

Introduction

Anorectal mucosal melanoma (ARM) is an uncommon and aggressive malignancy that is often found at advanced stages of the disease, primarily due to its non-specific presentation.

Primary ARM accounts for less than 2% of all melanomas. Because of its rarity, it is often misdiagnosed as benign conditions such as hemorrhoids or polyps on initial exam, thus leading to a delay in diagnosis and treatment.

Here, we present a 73 year old male with a rectal mass originally diagnosed as hemorrhoids, who was subsequently diagnosed with ARM.

Case Report

A 73-year-old Caucasian male with a past medical history of external hemorrhoids presented to his primary care provider after developing rectal pain that did not resolve with sitz baths and over-the-counter topical therapy.

A physical exam revealed three friable circumferential masses, believed to be nonreducible hemorrhoids.

The patient was then evaluated by colorectal surgery for hemorrhoidectomy. Following hemorrhoidectomy, pathology report revealed ARM with positive margins and lymphovascular invasion.

Immunohistochemistry performed showed diffuse immunoreactivity for melan-A and SOX10 [A, B]. There was no BRAF mutation detected. A PET CT performed was negative for metastatic disease and lymphoscintigraphy did not visualize a sentinel node. It was determined that patient had locally invasive disease without metastasis and that surgical therapy was indicated.

One month after diagnosis the patient elected to undergo abdominoperineal resection (APR) with colostomy. During surgery, there was what appeared to be a small, residual malignant nodule in the right anal canal that was resected as well as the entire anal canal during the procedure. Pathology showed negative margins in the sigmoid, rectum and 12 lymph nodes. One specimen contained melanoma in situ and invasive melanoma involving the anus [C].

The patient recovered well after surgery and began adjuvant pembrolizumab with close follow up.

Figures

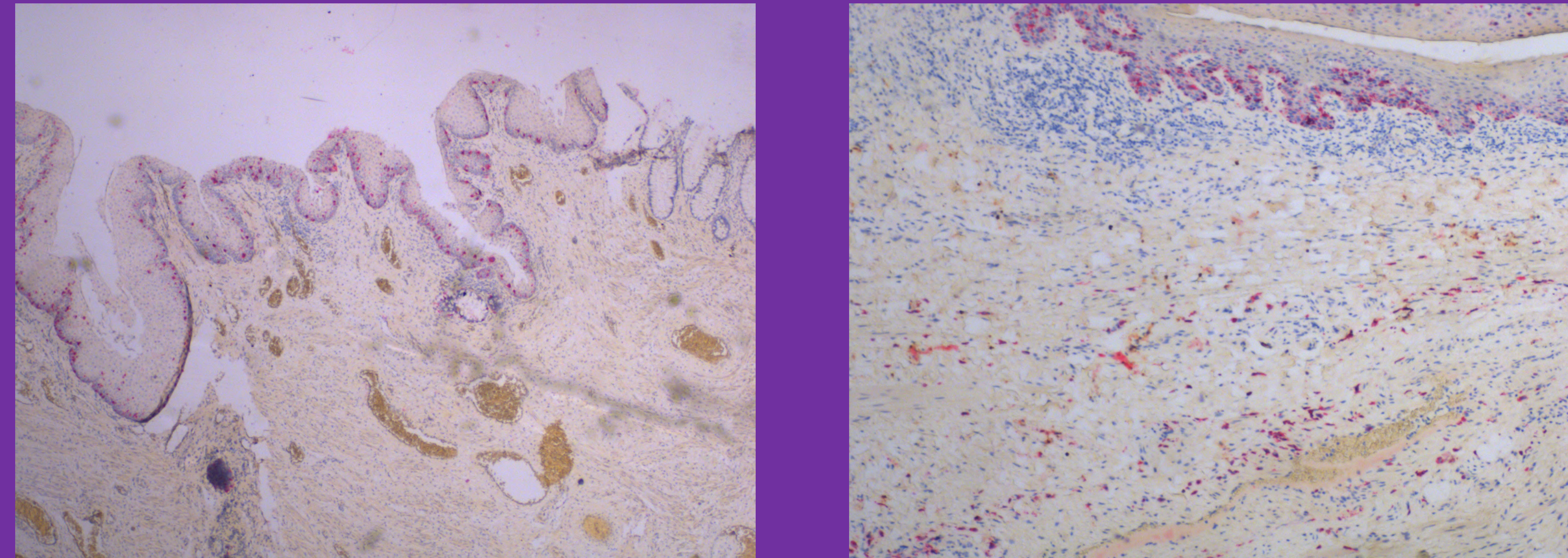


Figure A, B: Immunohistochemical stain demonstrating positivity for melan-A, confirming the diagnosis of anorectal mucosal melanoma

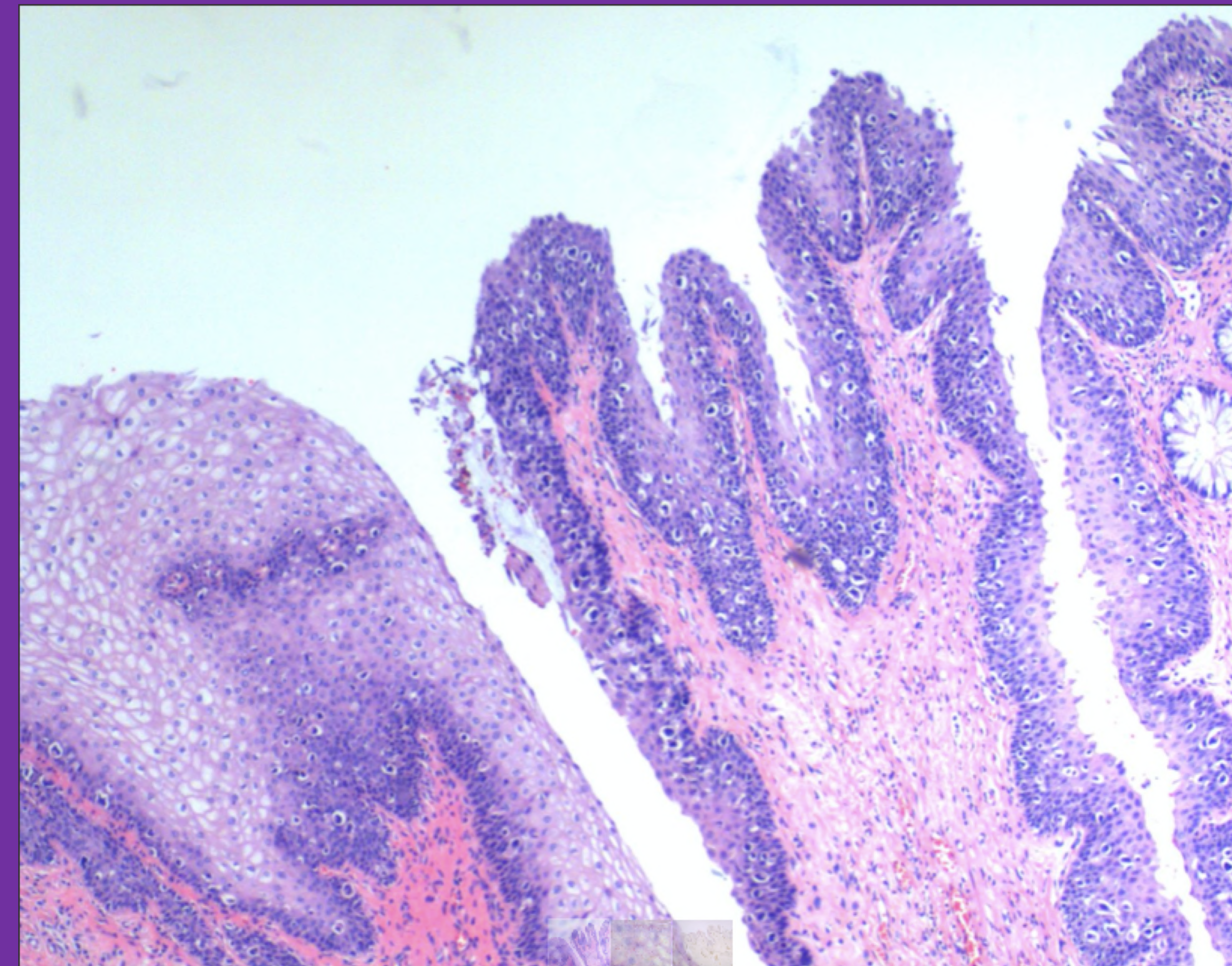


Figure C: Histopathology demonstrating melanoma in situ post hemorrhoidectomy and abdominal perineal resection

Discussion

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.

The lesions on examination usually lack typical melanoma pigmentation in up to 80%, and are amelanotic in 20-30% of the cases.

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.

This diagnostic delay results in an overall 5 year survival rate around 10-20%.

Conclusion

This case highlights the importance for clinicians to maintain a high index of suspicion for ARM and should have a low threshold to biopsy suspicious lesions.

Increasing awareness for ARM will hopefully improve the time to diagnosis and in turn, improve survival and quality of life.

References

- [1] McBrearty, A., D. Porter, and K. McCallion. "Anal melanoma: A general surgical experience." *J Clin Case Rep* 5.493 (2015): 2
- [2] Felz MW, Winburn GB, Kallab AM, Lee JR. Anal melanoma: an aggressive malignancy masquerading as hemorrhoids. *South Med J*. 2001 Sep;94(9):880-5. PMID: 11592745.
- [3] Stefanou A, Nalamati SP. Anorectal melanoma. *Clin Colon Rectal Surg*. 2011
- [4] Weinstock MA. Epidemiology and prognosis of anorectal melanoma. *Gastroenterology*. 1993 Jan;104(1):174-8. doi: 10.1016/0016-5085(93)90849-8. PMID: 8419240.
- [5] Brady MS, Kavolius JP, Quan SH. Anorectal melanoma. A 64-year experience at Memorial Sloan-Kettering Cancer Center. *Dis Colon Rectum*. 1995 Feb;38(2):146-51. doi: 10.1007/BF02052442. PMID: 7851168.