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Anorectal Mucosal Melanoma Discovered During Hemorrhoidectomy

Ben Hassine Basma^{a,c}, Rapti Souphia^{a,b,*}, Haloui Nabil^{a,b},
Ben Marzouk Saoussen^{a,b}, Lahouar Omar^{a,d}, Khalifa
Mohamed Bechir^{a,b}

^aGeneral Surgery Department, Military Hospital of Tunis, Mont Fleury-1008, Tunis, Tunisia

^bFaculty of Medicine of Tunis, 15, Djebel Lakhdhar Street – 1007 Bab Saadoun, Tunis, Tunisia

^cFaculty de Medecine Ibn El Jazzar of Sousse, Mohamed Karoui Street - 4002, Sousse, Tunisia

^dFaculty of medicine of Monastir, Avenue Avicenne- 5000

*Corresponding Author:

Rapti Souphia

General Surgery Department, Military Hospital of Tunis, Mont Fleury-1008, Faculty of Medicine of Tunis, 15, Djebel Lakhdhar Street – 1007 Bab Saadoun, Enkhilet, Ariana 2083, Tunisia

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1. Abstract

1.1. Introduction: Anorectal melanoma is a rare and aggressive malignant neoplasm, accounting for 0.4% to 1.6% of melanomas. It is often diagnosed at an advanced stage due to its nonspecific presentation, and frequently mimics benign conditions, with hemorrhoids being the most common diagnostic pitfall. We report a case incidentally discovered during a hemorrhoidectomy, highlighting the diagnostic and therapeutic challenges.

1.2. Case report: A 61-year-old patient presented with recurrent anal pain. A physical examination revealed the presence of three circumferential masses at the anal margin, which were initially suspected to be hemorrhoids. However, subsequent histopathological analysis confirmed a malignant melanoma. Further investigation revealed generalized peritoneal carcinomatosis, which resulted in the decision to discontinue any surgical resection.

1.3. Discussion: Anorectal melanoma is a rare tumor that should be prioritized when an atypical anorectal lesion is encountered. The diagnosis is confirmed through biopsy, considered the gold standard. Management requires a multidisciplinary approach, with therapeutic options ranging from wide local excision to abdominoperineal resection. Prognosis is generally poor, with tumor stage at diagnosis being the main prognostic factor.

1.4. Conclusion: This case highlights the rarity of anorectal melanoma and emphasises the importance of including it in the differential diagnosis of anorectal pathology, as delayed diagnosis is associated with a particularly poor prognosis.

2. Keywords: Melanoma, anorectal, haemorrhoids, Diagnosis, Multidisciplinary management, treatment.

3. Introduction

Anorectal melanoma is a rare malignancy accounting for approximately 0.4% to 1.6% of all melanomas and 1% of anorectal malignancies[1,2].

It predominantly affects older patients, typically between the sixth and eighth decades of life, with a higher incidence in women[3]. This disease is notable not only for its rarity, but also for the lack of standardized treatment guidelines and its poor prognosis, with a five-year survival rate of less than 20%[4,5].

Early diagnosis is essential to improve patient outcomes. Through a clinical case and a review of the literature, we analyze the clinical, therapeutic, and prognostic features of this uncommon pathology. This work has been reported in line with the SCARE criteria [6].

4. Case report

A 61-year-old female patient with a history of external hemorrhoids initially consulted a gastroenterologist for recurrent anal pain persisting for six months, of moderate intensity and unresponsive to symptomatic treatment, associated with rectal bleeding without alteration of bowel habits.

On physical examination, her body mass index (BMI) was 24 kg/m², and her abdomen was soft, depressible, and non-tender, with no organomegaly. Lymph node examination did not reveal any adenopathy. Evaluation of the anal margin revealed three circumferential masses, initially suspected to be hemorrhoids, with no palpable rectal mass (Figure 1).



Figure 1: Examination of the anal margin after lesion excision

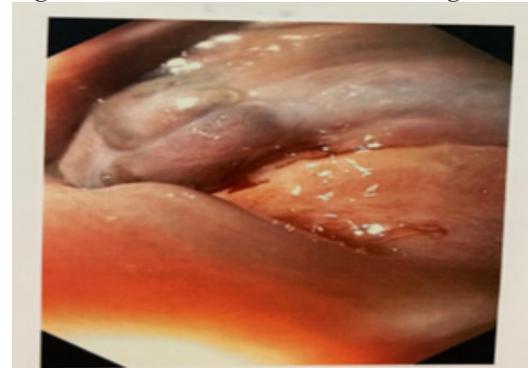


Figure 2: Colonoscopic image showing the persistence of a 2.5 cm lesion

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cm tumor mass.

The excision of the lesions was followed by a histopathological analysis, which confirmed the diagnosis of malignant melanoma with positive margins. A colonoscopy revealed a persistent tumor mass of 2.5 cm at the anal margin, with no other abnormalities (Figure 2).

The staging work-up revealed a suspicious 13 mm carcinomatosis nodule in the vesico-uterine space (Figure 3). Intraoperative exploration demonstrated generalized peritoneal carcinomatosis with large vesico-uterine nodules, leading to the abandonment of any abdominoperineal resection (APR).

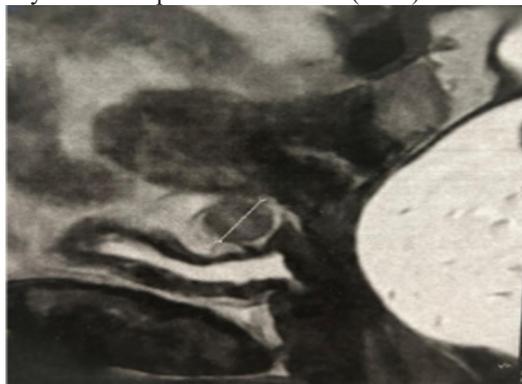


Figure 3: Pelvic MRI revealing a suspicious 13 mm carcinomatosis nodule in the vesico-uterine space.

5. Discussion

Anorectal malignant melanoma (AMM) is an aggressive malignancy that originates from melanocytic cells in the anorectal mucosa[7]. As a subtype of mucosal melanomas, AMM is characterized by a poor prognosis and distinct molecular features[8]. Unlike cutaneous melanomas, which frequently harbor BRAF mutations, mucosal melanomas, including AMM, are more commonly associated with activating mutations in the receptor tyrosine kinase c-KIT[7].

The symptoms of anorectal melanoma, such as rectal bleeding, tumor mass and anal pain, are often non-specific and can be mistaken for other benign or malignant anorectal conditions, particularly hemorrhoids, leading to frequent diagnostic errors[5,9]. This confusion results in delays in diagnosis and early management, as observed in our patient.

On clinical examination, the tumor typically presents as an ulcerative and exophytic mass or a polypoid and pedunculated lesion with characteristic black pigmentation[4,10]. However, some amelanotic forms further complicate diagnosis[11]. These tumors are often detected at an advanced stage, with a median size ranging from 2.2 to 3.2 cm[12].

Histological and immunohistochemical examination remains the gold standard for diagnosis[10].

A complete colonoscopy is recommended to rule out synchronous lesions. Endoscopic ultrasound or pelvic MRI allow for local staging, while systemic staging relies on thoraco-abdominopelvic CT scans or PET-CT exams, which help detect nodal and distant metastases[13].

AMM is classified into three stages: localized disease (stage I), involvement of regional lymph nodes (stage II), and distant metastases (stage III)[1]. Tumor stage is an independent prognostic factor for overall survival, highlighting the need for thorough

assessment to optimize management[14,15].

To date, there is no consensus regarding the optimal surgical strategy for the treatment of locoregional anorectal melanoma. While surgery remains the standard of care, the choice between local excision (LE) and abdominoperineal resection (APR) remains controversial[7,16].

LE reduces surgical morbidity and preserves continence, but carries a high risk of local recurrence. APR appears to provide better local control; however, no survival benefit has been demonstrated and metastatic progression remains the leading cause of mortality.[17]

Treatment decisions should be individualized and discussed within a multidisciplinary team[12]. The role of adjuvant therapies remains uncertain, as neither chemotherapy nor radiotherapy has shown a survival benefit, although the latter may be considered for palliative care[4].

6. Conclusion

Anorectal melanoma is a rare and highly aggressive malignancy. It is typically diagnosed at advanced stages, which adversely affects prognosis[7].

This case highlights the complexities of its management and underscores the importance of a multidisciplinary approach. Although surgery remains the primary treatment option, it does not significantly impact survival[4]. The role of adjuvant therapies is still debated. Early diagnosis and the exploration of novel therapeutic strategies, particularly immunotherapy, are essential for improving clinical outcomes[8].

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8. Ethical Approval

Ethical approval for this study (Ethical Committee N° MHOT 2024) was provided by the Ethical Committee of the Military Hospital of Tunis, Mont Fleury-1008, Tunis, Tunisia, on 15 February 2024.

9. Patient Consent

Written informed consent was obtained from the patient for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

10. Research Registration

As per the World Medical Association's Declaration of Helsinki 2013, all research studies involving human subjects should be registered in a publicly accessible database before recruitment. This case report has been retrospectively registered at www.researchregistry.com with the Unique Identifying Number (UIN)

Guarantor

Rabti Souphia

Ethics Approval and Consent to Participate

Ethical approval is not required for this case report. Institutional

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policy does not mandate ethical approval for case reports involving retrospective observations of previously treated patients.

Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for the Editor-in-Chief upon request.

Availability of Data and Material

All relevant data are included in the manuscript. Additional information can be provided upon reasonable request.

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11. Author contribution

Rabti Souphya and Ben Hassine Basma contributed to manuscript writing and editing, and data collection; *, Haloui Nabil, Ben Marzouk Saoussen contributed to data analysis; Med Bachir Khalifa contributed to conceptualization and supervision; All authors have read and approved the final manuscript

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