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Primary Anorectal Melanoma: Report of Two Cases of a Very Rare Disease

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Abstract **Case Report**

Primary anorectal melanoma (PAM) is a rare and aggressive malignancy accounting for less than 1% of anorectal cancers and 1–3% of all melanomas. It typically presents with non-specific symptoms such as rectal bleeding, anal pain, and weight loss, often leading to delayed diagnosis. We report two clinical cases of PAM: the first in a 52-year-old man with advanced disease and poor response to immunotherapy, and the second in a 73-year-old woman treated with chemoradiotherapy due to surgical ineligibility. Diagnosis in both cases was confirmed by histopathological and immunohistochemical analyses (S100 and HMB45 positivity). Imaging revealed locally advanced tumors without initial distant metastases. These cases highlight the diagnostic challenges and lack of standardized therapeutic protocols in PAM. While surgical excision remains the mainstay of treatment, the roles of chemotherapy, radiotherapy, and immunotherapy are evolving and not yet fully established. Prognosis remains poor, with high metastatic potential and low survival rates. This report underscores the need for heightened clinical awareness and further research to optimize the management of this rare malignancy.

Keywords: Primary Anorectal Melanoma (PAM), Anorectal Cancer, Melanoma, Diagnosis, Treatment.

INTRODUCTION

Primary anorectal melanomas (PAM) are rare malignant tumors, representing less than 1% of anorectal cancers and 1-3% of malignant melanomas [1]. Their incidence is estimated to be around 0.3% of all cancers in the general population, with this particular form of melanoma typically occurring between the 5th and 7th decades of life, showing a slight female predominance [2]. Their prognosis is particularly poor due to their tendency to form early metastases, often before diagnosis is made [3]. Treatment remains controversial due to the lack of validated protocols and solid clinical studies [4]. This review presents two clinical cases of primary anorectal melanomas, discusses diagnosis, treatment, and prognostic challenges, and highlights existing therapeutic pathways.

CLINICAL CASES

Case 1:

A 52-year-old man, with no significant medical history, presents for an etiological evaluation of chronic rectal bleeding, proctalgia, and anal discharge, which have been ongoing for four months. These symptoms are accompanied by a general deterioration in health and weight loss.

Clinical Examination:

The clinical examination reveals a mass at the anal margin, measuring approximately 5 cm in its largest dimension (Figure 1). The lesion is non-bleeding on contact, and a digital rectal examination shows a non-circumferential proliferative process extending into the middle rectum.

Endoscopic and Histological Examination:

Colonoscopy reveals an ulcerated, bluish, proliferative mass, and biopsies show an undifferentiated malignant tumor proliferation with infiltration into the superficial stroma. Immunohistochemical tests show positive antibodies against S100 and HMB45 proteins, confirming a primary anorectal melanoma.

Imaging:

Pelvic MRI shows sphincter and prostatic infiltration, as well as locoregional lymphadenopathy (T4N2Mx stage). A thoraco-abdominopelvic CT scan does not reveal distant metastases. A PET scan was not

performed initially. The patient was offered abdominoperineal amputation (APA) but refused the procedure. He was put on immunotherapy, but after seven months, he developed bone metastases and passed away.

Case 2:

A 73-year-old non-smoking woman consults for symptoms of mucous diarrhea with 3-4 bowel movements daily, intermittent rectal bleeding, intense anorectal pain, and tenesmus. These symptoms are accompanied by weight loss.

Clinical Examination:

The clinical examination reveals a general deterioration in health and mucocutaneous pallor. A digital rectal exam shows bloody stools without a palpable mass.

Endoscopic and Histological Examination:

Colonoscopy reveals an ulcerated, friable, and hemorrhagic tumor located 8 cm from the anal margin, with luminal narrowing. Biopsies reveal atypical tumor cells with abnormal mitoses, suspected microinvasion of the underlying stroma, and areas of tumor necrosis. (Figure 2) Immunohistochemical study was consistent with primary anorectal melanoma.

Imaging:

Pelvic MRI reveals a rectal tumor located 8 cm from the anal margin, infiltrating the mesorectal fat and near the uterus, with multiple locoregional pelvic lymphadenopathies. (Figure 3) Thoracic, abdominal, and pelvic CT scans show no distal metastases. The tumor stage is T3N2M0.

Treatment:

Treatment was based on chemotherapy (cisplatin, fluorouracil) and external radiotherapy. Surgery was excluded, though an intervention could be considered in case of local recurrence.

DISCUSSION

Symptoms of primary anorectal melanoma are often nonspecific, delaying diagnosis. Patients may present with rectal bleeding, anorectal pain, weight loss, and tenesmus—symptoms that can easily be mistaken for more benign conditions like hemorrhoids or anal fissures [5]. Thus, it is crucial to consider primary anorectal melanoma in the context of a thorough diagnostic evaluation when persistent and unusual symptoms are observed. Diagnosis relies on biopsy and histopathological examination, which should include Fontana-Masson staining to identify melanin pigment, as well as immunohistochemical tests to confirm the presence of melanoma-specific markers (S100, HMB45, etc.) [6].

Imaging plays a critical role in determining the local extent of the tumor. Pelvic MRI and thoraco-abdominopelvic CT scans help detect local infiltrations, lymphadenopathies, and metastases [7]. However, PET scans remain controversial, though they may be useful in monitoring distant metastases [8].

Treatment of anorectal melanoma remains a complex area, mainly due to the rarity of this disease and the lack of randomized studies. Surgery, particularly abdominoperineal amputation (APA) combined with lymphadenectomy, remains the treatment of choice, although its relevance is limited when the tumor is already at an advanced stage with local or distant metastases. However, the absence of standardized treatment protocols makes management challenging [9].

Chemotherapy (cisplatin, fluorouracil) and external radiotherapy are used in cases of locally advanced tumors or recurrences. Their efficacy in curative treatment remains disputed. Additionally, immunotherapy, which has shown promising results in the treatment of other types of melanoma, is also being explored for anorectal melanoma, but its effectiveness remains to be confirmed through larger studies [10,11].

The prognosis of primary anorectal melanoma is extremely unfavorable. The five-year survival rate is less than 10%, due to rapid metastasis formation. These metastases may be local but are often distant, affecting lymph nodes, lungs, liver, and bones [12]. Early diagnosis can improve survival chances, but delayed diagnosis due to the rarity and nonspecific nature of symptoms worsens the prognosis.

CONCLUSION

Primary anorectal melanoma is a rare and aggressive condition with significant diagnostic and therapeutic challenges. Due to its low incidence, there are no standardized treatment protocols. Surgery remains the primary therapeutic option, but adjuvant treatments such as chemotherapy, radiotherapy, and immunotherapy are also used, although their effectiveness still needs further evaluation. The prognosis remains overall unfavorable, and further research is needed to improve the management of this rare and difficult-to-treat condition.

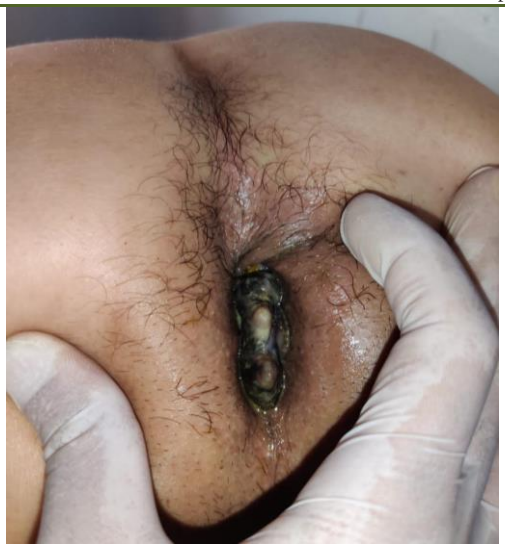

Figure 1: Mass at the anal margin, measuring approximately 5 cm

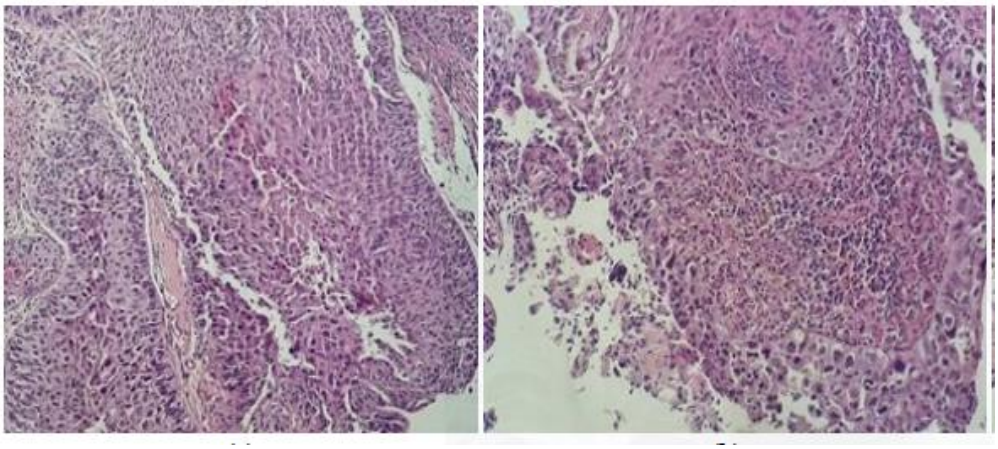

Figure 2: The tumor cells are large with anisokaryotic and hyperchromic nuclei, with irregular outlines and abnormal mitosis. The basement membrane is mostly continuous. There are localized foci of suspected microinvasion of the chorion

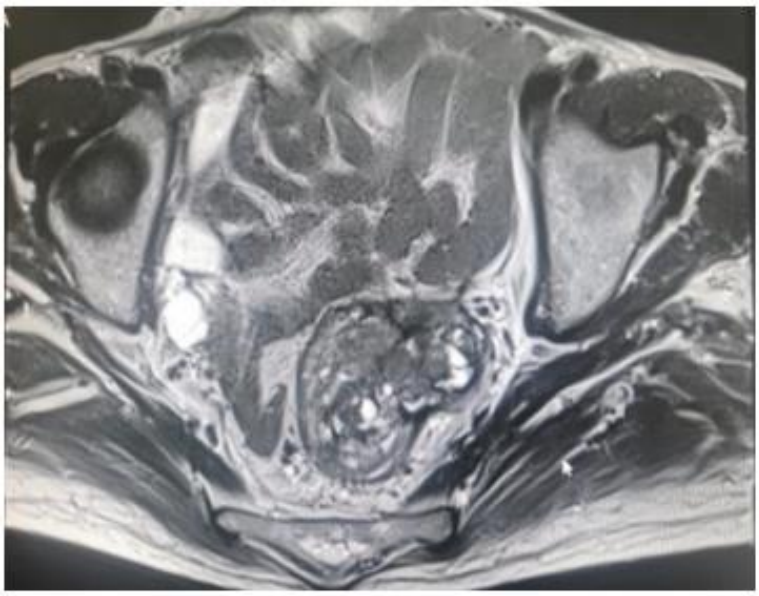

Figure 3: Abdominal imaging showing a mass evolving in the middle rectum

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Ethical Approval: Ethical approval is not required at our institution to publish an anonymous case report.

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