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IP Journal of Diagnostic Pathology and Oncology

Journal homepage: https://jdpo.org/

Case Report

Anorectal melanoma manifesting with bone metastases: A clinico-pathological surprise

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Abstract

Anorectal melanoma is a rare malignancy that is known to metastasize in the liver, followed by the lungs. Very rarely it can metastasize into bone, like in our case, which was an elderly man presenting to us with lower back pain and was thus clinically thought to be a case of metastatic prostatic carcinoma. However, radiological details and histopathological examination of the bone marrow biopsy revealed it to be anorectal melanoma with bone metastasis.
A 65-year-old male presented with lower backache for four months. Radiological imaging revealed marrow lesions in the lumbosacral-iliac region and right femur head. A bone marrow biopsy was performed. Its examination revealed sheets of markedly pleomorphic tumor cells containing brownish-black intra-cytoplasmic pigment suggesting metastatic deposits of melanoma. Immunohistochemistry further confirmed the diagnosis. The diagnosis of metastatic melanoma was rendered. Further workup disclosed the primary to be in the anorectal region. Thus, the case was concluded to be that of anorectal melanoma metastasizing to bone.

Keywords: Anorectal melanoma, Melanoma, Malignant melanoma, Anorectal region, Bone, Bone metastasis.

Received: 21-09-2025; **Accepted:** 21-10-2025; **Available Online**: 29-10-2025

1. Introduction

Anorectal melanoma (ARM) is a rare malignancy accounting for ~1.3% of all melanomas.[1] It often presents with gastrointestinal symptoms like anal-pruritus, rectal-pain, bleeding per-rectum, etc.[2,3] Other symptoms include rectal fullness, change in bowel habits and movements, and inguinal masses.[2] We put forward a rare presentation of ARM, which presented to us with lower back pain due to bone metastases in the lumbo-sacral-iliac region. It was initially thought to be a case of prostatic carcinoma with bone metastasis. But as the case workup progressed, the case emerged to be that of anorectal melanoma metastasizing to the bone.

2. Case Report

A 65-year-old male presented with lower backache for the past four months. His digital rectal examination and laboratory parameters including serum prostate-specific antigen (0.85ng/mL) were within normal limits. Magnetic resonance imaging (MRI) of the lower lumber spine revealed marrow lesions in lumbosacral-iliac region, and right femur head (**Figure 1**A). A bone marrow biopsy was performed. It revealed sheets of markedly pleomorphic tumor cells displaying brownish-black pigment in their cytoplasm (**Figure 1**B). On immunohistochemistry, tumor cells were positive for S100 and HMB-45 (**Figure 1**C-D). Thus, the diagnosis of metastatic melanoma was rendered.

A computerized tomography (CT) scan of the whole abdomen was then performed. It disclosed an ill-defined mass in the mid and lower rectum measuring 6.1x5.2x5cm,

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https://doi.org/10.18231/j.jdpo.20117.1761718895

along with right common-iliac station and retroperitoneal lymphadenopathy. Multiple lesions were noted in the bilateral liver lobes. Thus, the case was concluded to be that of anorectal melanoma with multiple metastases, including that in the bone.

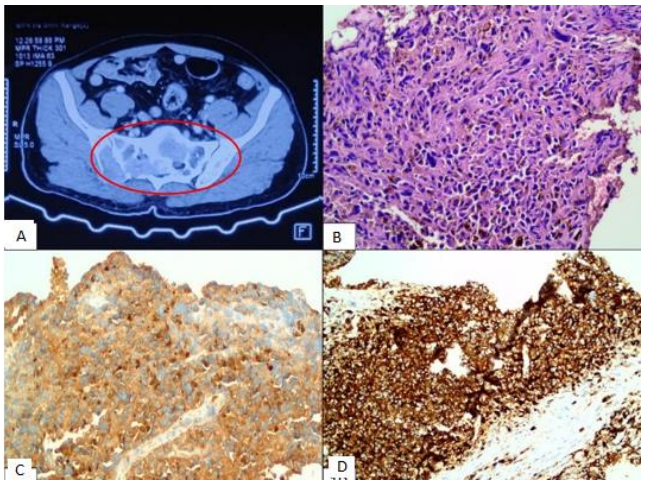

Figure 1: Bone metastases of ARMM; **A:** Magnetic Resonance Imaging (MRI) of lower lumber spine showing bony metastatic lesions (marked by circle); **B:** Bone marrow biopsy showing tumor cells displaying markedly pleomorphic nuclei, prominent nucleoli, and moderate amount of melanin containing cytoplasm (Hematoxylin & Eosin stain;200X); **C&D:** Immunohistochemistry displaying S-100(200X) and HMB-45(200X) positivity.

3. Discussion

Anorectal melanoma accounts for only ~16.5% of all mucosal melanomas, ~0.05% of all colorectal malignancies and 1% of all anal carcinomas.[1,4] They are mostly seen within 6 cm of the anal rim. However, they can occur anywhere in the anorectum.[5] Male: female ratio is 1:1.6-2.3.[2] They often present with organ specific symptoms like anal pruritus or rectal pain.[3] However, other symptoms like rectal fullness, change in bowel habits/movements, and inguinal masses may also draw medical attention.[2]

Colonoscopy and endoscopic ultrasound (EUS) help in tumor diagnosis and staging. Histopathological examination with immunohistochemistry is required for the diagnosis and is the gold standard. On histopathology, the tumor cells are epithelioid to spindle shaped, displaying markedly pleomorphic nuclei, prominent nucleoli and a variable amount of intracytoplasmic brownish-black melanin pigment. This pigment may be absent in ~40% cases of ARM.[6] On immunohistochemistry, tumor cells are positive for S100, HMB45, and Melan-A/Mart-1. Markers like BRAF and CD-117 further help to detect their specific mutations, which can dictate the targeted therapy. In ARM, CD117 mutations are seen more often than BRAF mutations.[1]

ARMs are staged as stage I, II, or III depending upon the extent of disease involvement: stage I is local disease; stage II is local disease with regional lymphadenopathy; and stage III is distant metastasis.[7] Liver metastasis is most common, followed by that in the lungs and bones.[2] Our case is one such rare case with bone metastasis, which led to the disease presentation. Prasad et al. categorized mucosal melanomas into levels 1 to 3 being localized tumor, regional tumor with laminal invasion, and disseminated disease, respectively.[8] The median survival varies with the level of involvement which is 138 months for level 1 and only 17 months for level 3. However, the prognosis is usually poor as ARMs usually present at the advanced stages.[2]

Surgical excision forms the typical treatment and may be abdomino-perineal resection (APR), wide local excision (WLE), or endoscopic mucosal resection (EMR). Further radiotherapy; chemotherapy; immunotherapy with interferon-α, interleukin-2, CTLA-4, or PD-1/PDL-1 inhibitors; targeted therapy for BRAF and KIT pathways; and antiangiogenetic therapy can be used. 5-year survival rate for ARM is <20%.[2] Factors like tumor size, depth of invasion, treatment modality, symptomatic duration, disease stage, lymphadenopathy, and molecular markers like proliferating cell nuclear antigen (PCNA) and Ki-67 proliferation index decide the prognosis.

4. Conclusion

Anorectal melanomas are rare and usually present with organ-specific symptoms like rectal pain and bleeding per rectum. Very rarely they may come to notice due to non-specific complaints like backache, which results from bone metastasis. Bone metastasis in ARMs is a very rare occurrence and, in such scenarios, deciphering the correct diagnosis is crucial for the proper patient management. Thus, this rare presentation of rare anorectal melanoma should always be suspected when coming across clinico-radiological features as described above in our case.

5. Conflict of Interests

No conflicts to declare.

6. Ethical Approval

Not Applicable.

7. Source of Funding

None.

8. Acknowledgements

Not Applicable.

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Cite this article: Singh A, Singh A, Arya A. Anorectal melanoma manifesting with bone metastases: A clinico-pathological surprise. *IP J Diagn Pathol Oncol*. 2025;10(3):138-140.