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Rare Differential Diagnosis of Hemorrhoidal Disease: Primary Anorectal Melanoma; a Case Report and Review of the Literature

Willy Stéphane Kengne[1*], Joseph Idriss Djoko[2], Gael Jacquemin[3], Julie Vandepapeliere[4], Lynn Gabrielle Alexis[4], Silva De Vulder-Cotrina[4], Adeline Donati[1], Marie-Cécile Nollevaux[5], Olivier Borgniet[1]

[1]Gastroenterology Department, CHU UCL NAMUR, Dinant, Belgium
[2]Plastic Surgery Department, CLINIQUE UNIVERSITAIRE SAINT LUC UCL, Bruxelles, Belgium
[3]General Surgery Department, CHU UCL NAMUR, Dinant, Belgium
[4]Oncology Department, CHU UCL NAMUR, Dinant, Belgium
[5]Department of Anatomopathology, CHU UCL NAMUR, Godinne, Belgium
Email: *stephiwilly@yahoo.fr

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Abstract

Anorectal melanoma is a rare tumor representing less than 1% of anorectal cancers and around 0.3% of malignant melanomas. Its prognosis is particularly poor due to the early occurrence of metastases. We report the case of a 65-year-old man presenting with rectorrhagia and anal pain, initially diagnosed as hemorrhoidal disease. Subsequent proctological examination revealed an ulcerating-bourging tumor, confirmed histologically as an anorectal melanoma. After a normal extension workup, an abdominoperineal amputation was performed. Anorectal melanoma is a pathology with a poor prognosis, requiring early diagnosis to improve chances of survival.

Keywords

Anorectal Melanoma, Diagnosis, Treatment, Prognosis, Anatomopathology, Surgery

1. Introduction

Anorectal melanoma is a rare tumour, accounting for between 1% and 3% of anorectal malignancies, and around 0.3% of malignant melanomas [1]. Its annual incidence is estimated at around 4.8 cases per 10 million people, and its prevalence has increased markedly in recent decades [1]. Although prevalence is rising, standards of diagnosis and management remain poorly defined. This tumor occurs

most frequently in elderly patients, with a predominance in Caucasian women [1]. Clinically, initial symptoms include rectal bleeding and changes in bowel habits, often confused with more common pathologies such as hemorrhoids. This confusion leads to delayed diagnosis, and around 60% of patients already have metastases at the time of diagnosis [1]. Treatment relies mainly on surgical excision, although there is no clear consensus on the management of distant metastases or the use of standardized adjuvant therapies. Survival of patients with anorectal melanoma remains unfavorable overall, with a median survival of around 24 months and a 5-year survival rate of just 10%, with the majority of patients succumbing to metastases [1]. We report a rare case of hemorrhagic anorectal melanoma mimicking hemorrhoidal disease.

2. Case History

We report the case of a 65-year-old Caucasian man with a history of hypertension and no family history of colorectal cancer or other malignancies. He had presented for three months with rectal discharge associated with anal pain and no other associated symptoms. A digital rectal examination (DRE) was performed and found to be normal. Proctological examination, supplemented by colonoscopy, revealed Goligher grade 3 hemorrhoids and two rectal polyps located between 5 and 25 cm from the pectineal line. These were resected and the hemorrhoidal packets ligated.

The patient was seen again two months later for persistent moderate anal pain and rectal discharge. On rectal examination, a flat, non-obstructive mass, measuring between 20 and 35 mm and located in the anterolateral part of the linea alba of the anal canal, was palpated. The mass bled on touch. Anorectoscopy revealed a rounded ulcerating lesion, mobile but fixed to the anterolateral wall of the anal canal (**Figure 1**). Biopsies were taken, revealing a primary anorectal melanoma.

Histological analysis confirmed the presence of cells expressing the immunohistochemical markers S100 and Melan-A (**Figure 2**), characteristic of melanoma. MRI of the rectum showed a lesion invading the entire circumference of the lower rectum over a length of 69 mm, with infiltration of the mesorectum (stage T3 CRM 0 N+) and lymph nodes. There was also involvement of the internal sphincter in the right posterolateral upper quarter (**Figure 3**). A PET scan was performed to assess the extent of the disease, revealing hypermetabolism in the recto-anal region, as well as para-rectal adenopathies and a hypermetabolic focus in the left tonsil. Following a multidisciplinary consultation meeting, in view of the size of the tumour and its locoregional infiltration, it was decided to carry out a neodajuvant treatment to reduce the size of the tumour and allow R0 resection. This consisted of two courses of nivolumab and ipilimumab. They had to be stopped due to grade 2 skin toxicity after the first course and immuno-induced encephalopathy after the second, necessitating hospital management. A PET scan showed regression of tumor hypermetabolism, and laparoscopic abdominoperineal amputation was performed. Postoperative outcome was favorable.

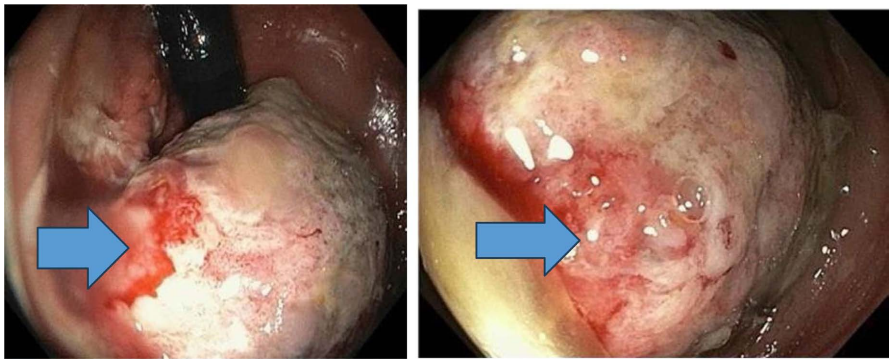

Figure 1. Retrovisional endoscopic image: visualization in the anal canal of an ulcerating-bourging mass attached to the anterolateral border of the lower rectum.

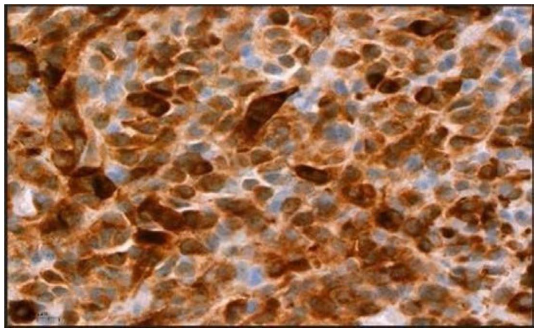

Figure 2. Visualization of MELAN A melanocytic marker expression by atypical cell sheets using immunohistochemistry.

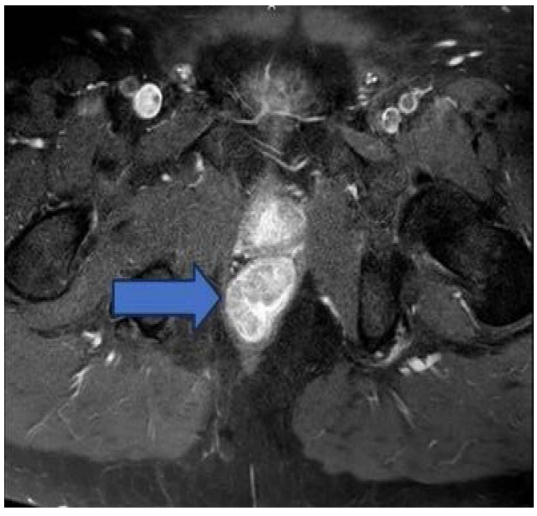

Figure 3. Axial and sagittal MRI slices showing a lower rectal lesion in, T2 hyper signal taking contrast heterogeneously with fat infiltration.

Histology of the surgical specimen revealed an ulcerated nodular lesion 7 cm long and 4.5 cm wide reaching the pectineal line, with metastatic infiltration of six out of twenty-six lymph nodes. Surgical margins were free of tumour cells:

resection in macroscopically and microscopically healthy margins. The patient received adjuvant treatment with nivolumab, but this was discontinued due to side effects. Oncological follow-up was initiated, with discussion of immunotherapy or anti-PD-L1 in case of relapse.

3. Discussion

Anorectal melanoma is a rare malignant tumor arising from the transformation of melanocytes derived from the neural crest [1]. It accounts for around 0.3% of all melanomas and less than 1% of anorectal cancers [1] [2]. The disease mainly affects elderly patients, with peak incidence between the 5th and 7th decades, and a predominance in Caucasian women. It often results from infiltration of the rectal mucosa by a process initially located at anal level, originating from melanocytes normally present in the squamous epithelium of the pectinate zone as well as in the transitional epithelium above this line [3]-[5]. Anorectal melanoma has a poor prognosis, with an average survival of 20 months after treatment.

Initial symptoms are often non-specific, dominated by anemia and rectal discharge, leading to frequent misdiagnosis with more benign pathologies such as hemorrhoids or rectal polyps. As a result, the diagnosis is often made late, with around 60% of patients presenting with metastases at the time the tumour is discovered; these metastases are of multiple local or distant location, as they disseminate via the hematogenous and lymphatic routes. A full extension work-up is essential, including colonoscopy to look for synchronous lesions, rectal echo-endoscopy or pelvic MRI to assess parietal and lymph node infiltration, and thoraco-abdominopelvic CT or PET scan to identify distant metastases [6]. Diagnostic confirmation is based on histology, including specific staining (FONTANA) and immunohistochemistry, such as positivity for S100 and Melan-A markers [7] [8].

The mainstay of treatment for anorectal melanoma is surgery, with abdominoperineal amputation with lymph node courage the procedure of choice in locally advanced forms. However, recommendations on the management of distant metastases and the use of adjuvant therapies, such as immunotherapy, remain unclear in the absence of data from randomized clinical trials [9] [10]. In this case, although surgery has been successfully performed, the presence of lymph node metastases underlines the aggressive nature of this pathology, justifying close monitoring of the patient.

Therapeutic perspectives include immune checkpoint inhibitors such as nivolumab and ipilimumab, which have shown promising results despite their potential toxicity [11] [12].

Prolonged follow-up is necessary to assess the efficacy of these treatments and prevent recurrence, given the generally unfavorable prognosis of this type of tumor.

4. Conclusion

Anorectal melanoma is a rare disease with a particularly poor prognosis, due to

the frequency of metastases at the time of diagnosis. Early diagnosis is crucial to improve prognosis, although the non-specific clinical presentation makes recognition difficult. Treatment relies mainly on surgery, but adjuvant therapies, particularly immunotherapy, offer new perspectives. It is essential for practitioners to consider this pathology in the differential diagnosis of rectal bleeding, and to monitor for signs of metastatic recurrence.

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Ethics Approval and Consent to Participate

Consent for publication was obtained from the patient.

Consent for Publication

All authors approved the final version of the manuscript and agreed for publication.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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