a preliminary diagnosis of "rectal melanoma" for further evaluation and management.Physical examination revealed that the patient's vital signs were stable, and no obvious abnormalities were detected in the chest or abdomen. During digital rectal examination performed in the knee-chest position, no perianal masses, erythema, or fistulas were identified; however, mild tenderness was noted during the examination. No significant lesions were palpated within the rectum, and digital passage through the stenotic ring was unobstructed.Laboratory tests did not reveal any notable abnormalities. Contrast-enhanced CT and MRI of the rectum (Fig. 1a and b) revealed: Localized thickening of the distal rectum, along with diffuse mild thickening of the middle and upper rectal walls; Multiple small lymph nodes in the sigmoid mesocolon.

After completion of preoperative evaluations, the patient underwent a laparoscopic Miles procedure under general anesthesia. The resected specimen included a segment of rectum with the anus, measuring approximately 20 cm in length (Fig. 1c and d). Approximately 4.5 cm from the anal marginand 2 cm from the dentate line, the mucosa exhibited grayish-black discoloration over an area of about 2×1.5 cm. Within this region, a focal area of deeper pigmentation (approximately 0.5×0.4 cm) with slightly increased firmness was observed.Immunohistochemical staining results were as follows: CK (−), Vim(+),HMB-45 (+), Melan-A (+), S-100 (+), and Ki-67 (approximately 30%+).Histopathological assessment confirmed rectal malignant melanoma(Fig. 2), with metastasis identified in 1 of 18 lymph nodes (1/18) dissected from the sigmoid mesocolon and perirectal adipose tissue.

The patient received adjuvant radiotherapy to the abdominal and pelvic cavity (GTVtb 50 Gy, CTV 45 Gy), in combination with tislelizumab-based targeted therapy and supportive care including gastric protection. During treatment, grade II bone marrow suppression occurred, which improved following leukocyte-stimulating therapy. The patient was subsequently discharged without complications.At the time of writing, the patient has been followed up for 48 months post-treatment, with no evidence of disease recurrence. Follow-up remains ongoing.

Discussion and conclusions

Rectal malignant melanoma is a rare subtype of mucosal melanoma, with a significantly lower incidence compared to the more common cutaneous melanoma [4]. Epidemiological studies have shown that it occurs more frequently in Asian populations than in Caucasians, and is more commonly diagnosed in women, typically between the ages of 53 and 69. Approximately 70–90% of cases arise in the anal canal near the dentate line, while the remaining tumors are located in the perianal skin [5].The early clinical manifestations of rectal malignant melanoma—such as rectal bleeding and abdominal pain—are often nonspecific and can easily be mistaken for more common benign conditions like hemorrhoids or rectal adenocarcinoma [6].Therefore, clinicians should maintain a high level of suspicion when evaluating patients presenting with such symptoms. Comprehensive imaging evaluation and histopathological biopsy are essential for reducing the risk of misdiagnosis.Definitive diagnosis depends on histopathological examination, which may reveal features including nuclear atypia, mitotic activity, and variable pigmentation. Immunohistochemical markers such as S-100, SOX-10, HMB-45, and Melan-A are also valuable in confirming the diagnosis, particularly in amelanotic variants.Currently, surgical resection remains the primary treatment modality, often followed by adjuvant therapies such as chemotherapy or radiotherapy. For patients with high mutational burden or specific biomarker expression, immunotherapy may represent a promising treatment strategy and warrants further exploration.

This patient presented with internal hemorrhoids and rectal bleeding, and was incidentally diagnosed with malignant melanoma following hemorrhoidectomy. Imaging studies revealed localized thickening of the

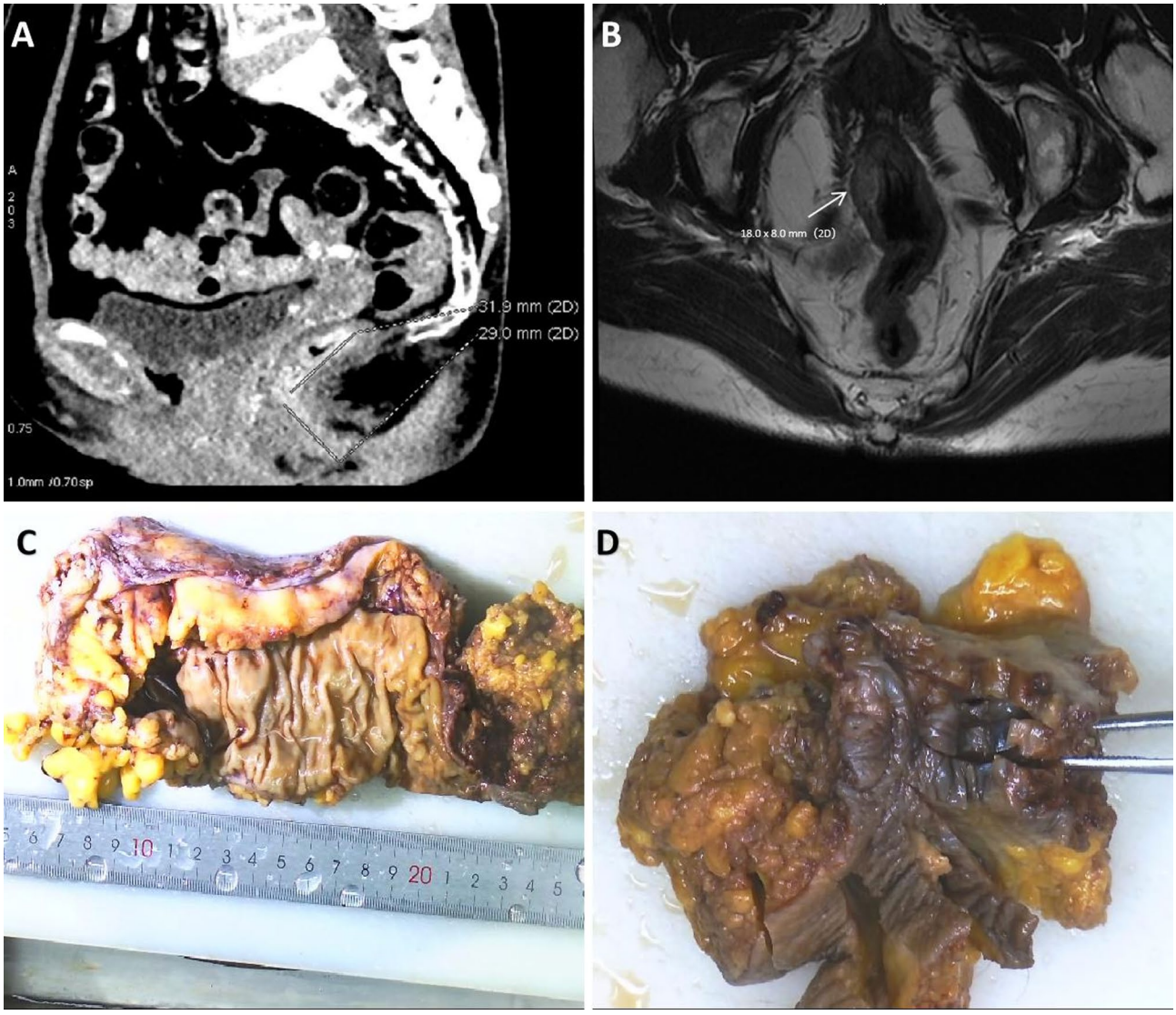

Fig. 1 **a** Preoperative contrast-enhanced CT image.**b** Preoperative contrast-enhanced MRI image.**c** Resected specimen.**d** Mucosa showing gray-black discoloration

rectal wall, which provided an important diagnostic clue. The final postoperative pathological examination confirmed the diagnosis.It is worth emphasizing that during histopathological evaluation, tumor biopsy should not be limited to simple incisional sampling. Instead, complete excision of the lesion is recommended for accurate diagnosis, as partial biopsy may carry the risk of iatrogenic tumor dissemination.Under microscopic examination, rectal malignant melanoma can exhibit various histological patterns, including epithelioid, spindle cell, or mixed subtypes [7]. In this case, the tumor was predominantly composed of spindle-shaped cells, with evident melanin deposition. However, it is important to note that not all cases demonstrate obvious pigmentation; some tumors may be amelanotic (i.e., "non-pigmented melanomas") [8].Immunohistochemical staining in this case showed positive expression of S-100, HMB-45, and Melan-A, further supporting the diagnosis. As rectal malignant melanoma originates from melanocytes derived from neural crest cells that migrate to the mucosa, it is highly aggressive and prone to early local invasion and distant metastasis, particularly to the liver, lungs, and brain [9]. This case exhibited typical histopathological features, sufficient immunohistochemical evidence, and lymph node metastasis, all of which are consistent with the known characteristics of the disease, confirming a clear diagnosis.In this case, the patient underwent laparoscopic Miles' procedure and was subsequently treated with adjuvant radiotherapy (GTVtb 50 Gy, CTV 45 Gy) combined with targeted therapy using tislelizumab. This multimodal approach yielded favorable therapeutic outcomes. The literature supports the use of immune

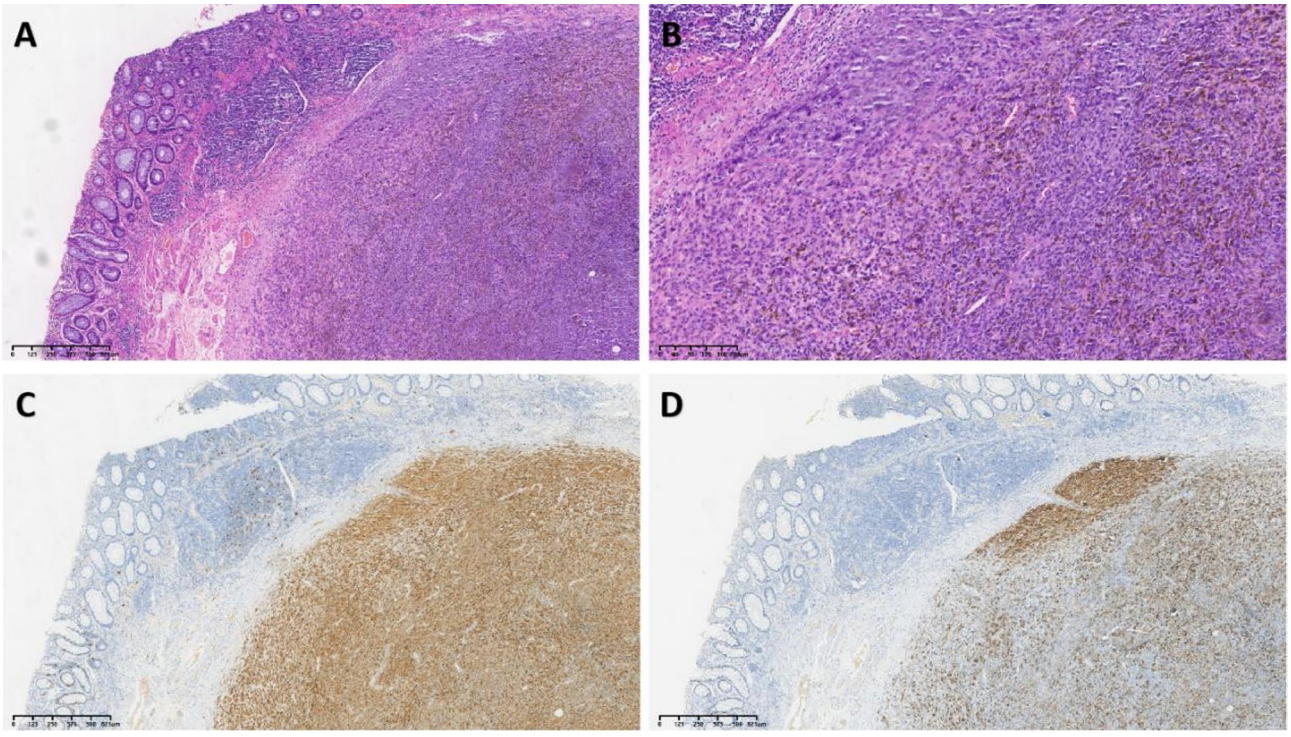

Fig. 2 **a** Submucosal infiltration by numerous tumor cells with indistinct margins between the tumor and surrounding tissue (HE, ×40).**b** Tumor cells showing epithelioid and spindle shapes, with visible pigmentation (HE, ×100).**c** HMB45 (+), EnVision,×40.**d** Melan-A (+), EnVision,×40

checkpoint inhibitors, such as PD-1/PD-L1 inhibitors, in the treatment of melanoma [10]. Studies have shown that the combination of toripalimab and axitinib achieves a notable efficacy in patients with advanced mucosal melanoma. Specifically, this regimen demonstrated an objective response rate (ORR) of 48.3% (95% CI, 29.4–67.5), with a median progression-free survival (PFS) of 7.5 months (95% CI, 3.7 months to not reached) [11]. Furthermore, another study reported an ORR of 36.0% (95% CI, 18.0–57.5) and a median PFS of 6.7 months (95% CI, 4.1 months to not estimable) [12], highlighting its potential as an effective treatment option for this challenging patient population.Despite promising advances in treatment, the overall prognosis for rectal malignant melanoma remains poor. Most studies report a 5-year survival rate of less than 20%, with a median overall survival of 8 to 19 months after diagnosis of primary rectal malignant melanoma [13]. Early diagnosis and treatment can significantly improve survival rates; conversely, later stages, particularly when the cancer has metastasized to lymph nodes or spread to other organs, are associated with a worse prognosis [14].Complete surgical resection (R0 resection) of the tumor and its margins plays a crucial role in determining prognosis. Patients who undergo complete removal without residual disease tend to have longer survival periods [15]. Additionally, the presence or absence of specific genetic mutations or molecular markers can influence disease progression and treatment response [16]. For example, *BRAF V600E* and *KIT* (*V555I* and *K642E*) mutations are commonly observed in rectal melanomas, and targeted therapies are available for these mutations. In contrast, *NRG1* deletions are associated with a poorer prognosis [17].By deepening our understanding of the underlying disease mechanisms and applying novel treatment strategies, we aim to improve the prognosis for patients with rectal malignant melanoma, extending their survival and enhancing their quality of life.

In conclusion, this case of rectal malignant melanoma was incidentally diagnosed at an early stage during hemorrhoidectomy. The patient was treated with a combination of laparoscopic Miles' procedure and targeted immunotherapy, achieving 48 months of disease-free survival — an outcome that is extremely rare in the existing literature. This case report not only helps to further raise clinicians' awareness of this rare disease but also provides valuable insights for the management and treatment of similar cases.

Abbreviations

CT Computed tomography
MRI Magnetic resonance imaging

Acknowledgements
Not applicable.

Author contributions
Huidong Guo and Haitao Huang designed the study under the supervision of Huidong Guo, Haitao Huang, Jiachen Xie, and Yiqun Kang. Jiachen Xie compiled the references. Huidong Guo, Hai Xiao and Yanquan Liu drafted the manuscript. Haitao Huang performed the surgical procedures, with assistance from Yiqun Kang. Huidong Guo and Ye Li conducted the pathological review. All authors have read and approved the final version of the manuscript.

Funding
None.

Data availability
No datasets were generated or analysed during the current study.

Declarations

Consent for publication
Written informed consent was obtained from the patient for publication of this case report, and the accompanying images.

Competing interests
The authors declare no competing interests.

Received: 27 April 2025 / Accepted: 27 May 2025
Published online: 07 June 2025

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