



Anorectal Mucosal Melanoma: A Case Series

Jeremy Polman DO MS MBA, Gabrielle Sanford MD, Marshall Stagg MD, Rameela Mahat MD

Baton Rouge General Internal Medicine Residency Program



Introduction

- Anorectal Mucosal Melanoma (ARM) is a rare and aggressive malignancy that is poorly described and infrequently studied.
- ARM differs significantly from cutaneous melanoma in presentation, genetic profile, staging, treatment response, and progression patterns.
- ARM is a challenge for physicians because of its rarity, unclear pathogenesis, non-specific presentation, and aggressive disease course leading to an inferior prognosis.
- This report presents three clinical cases, all of which presented with relatively benign symptoms.

Case Descriptions

- The first patient is a 73 year old Caucasian male with a history of external hemorrhoids who presented to his primary care provider after developing rectal pain that did not resolve with sitz baths and over the counter topical therapy. Physical exam revealed three masses, believed to be hemorrhoids. The masses were then excised and sent for pathology, which revealed ARM.
- The second patient is a 63 year old Caucasian female with a past medical history of psoriatic arthritis who presented to the hospital with one month of intermittent, painless hematochezia during defecation. Physical exam findings were remarkable for a large anterior anal mass on digital rectal examination. Colonoscopy revealed a 2 cm submucosal, actively bleeding, friable, non-pigmented mass on a broad stalk in the anal canal that extended to the dentate line [Fig 1]. Colorectal surgery performed a full-thickness trans-anal resection of the mass. Immunohistochemical studies were positive for SOX-10 and HMB-45 [Fig 2, 3], confirming ARM.
- The third patient is a 71 year old African American male with a past medical history of cerebrovascular accident with residual ataxia who presented to his gastroenterologist with worsening painless hematochezia. The patient describes taking aspirin and clopidogrel secondary to recent cerebrovascular accident. Since initiation of these medications, the patient has noticed hematochezia three to four times a week. The patient underwent colonoscopy, which revealed a 14 mm oozing, polypoid, semi-pedunculated lesion in the distal rectum [Fig 4].

Figures

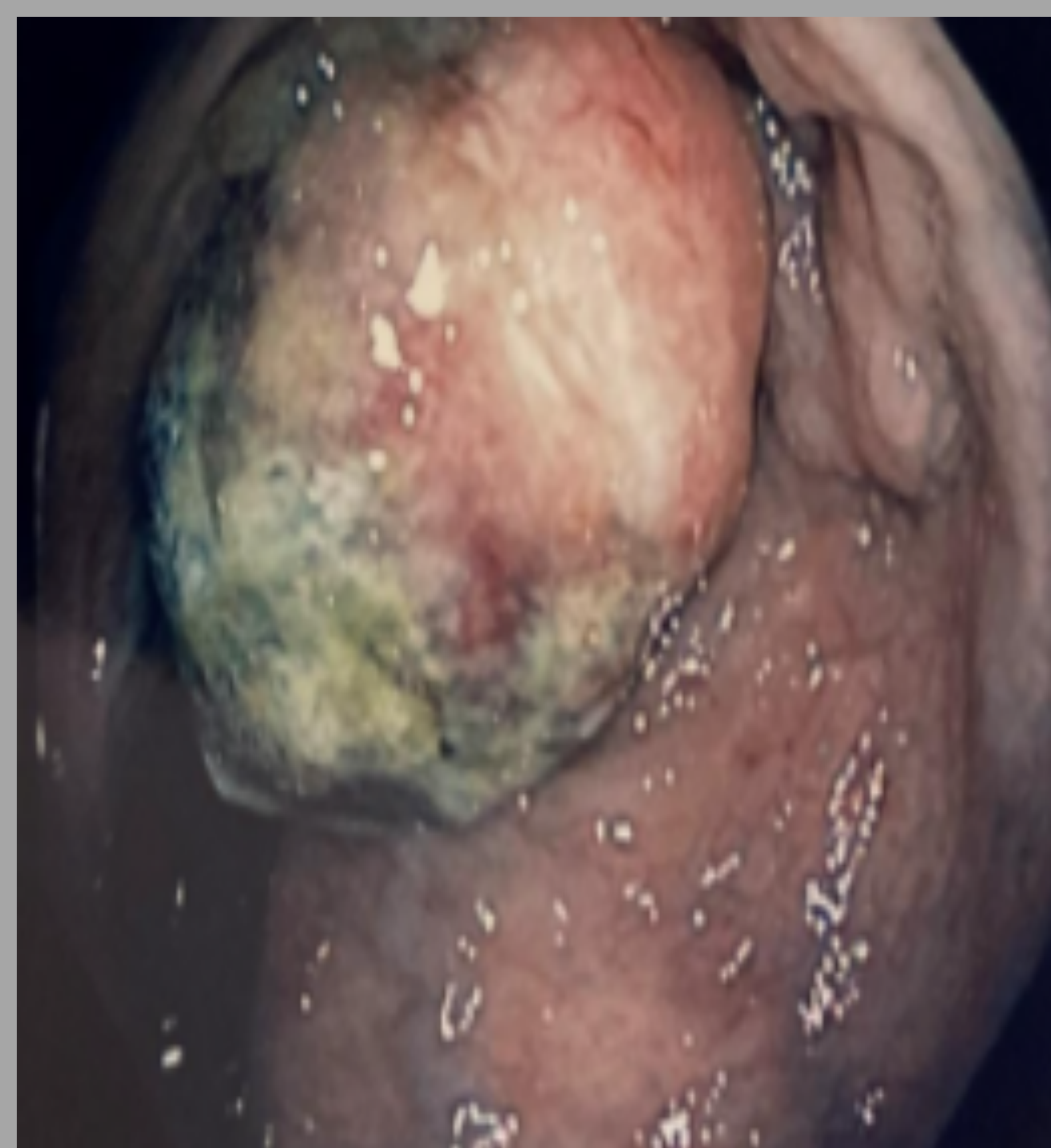


Figure 1: 1x2cm submucosal, bleeding mass in anal canal

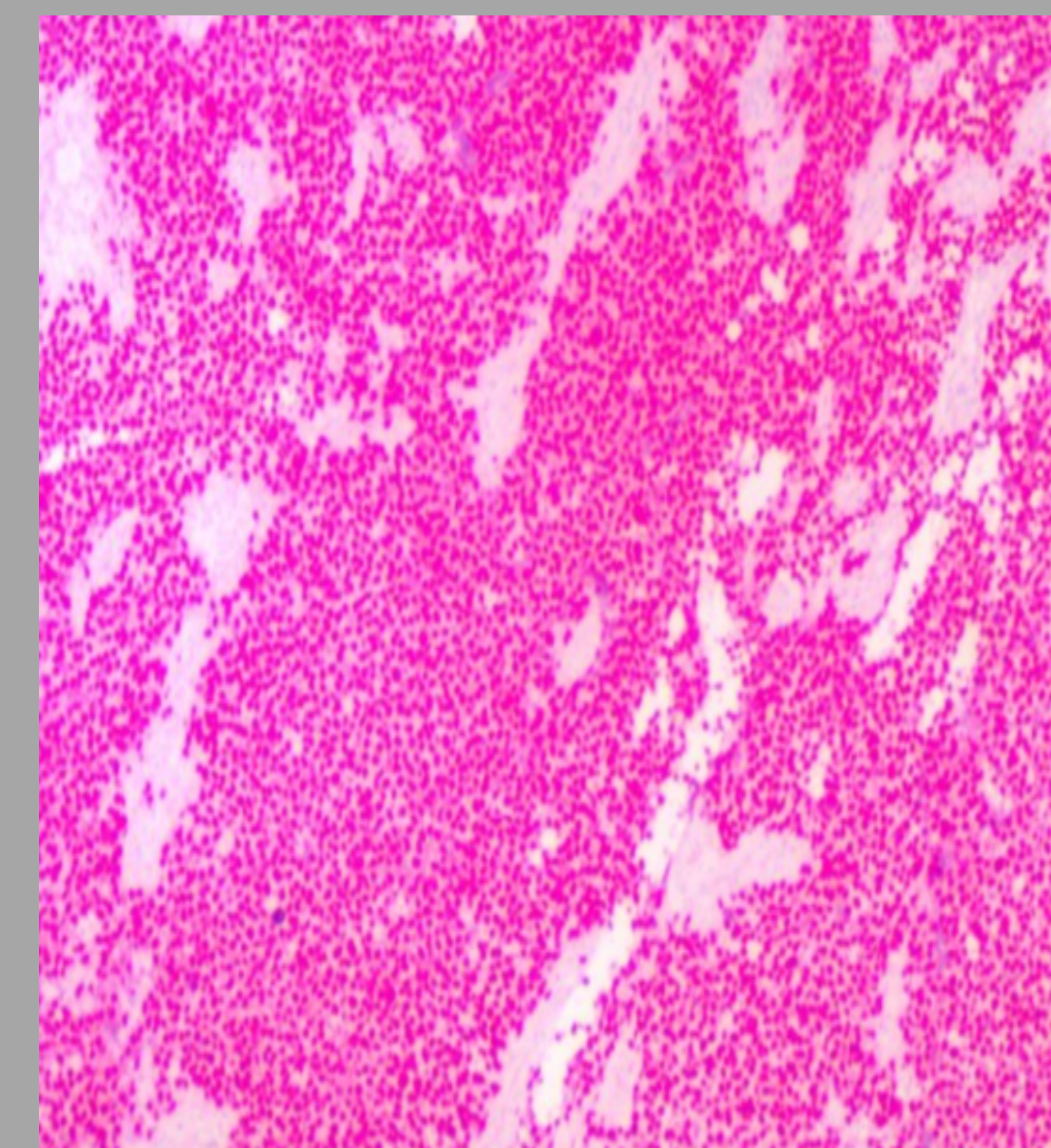


Figure 2: Positive immunohistochemical stain for SOX-10, a common marker for malignant melanoma

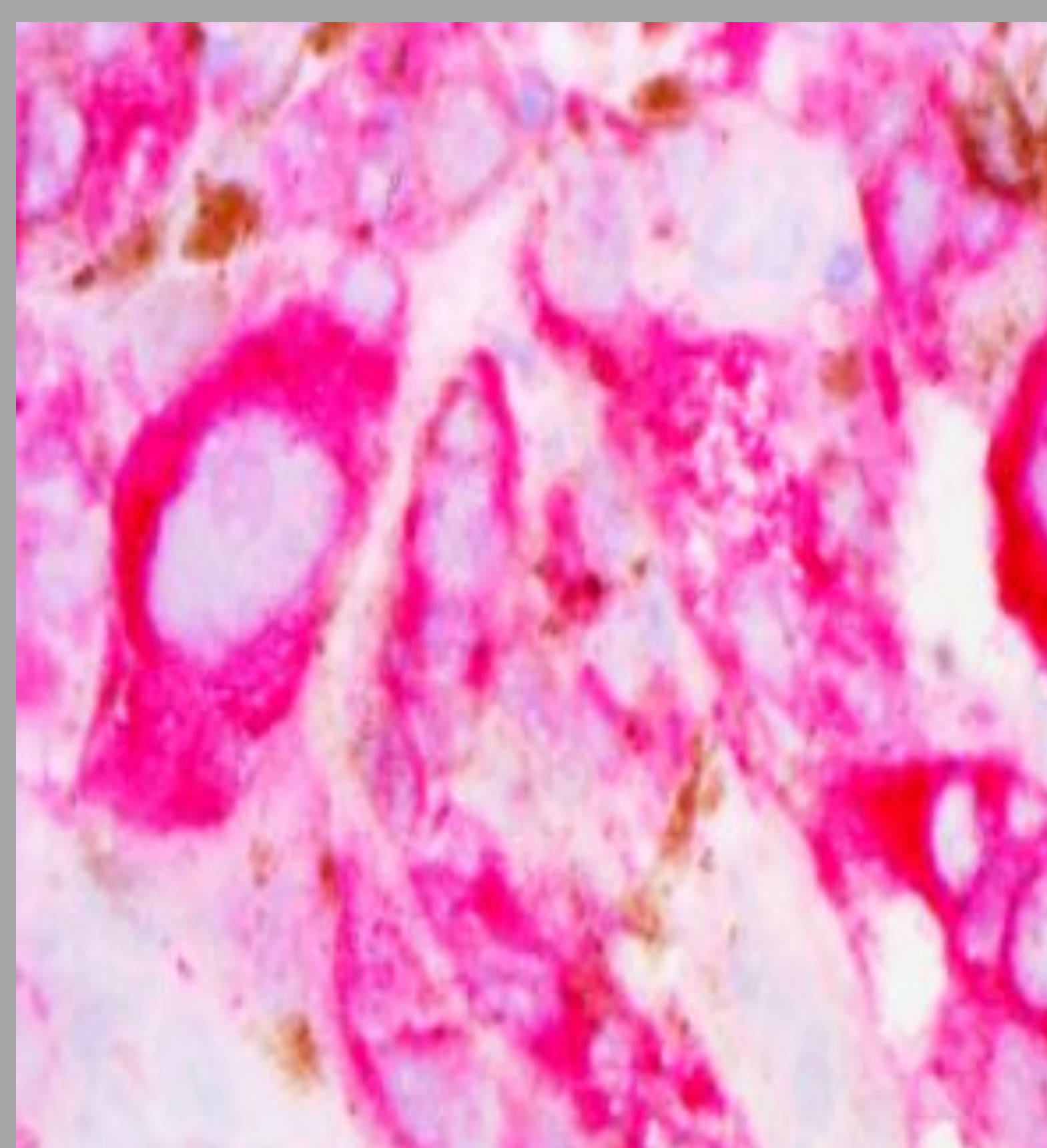


Figure 3: Positive immunohistochemical stain for HMB-45



Figure 4: 14mm polypoid, semi-pedunculated, oozing mass in distal rectum

Discussion

- Malignant melanomas develop from melanocytes that originate from pluripotent neural crest stem cells.
- Rectal bleeding is the most common initial symptom of ARM, but tenesmus, incontinence, change in bowel habits, anorexia, and weight loss may also be seen [1].
- The lesions on examination are usually firm, polypoid, submucosal, lack typical melanoma pigmentation in up to 80%, and are amelanotic in 20-30% of the cases.
- ARM presenting in a non-specific fashion is rare, aggressive, and may be amelanotic [2].
- The lesions are often mistaken for hemorrhoids or polyps, leading to delayed diagnosis. These factors, along with the location of the disease, may lead to diagnostic delay or misdiagnosis.
- Consequently, most ARM cases are locally advanced or metastatic at the time of diagnosis, resulting in an overall 5 year survival rate around 10-20% [3].
- Therefore, a high index of suspicion leading to a biopsy is needed to find early-stage disease.

Conclusion

Increased awareness of ARM with an increased index of suspicion and a low biopsy threshold are needed to make an early diagnosis. Approaching lesions in this fashion may improve outcomes by helping to mitigate the high burden of late diagnoses and advanced disease.

References

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