Case Reports and Reviews

Case Report



Anal Melanoma: When The Uncommon Becomes A Diagnostic Challenge Case Report And Brief Review

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Abstract

Anal melanoma is an uncommon neoplasm (~1 % of anal tumours) but highly aggressive, with a 5-year overall survival below 20 %. We present the case of a 74-year-old woman who reported a rapidly growing exophytic anal mass, pain, and haematochezia. Colonoscopy revealed a lobulated tumour 3 cm from the anal verge; the initial biopsy showed a poorly differentiated carcinoma. Positive immunohistochemistry for SOX10, HMB45, and Melan-A confirmed melanoma. Abdominopelvic MRI demonstrated infiltration of the mesorectal fascia and rectovaginal septum without nodal involvement, and ^{18}F-FDG PET/CT ruled out distant metastases, classifying the disease as stage II (T4b N0 M0, AJCC 8th ed.). After multidisciplinary discussion, radical proctosigmoidectomy, total hysterectomy, and abdominopelvic lymphadenectomy with end colostomy were performed, achieving an R0 resection. Owing to the high risk of relapse, adjuvant therapy with pembrolizumab was initiated. This case underscores the diagnostic challenge of anal melanoma, the importance of immunohistochemistry in undifferentiated lesions, and the value of comprehensive imaging staging to select radical surgery and/or immunotherapy. It also highlights the necessity of including melanoma in the differential diagnosis of anal masses and adopting an early multidisciplinary approach to optimise oncological and functional outcomes.

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Keywords

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Introduction

Anal malignancies account for a small proportion of gastrointestinal neoplasms, representing approximately 4% of lower gastrointestinal tract cancers [1,2]. Histologically, approximately 80% are squamous cell carcinomas, whereas adenocarcinomas account for 10%, and the remaining 10% comprise other histological subtypes, including melanoma, which represents approximately 1% of cases [3]. Melanoma may present as either a pigmented (melanotic) or nonpigmented (amelanotic) lesion. Although it can arise in any tissue containing melanocytes, the skin is its most common primary site [3,4].

Anorectal melanomas predominantly arise near the dentate line of the anal canal, with extension into the submucosa [4,5]. Approximately 20–30% of cases involve the rectum. Clinical presentation is often nonspecific, making early diagnosis challenging [3,4,6]. The most common manifestations include perianal pain, rectal bleeding, and perianal swelling, symptoms that are frequently misattributed to benign conditions such as hemorrhoidal disease [3–5]. Less commonly, patients may initially present with inguinal lymphadenopathy or metastatic disease, findings associated with advanced-stage disease and an unfavorable short-term prognosis [3,4].

The diagnostic approach requires a comprehensive evaluation. Colonoscopy plays a key role in the direct visualization of the lesion, while physical examination and complementary imaging modalities, such as pelvic magnetic resonance imaging (MRI) or positron emission tomography–computed tomography (PET/CT), allow for a more accurate assessment of disease extent [7]. From a therapeutic perspective, surgical resection remains the mainstay of treatment, although the optimal extent of surgery continues to be debated, particularly in advanced cases, in which balancing oncological control against functional preservation is critical [8]. Furthermore, the 5-year overall survival rate in these patients does not exceed

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20%, reflecting the aggressive nature of the disease [6].

Clinically, anorectal melanoma can be classified into three stages:

- Stage I: Localized disease.
- Stage II: Locoregionally advanced disease with regional lymph node involvement.
- Stage III: Presence of distant metastases.

This classification not only guides therapeutic management but also has prognostic implications, emphasizing the need for early diagnosis and a multidisciplinary approach to improve clinical outcomes [3,4].

We report the case of a woman in her eighth decade of life who presented with an exophytic anal lesion. Following a comprehensive diagnostic evaluation, the definitive diagnosis of anal melanoma was established.

Case Presentation

We report the case of a 74-year-old woman with no significant past medical history, who presented to the emergency department with a progressively enlarging anal mass of approximately two months' duration. Initially asymptomatic, the lesion gradually increased in size and became associated with severe anal pain, intermittent episodes of hematochezia, and changes in bowel habits characterized by constipation.

On admission, the patient was hemodynamically stable, with a blood pressure of 125/71 mmHg and a heart rate of 78 beats/min. Anorectal examination revealed an approximately 10 × 5 cm indurated lesion located within the anal canal, characterized by a rock-hard consistency, evident areas of necrosis, tenderness on palpation, and friability (Figure 1).



Figure 1. *Exophytic lesion protruding through the anal canal, measuring approximately 10 × 5 cm in its externally visible portion.*

Given the physical examination findings and the suspicion of an underlying malignancy, colonoscopy was performed, revealing a polypoid, lobulated tumor located 3 cm from the anal verge, with a hyperemic, friable, and edematous appearance. Initial histopathological examination revealed a poorly differentiated, ulcerated anorectal tumor. For further characterization, immunohistochemical analysis was performed. Histopathological evaluation demonstrated a malignant neoplasm involving the lamina propria, composed of a diffuse proliferation of small cells with scant cytoplasm, hyperchromatic nuclei, and increased mitotic activity. Immunohistochemical staining showed diffuse positivity of the tumor cells for SOX10, HMB45, and Melan-A, confirming the histopathological and immunophenotypic diagnosis of melanoma.

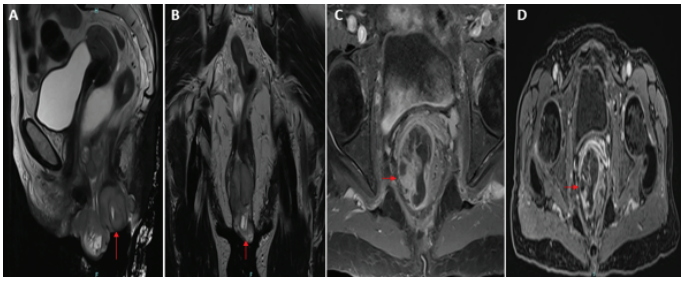


Figure 2*. Abdominopelvic magnetic resonance imaging (MRI). MRI demonstrating a solid anorectal mass with complete circumferential involvement, infiltration of the mesorectal fascia and the upper third of the rectovaginal septum, and severe luminal stenosis exceeding 80%, consistent with locally extensive disease.*

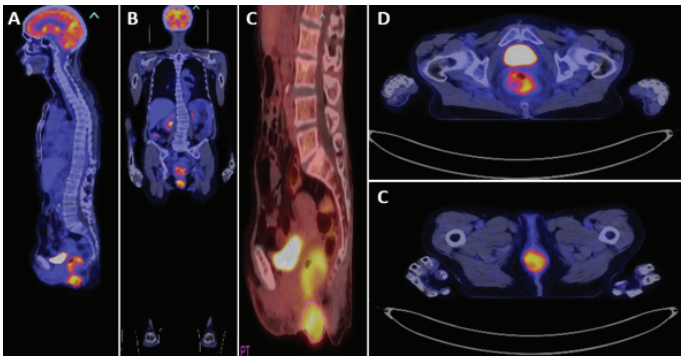


Figure 3. *^{18}F-fluorodeoxyglucose positron emission tomography/computed tomography (^{18}F-FDG PET/CT). PET/CT demonstrating a hypermetabolic anorectal mass corresponding to the primary tumor, without evidence of regional lymph node involvement or distant metastatic disease.*

As part of the staging workup, brain magnetic resonance imaging (MRI) was performed, revealing chronic microangiopathic and involutional changes, without reported evidence of intracranial metastases. Subsequent abdominopelvic MRI demonstrated a solid anorectal mass with complete circumferential involvement, infiltration of the mesorectal fascia and the upper third of the rectovaginal septum, and severe luminal narrowing exceeding 80% (Figure 2). Finally, ^{18}F-fluorodeoxyglucose positron emission tomography/computed tomography (^{18}F-FDG PET/CT) demonstrated a hypermetabolic anorectal lesion, with no evidence of regional lymph node involvement or distant metastatic disease (Figure 3).

Following multidisciplinary evaluation, radical surgical management was undertaken, consisting of proctosigmoidectomy, total hysterectomy, radical abdominopelvic lymphadenectomy, and partial omentectomy, with colostomy formation. Subsequently, adjuvant therapy with pembrolizumab, a programmed cell death protein 1 (PD-1) inhibitor, was initiated under the supervision of the medical oncology team.

Discussion

Anal melanoma is a rare but biologically aggressive malignancy whose management continues to pose significant challenges in contemporary oncological practice. The

present case illustrates the clinical characteristics, diagnostic challenges, and therapeutic complexities associated with this disease [3,5,6].

Diagnostic Challenges and Clinical Findings

As observed in our 74-year-old patient, anal melanoma frequently presents with nonspecific symptoms, including anal pain, hematochezia, and changes in bowel habits, which may mimic benign anorectal conditions such as hemorrhoids or anal fissures. This overlap in clinical presentation may contribute to diagnostic delays, with up to 80% of cases reportedly diagnosed at advanced stages (stage II or III), characterized by locoregional involvement or metastatic disease at presentation [3,9]. The 10 × 5 cm exophytic lesion with necrotic areas observed in our patient is consistent with published series reporting a mean tumor size exceeding 4 cm at diagnosis.

Immunohistochemical analysis using HMB45, Melan-A, and SOX10 was essential for establishing the definitive diagnosis, particularly given the poorly differentiated morphology observed on initial histopathological examination. This is especially relevant because up to 30% of anorectal melanomas are amelanotic, increasing the risk of diagnostic misclassification. Imaging studies, including pelvic magnetic resonance imaging (MRI) and positron emission tomography/computed tomography (PET/CT), served complementary purposes by delineating the extent of local invasion, including rectovaginal septal involvement and luminal stenosis exceeding 80%, while excluding detectable metastatic disease [10–12].

Controversies in Surgical Management

The radical surgical approach undertaken in this case, including proctosigmoidectomy, lymphadenectomy, and hysterectomy, reflects a central therapeutic dilemma in anorectal melanoma. Although wide local excision with margins of at least 2 cm may improve local disease control, a corresponding overall survival benefit has not been consistently demonstrated [8,13]. Retrospective studies, including that by Matsuda et al., suggest that abdominoperineal resection does not confer a survival advantage over wide local excision, although it may reduce local recurrence rates [13]. In the present case, the decision to pursue radical surgery was based on extensive circumferential tumor involvement and invasion of the rectovaginal septum, with the aim of achieving complete resection with negative margins (R0).

Advances in Systemic Therapy

The incorporation of pembrolizumab as adjuvant therapy represents an evolving therapeutic strategy. Evidence from clinical trials involving cutaneous melanoma has demonstrated that programmed cell death protein 1 (PD-1) inhibitors reduce the risk of recurrence in selected patients with resected high-risk disease [14]. However, evidence specifically supporting their use in anorectal melanoma remains limited. Small clinical series have reported encouraging responses to anti-PD-1 therapy in patients with metastatic mucosal melanoma [15]. In our patient, who had no detectable distant metastases at diagnosis, adjuvant pembrolizumab was initiated with the aim of reducing the risk of disease recurrence, although its specific benefit in this setting remains uncertain.

Prognosis and Predictive Factors

The histopathological characteristics observed in this case, particularly increased mitotic activity and a diffuse tumor growth pattern, suggest aggressive biological behavior. The absence of radiologically detectable regional lymph node involvement represents a potentially favorable finding; however, extensive local invasion involving the rectovaginal septum indicates a substantial risk of recurrence despite the absence of distant metastatic disease.

Conclusion

Although rare, anal melanoma is a highly aggressive malignancy associated with a poor prognosis, as illustrated by the present case of a 74-year-old woman with locally advanced disease and extensive invasion of adjacent structures. Its nonspecific clinical presentation and frequent resemblance to benign anorectal conditions may delay diagnosis, as reflected by the large tumor size (10 cm) at presentation. Definitive diagnosis required a multimodal approach, highlighting the essential role of immunohistochemical markers (HMB45, Melan-A, and SOX10) in identifying melanoma in poorly differentiated tumors. Surgical resection remains the cornerstone of treatment for localized disease, although the survival benefit of extensive radical surgery remains uncertain. The use of adjuvant pembrolizumab reflects advances in immunotherapy; however, its efficacy specifically in anorectal melanoma requires further investigation. This case underscores the importance of considering melanoma in the differential diagnosis of atypical anal masses and emphasizes the need for a multidisciplinary approach to facilitate timely and individualized therapeutic decision-making.

Declaration of Artificial Intelligence

Use Artificial intelligence (AI) tools were used exclusively to assist with language editing, grammatical corrections, improvements in sentence structure and readability, and translation of the manuscript from Spanish into English. AI tools were not used to generate clinical data, interpret diagnostic findings, conduct statistical analyses, or formulate scientific conclusions. All AI-assisted modifications were reviewed and validated by the authors, who assume full responsibility for the accuracy, integrity, and scientific content of the final manuscript.

Conflicts of Interest

The authors declare no conflicts of interest.

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