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Primary Anorectal Melanoma: A Rare and Aggressive Condition: A Comparative Study with other Published Series

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Abstract **Case Report**

Primary anorectal melanoma is a rare malignant tumor, accounting for less than 1% of anorectal cancers and 1–3% of all melanomas. It is characterized by marked aggressiveness and a high potential for early metastasis. Diagnosis is often delayed due to nonspecific clinical manifestations. We report two case reports illustrating the diversity of clinical presentations and treatment modalities. The first case involves a 52-year-old male patient with a locally advanced form of the disease who had an unfavorable course during immunotherapy. The second case involves a 73-yearold female patient treated with chemoradiotherapy due to a surgical contraindication. Diagnosis relies on histopathological examination supplemented by immunohistochemistry. Standard treatment protocols remain undefined. The prognosis remains poor, with very limited overall survival.

Keywords: Anorectal melanoma, Mucosal melanoma, Immunohistochemistry, KIT mutation, Surgical resection, Immunotherapy.

INTRODUCTION

Primary anorectal melanoma (PRM) is a rare tumor arising from melanocytes in the anorectal mucosa. It represents an exceptional location for mucosal melanomas, whose overall incidence remains low compared to cutaneous melanomas.

This condition primarily affects older individuals, between the ages of 50 and 70, with a slight predominance in women. PRAM is characterized by: rapid progression early metastatic spread a lack of therapeutic consensus

These characteristics explain its particularly poor prognosis.

Pathophysiology and histogenesis

MAP develops from melanocytes present in the anorectal junction. Unlike cutaneous melanomas, its development is not linked to sun exposure.

At the molecular level:

- BRAF mutations: rare
- KIT mutations: more common
- NRAS alterations: possible

These characteristics partly explain the variable response to targeted therapies and immunotherapy.

CLINICAL OBSERVATIONS

Case 1

A 52-year-old patient with no significant past medical history, presenting with chronic rectal bleeding associated with anal pain and anal discharge that has been present for four months.

Clinical examination

A 5-cm anal mass was present, firm, and non-bleeding on palpation. Digital rectal examination revealed tumor extension into the middle rectum.

Additional investigations

- **Endoscopy**: ulcerated, nodular, pigmented mass
- **Histology**: undifferentiated tumor proliferation

Immunohistochemistry: S100+, HMB45+

- **Pelvic MRI**: Sphincter and Prostate Invasion

- **Staging**: T4N2Mx

COURSE

Refusal of surgery. Treatment with immunotherapy, followed by rapid progression to bone metastases and death at 7 months.

Case 2

A 73-year-old female patient presented with mucoid diarrhea, rectal bleeding, anorectal pain, and tenesmus.

Clinical examination

Impaired general condition, pallor, no palpable mass on digital rectal examination.

Additional investigations

- **Endoscopy**: ulcerated and stenotic tumor
- **Histology**: marked atypia with numerous mitoses
- **Pelvic MRI**: mesorectal invasion
- **Stage**: T3N2M0

Treatment

Radiochemotherapy (cisplatin + 5-FU) with deferred surgery indicated in case of disease progression.

Diagnostic findings

Clinical

Symptoms are nonspecific:

- rectal bleeding
- anal pain
- tenesmus
- bowel movement disorders
- weight loss

These symptoms are often mistaken for benign conditions, delaying diagnosis.

Differential diagnosis

- hemorrhoids
- anal fissure
- rectal adenocarcinoma
- lymphoma
- rectal polyps

Histology and immunohistochemistry

The diagnosis is based on: proliferation of atypical cells presence or absence of melanin pigment immunohistochemical staining:

- S100 (highly sensitive)
- HMB45 (specific)
- Melan-A

Imaging

Pelvic MRI: key test for assessing local spread
Thoraco-abdominal-pelvic CT scan: metastatic evaluation
PET scan: useful but not routine

Therapeutic management

Surgery Standard of care:

extensive local excision abdominal-perineal resection
The choice depends on: the size of the tumor the local extent the patient's general condition

Radiotherapy

Indicated:

- as adjuvant therapy
- or in cases of unresectable tumors

Chemotherapy

Protocols based on:

- cisplatin
- dacarbazine 5-fluorouracil
- Limited efficacy.

Immunotherapy

anti-PD1 / anti-CTLA4 promising results but not yet sufficiently validated in MAP

Prognosis and prognostic factors

The prognosis is very poor:

- median survival: 12 to 24 months
- 5-year survival < 10%

Factors associated with poor prognosis:

- large tumor size
- lymph node involvement
- distant metastases
- delayed diagnosis

DISCUSSION

MAP remains a poorly studied condition due to its rarity. The two reported cases illustrate:

- the variability of clinical presentations
- the lack of a standardized treatment strategy
- the rapidly deteriorating course

Surgery remains the primary treatment, but its impact on overall survival is debated. Conservative approaches combined with systemic therapies may represent an alternative in certain cases.

Immunotherapy is a promising avenue, particularly in light of the results obtained in cutaneous melanomas, but requires studies specific to MAP.

Comparative discussion with published series

The findings observed in our two cases are generally consistent with data from the international literature, which confirm the rare, aggressive, and poor prognosis of primary anorectal melanoma (PRAM).

Clinical characteristics and time to diagnosis

Several retrospective series report that initial symptoms are dominated by rectal bleeding, anal pain, and the presence of a mass, often mistaken for benign

conditions. In a series of 19 patients, the average time to diagnosis was approximately 7.8 months.

Our observations confirm this diagnostic delay, with nonspecific initial symptoms in both cases, likely contributing to an advanced stage at the time of diagnosis.

Comparison of Surgical Outcomes

The role of surgery remains debated in the literature:

- A single-center study showed significantly longer survival after abdominoperineal resection (APR) compared to local excision (64 months vs. 11 months).
- In contrast, a recent meta-analysis including 1,858 patients found no significant difference in overall survival between radical surgery and local excision, regardless of tumor stage.
- These conflicting results perfectly illustrate the current controversy. In our series:
- the first patient refused radical surgery and had a rapidly fatal course
- the second was inoperable, illustrating the real limitations of surgical indications
- Thus, as suggested in recent studies, surgery must be individualized based on stage and general condition.

Survival outcomes and prognostic factors

Data from the literature show very limited overall survival:

- median survival: 12 to 24 months
- 5-year survival: between 10% and 20%

In a series of 56 patients:

- median survival ≈ 21–22 months
- 5-year survival ≈ 20%

These results are consistent with our first case, which was marked by a rapid fatal outcome within less than a year.

The main prognostic factors identified in the series are:

- depth of tumor invasion
- lymph node involvement
- stage at diagnosis

CONCLUSION

Primary anorectal melanoma is a rare, aggressive tumor with a poor prognosis.

Diagnosis is difficult due to nonspecific symptoms, which often lead to delayed treatment. Surgery remains the mainstay of treatment, but combined strategies incorporating immunotherapy and radiation therapy may improve the prognosis in the future.

A better understanding of this condition and multicenter studies are needed to optimize its management.

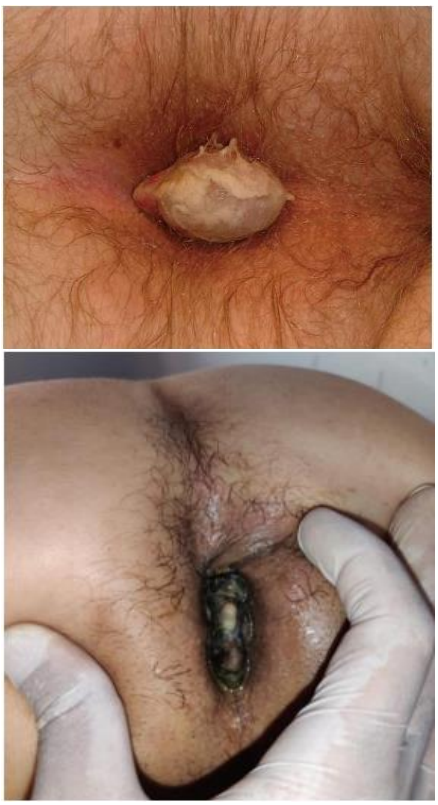

Figure 1: Mass at the anal margin, measuring approximately 5 cm

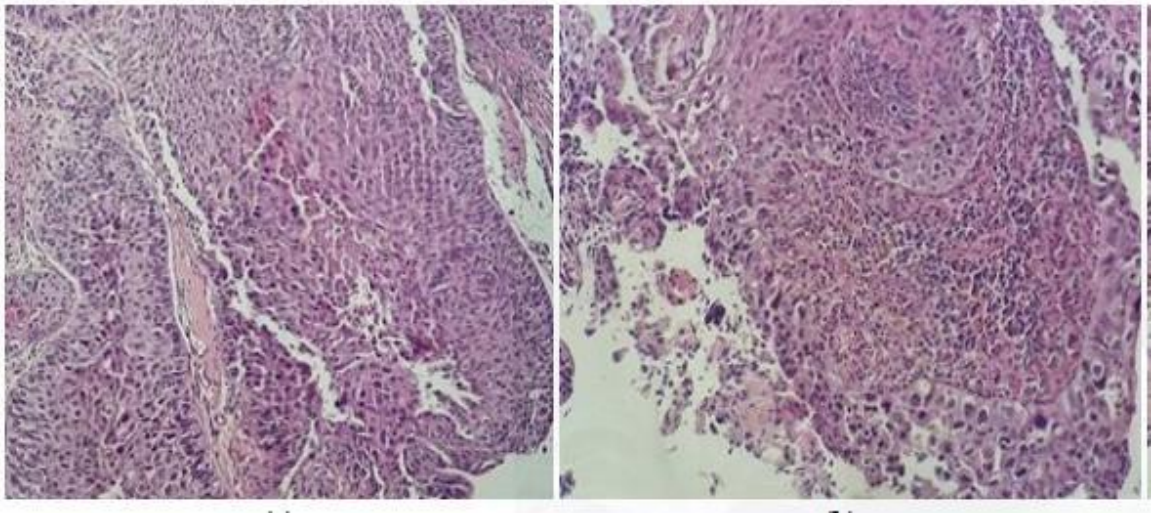

Figure 2: The tumor cells are large with anisokaryotic and hyperchromic nuclei, irregular outlines, and abnormal mitosis. The basement membrane is mostly continuous. There are localized foci of suspected microinvasion of the chorion

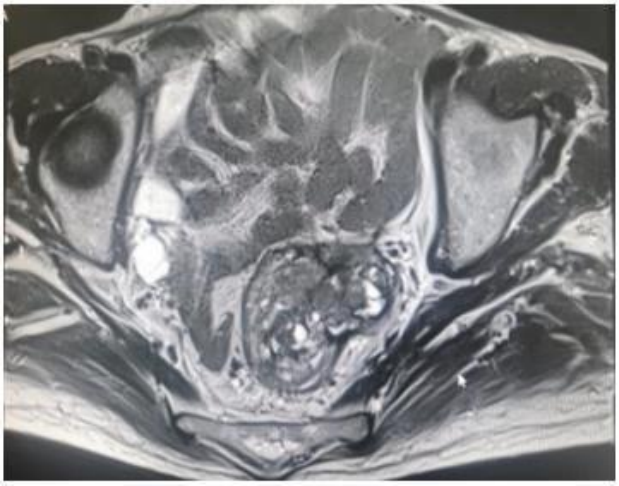

Figure 3: Abdominal imaging showing a mass developing in the middle rectum

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

REFERENCES

1. Shia J, *et al.,* Malignant melanoma of the anorectum: A 30-year experience at a single institution. Dis Colon Rectum. 2005;48(9):1707-1715. doi: 10.1007/s10350-005-0130-5.
2. Finkelstein DM, *et al.,* Anorectal melanoma: Clinical and pathological features. Ann Surg Oncol. 2014;21(3):1267-1273. doi: 10.1245/s10434-013-3389-x.
3. Lasota J, *et al.,* Immunohistochemistry in the diagnosis of anorectal melanoma. Am J Surg Pathol. 2006;30(2):163-172. doi: 10.1097/01.pas.0000171393.37046.8a.
4. Ternes N, *et al.,* Clinical management of anorectal melanoma: A review of treatment and outcomes. Surg Oncol. 2018;27(4):564-570. doi: 10.1016/j.suronc.2018.07.016.
5. Berman K, *et al.,* Outcomes in anorectal melanoma: A 15-year institutional review. J Surg Oncol. 2017;116(4):395-400. doi: 10.1002/jso.24598.
6. O'Donnell ME, *et al.,* Anorectal melanoma: A case report and review of the literature. J Clin Oncol.
7. 2010;28(4): e58-e61. doi: 10.1200/JCO.2009.24.3477.
8. Valdivieso R, *et al.,* Treatment options in anorectal melanoma: A review of the literature. J Gastrointest

Cancer. 2019;50(4):763-770. doi: 10.1007/s12029-018-00414-1.

9. Saeed H, *et al.,* Role of PET scan in anorectal melanoma: A review of the literature. World J Surg Oncol. 2017;15(1):127. doi: 10.1186/s12957-017-1254-7.
10. Jackson MJ, *et al.,* Abdominoperineal resection for anorectal melanoma: A 10-year experience. Am Surg. 2014;80(6):553-557.
11. Taal BG, *et al.,* Chemotherapy in anorectal melanoma: A systematic review. Cancer Treat Rev. 2016; 47:48-54. doi: 10.1016/j.ctrv.2016.05.003.
12. Heinzerling L, *et al.,* Immunotherapy in anorectal melanoma: New prospects for treatment. J Clin Oncol. 2017;35(5):1845-1853. doi: 10.1200/JCO.2016.69.1532.
13. Ternes N, *et al.,* Prognostic factors in anorectal melanoma: A multi-center study. Ann Surg Oncol.
14. 2019;26(5):1389-1395. doi: 10.1245/s10434-019-07527-2.