Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Anorectal Melanoma: Case Report and Literature Review

Z. Maflah[1*], Pr B. Slioui[1], Pr S. Bellasri[1], Pr N. Hammoune[1], Pr A. Mouhsine[1], Pr M. Atmane[1]

[1]Department of Radiology, Avicenne Military Hospital, Marrakech, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i05.065 | **Received:** 04.04.2025 | **Accepted:** 12.05.2025 | **Published:** 19.05.2025

***Corresponding author:** Z. Maflah
Department of Radiology, Avicenne Military Hospital, Marrakech, Morocco

Abstract **Case Report**

Anorectal mucosal melanoma is an uncommon and highly aggressive form of mucosal melanoma. Its rarity makes clinical diagnosis difficult, and its initial symptoms are generally non-specific, such as rectal bleeding (the most frequent symptom), anal pain or the presence of an anal mass. The prognosis for this disease is generally poor, and its incidence seems to be increasing every year. Anorectal mucosal melanoma often goes undetected and/or is already metastasized at the time of diagnosis. We present a case report of a patient who initially presented with nonspecific symptoms of anemia and rectorrhagia and was later found to have melanoma of the anorectal region. There is a notable paucity of literature on this disease, resulting in a lack of overall understanding of its nature. Most of the information available is in the form of isolated case reports rather than comprehensive studies. Although surgical resection remains the primary treatment approach, the majority of patients (over 80%) will die from distant metastases within five years of surgery. The five-year survival rate for anorectal melanoma is estimated at between 6% and 22%.

Keywords: Melanoma Skin Cancer, Initial Presentation of Malignant Melanoma, Mucosal Malignant Melanoma, Rectal Malignant Melanoma, Anorectal Melanoma.

INTRODUCTION

Skin cancer is the most common type of cancer, with invasive melanoma accounting for around 1% of all skin cancers. Although most melanomas occur on the skin (i.e. cutaneous melanomas), there are also mucosal melanomas that develop in the mucous membranes of the body, including the sinuses, nasal passages, oral cavity, vagina, anus and rectum1. Mucosal melanomas account for around 1.4% of all melanomas, of which around 50% originate in the head and neck. A larger proportion of the remaining cases occur in the anorectal region and female genitalia, while a smaller percentage affect the oesophagus, gallbladder, intestine, conjunctiva and urethra2. Anorectal mucosal melanoma (AMM) accounts for between 0.4% and 14.6% of all malignant melanomas, and 4% of all anal malignancies.

Melanoma incidence has risen steadily over the past few decades; according to the Centers for Disease Control and Prevention, the incidence rate in the United States from 2012 to 2016 was 21.8 cases per 100,000 individuals. The highest incidence was seen in non-Hispanic Caucasian males, while the lowest incidence was reported in individuals of African descent. In 2023, the American Cancer Society estimates that around 186,680 new cases of melanoma will be diagnosed in the United States, of which 97,610 will be invasive melanoma (58,120 in men and 39,490 in women). The society also predicts that 7,990 people (5,420 men and 2,570 women) will die from the disease [4].

Melanoma develops when melanocytes undergo mutations, leading to their uncontrolled proliferation and transformation into cancerous cells. These mutations can be acquired sporadically (the most common) or inherited through germline mutations. Exposure to ultraviolet (UV) light, mainly from sunlight, is identified as the main risk factor for the development of melanoma, according to the American Cancer Society [5]. Other risk factors include a family history of the disease, male gender, fair skin, fair hair, immunocompromised subjects, aging and conditions such as xeroderma pigmentosum. Race is also an important predictor of melanoma development [2].

Melanoma treatment varies according to the stage of the disease (i.e. stages I to IV), and is most effective in the early stages when it is confined to the epidermal layer of the skin. Thanks to a better understanding of the pathogenesis, progression and immunology of the disease, treatment modalities have rapidly evolved. Options include immunotherapy, targeted therapy, radiotherapy, chemotherapy and

surgical resection, with surgery being the preferred first-line therapy.

The aim of this report is to highlight a highly morbid type of melanoma in the hope of promoting earlier detection and, consequently, improving prognosis.

CASE PRESENTATION

We report the case of a 66-year-old female patient with no previous medical or surgical history. She presented with moderate rectal discharge for 9 months and anemia of 9 g/dl. A physical examination revealed an anal nodule. A radiological workup and biopsy were performed. Pelvic MRI showed a lesional process centred on the anal canal, lateralized to the left, budding endoluminal and extending to the lower and middle rectum, with lobulated contours, heterogeneous hypersignal in T2, iso signal in T1, diffusion restriction and strong, heterogeneous enhancement after injection of gadolinium (Figure 1).

This process infiltrates the left pubo-rectal fascicle and levator ani muscle, bulges into the ischial fossa and prolapses its lower pole through the anal orifice. It also infiltrates the internal sphincter on its left lateral side.

Mesorectal lymph nodes of regular outline and sub-centimetre size are associated.

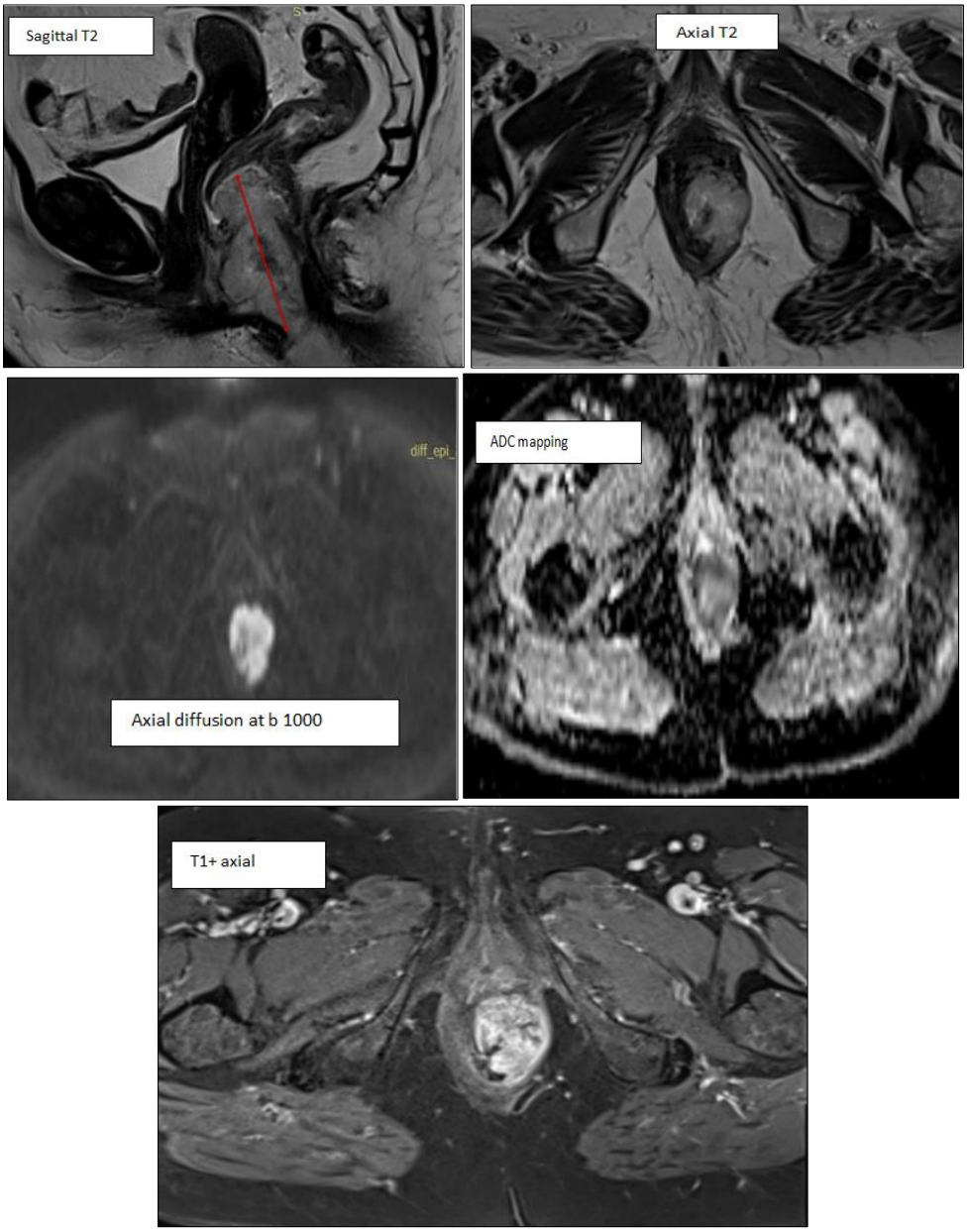

Figure 1: Pelvic MRI in sagittal and axial sections: lesional process centered on the anal canal lateralized to the left, budding endoluminal and extending to the lower and middle rectum

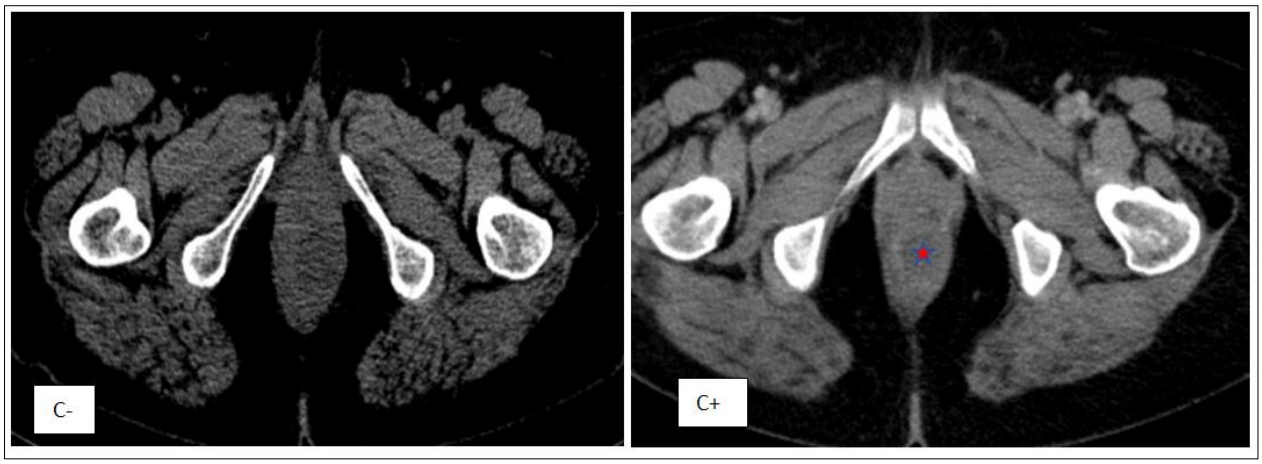

Figure 2: Abdominal-pelvic CT scan in axial section without and with injection of contrast medium: shows heterogeneous enhancement of the lesional process centred on the anal canal, delimiting a central necrotic zone (asterisk)

A thoraco-abdomino-pelvic CT scan was performed as part of the extension work-up, showing no secondary localization.

A rectal biopsy revealed a largely reorganized undifferentiated tumour proliferation. A complementary immunohistochemical study revealed positive anti-MDM2, anti-PS100, anti-HMB45, anti-Melan A and anti-SOX-10 antibodies. The immunohistochemical appearance favoured rectal localization of a melanoma.

To this end, the patient underwent abdominoperineal amputation with lymph node curage.

DISCUSSION

MSM are often delayed due to their initial non-specific presentation. One study found that 41% of these tumors had already spread regionally, and 22% had distant metastasis by the time they were detected [6. The lungs, liver, brain and bone are typical sites of metastasis [7].

The most common initial symptom of MSA is painless rectal bleeding. Due to the rarity of this neoplasm, other conditions, such as hemorrhages, polyps and even squamous cell carcinomas, are usually considered first and excluded. In larger tumors with deeper infiltration, symptoms may become more noticeable, including bleeding, the presence of an anal mass, pain, constipation and weight loss [8].

Although melanomas are usually pigmented, if a dark mass is seen at the anal verge, it may be mistaken for thrombosed hemorrhage. In addition, MSAs can present as amelanomatous melanomas, lacking pigmentation and requiring histopathological evaluation for diagnosis [9].

Melanoma originates from melanocytes, the pigment-producing cells in the skin. Melanocytes are found in the basal layer of the epidermis and are responsible for the production of melanin, the pigment that gives color to the skin and provides protection against UV radiation, acting as a natural "sunscreen". In mucous membranes, melanocytes also play a role in antimicrobial defense and immune responses [10].

The exact pathogenesis of MSA remains limited and is not fully understood [10]. Several theories exist concerning its development. Some propose a relationship with oxidative stress in the anal region and/or immunosuppression. Others suggest derivation from Schwannian neuroblastic cells or from the uptake and decarboxylation of the gut amine precursor and decarboxylation system [11]. The KIT receptor tyrosine kinase has also been implicated in the development of malignant melanoma, including MSA. Loss-of-function mutations, as well as activating mutations in genes such as KIT, BRAF and NRAS, have been associated with melanoma development [2].

Due to the rarity of MSA, there are insufficient randomized trials and standardized treatment plans for the disease. However, surgical interventions are commonly used [10]. WLE resection or abdominoperineal (APR) are the preferred surgical approaches, with similar results. Given the morbidity associated with APR, WLE is generally recommended [12]. Mucinous melanoma of intermediate thickness may require sentinel lymph node biopsy [13].

Various adjuvant therapies have been used in the literature. Interferon alfa immunotherapy has been shown to increase survival and decrease recurrence rates in node-positive patients. Other treatments, such as 117-Cesium brachytherapy and chemotherapy protocols containing dacarbazine, vincristine and nimustine hydrochloride, have been used with variable success rates [14]. Treatment decisions must take into account the patient's quality of life and comorbidities, particularly in the case of metastatic MSA.

Immunotherapy has shown promising results in recent years [10]. Agents such as ipilimumab (anti-

CTLA-4 monoclonal antibodies), nivolumab and pembrolizumab (anti-PD1 monoclonal antibodies) have been used to target T-cell-mediated antitumor immune responses [10-15]. Imatinib mesylate has shown encouraging results in patients with KIT-mutated rectal melanoma, and polytherapies (e.g. nivolumab and ipilimumab) have proved effective in advanced melanoma [16, 17]. Sunitinib has been used to achieve complete remission in patients with KIT-mutated melanoma [18]. Radiotherapy has not been extensively studied, but has been used for haemostasis in some cases [19].

Early diagnosis and arrest are crucial factors influencing the prognosis of MSA20. Ottaviano *et al*., have proposed a flowchart for the diagnosis, staging and treatment of MSA20. Unfortunately, the prognosis for MSA is generally poor, with an estimated five-year survival rate of around 20%, regardless of the treatment modality used. The main aim of surgery is to improve the patient's quality of life, as many people with MSA already have numerous metastases at the time of diagnosis. Peri-neural invasion, as identified on histopathology, is the most important factor affecting prognosis and the likelihood of recurrence [2].

CONCLUSION

Anorectal mucosal melanoma is an extremely aggressive disease characterized by its rarity and non-specific initial presentation, making early detection difficult. Currently, surgical resection is the preferred treatment option, but there is a lack of in-depth research into the disease itself. Our aim was to contribute to the current body of MSA case studies, raise awareness of its high morbidity and mortality rates, and help develop guidelines for its management.

The diagnosis of skin cancer, including MSA, relies heavily on patient detection. However, this approach often leads to missed lesions, especially considering the obscurity of MSA. To address this problem, we propose to implement whole-body skin surveys as part of annual physical examinations. Given that skin cancers account for around a third of reported cancers worldwide, incorporating regular skin examinations can help with early detection. In addition, we recommend annual anoscopies, paying particular attention to the dentate line due to the aggressive nature of melanomas in the anorectal region.

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