A rare case of amelanotic anorectal melanoma

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ABSTRACT

We report an exceedingly rare case of amelanotic anorectal melanoma misdiagnosed as hemorrhoids. A 74-year-old man presented with a week's history of "blood on toilet tissue" without bowel complaints or history of cutaneous melanoma. Skin evaluation was normal. Rectal exam was negative for blood, but revealed an anal nodule, interpreted as a hemorrhoid. Hemoglobin was normal; bleeding persisted. After one month, colonoscopy detected a non-pigmented anal lesion. Biopsy showed melanoma.

Noncutaneous mucosal melanoma represents 0.03% of new cancer diagnoses Anorectal melanoma accounts for 1% of melanomas and 0.4% of anal malignancies. Uncommonly, this malignancy lacks melanin pigment, complicating detection. Presenting complaints are non-specific rectal bleeding, pain, itching or incontinence, mimicking more common disorders.

Dangerous misdiagnosis occurs when benign disease, not malignancy, is suspected. Risk factors for cutaneous melanoma are less frequent. Mucosal melanoma has different genetics. Clinicians must be aware of diagnostic difficulties of anorectal melanoma, especially when amelanotic.

KEYWORDS: anorectal melanoma, GI bleeding, mucosal melanoma

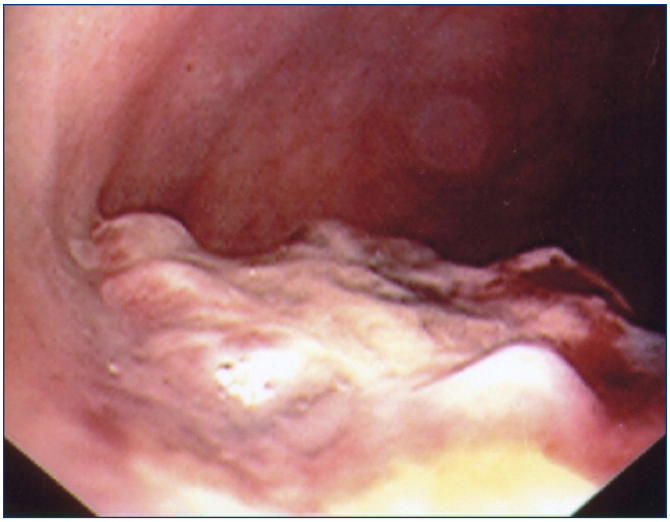
INTRODUCTION

Noncutaneous mucosal melanoma, including ocular, anorectal, vaginal and nasal sites, is rare, representing 0.03% of all new cancer diagnoses based on data from the National Cancer Data Base Report on Cutaneous and Noncutaneous Melanoma.[1] Anorectal melanoma accounts for as few as 1% of melanomas and 0.4% of anal malignancies.[2,3] Uncommonly, this malignancy lacks melanin pigment (amelanotic melanoma), complicating detection.[4] This tumor manifests as non-specific symptoms, including small amounts of rectal bleeding, pain, itching or incontinence, mimicking more common disorders with potential catastrophic delayed diagnosis. We describe an exceedingly rare case of amelanotic anorectal melanoma initially misdiagnosed as hemorrhoids.

CASE REPORT

A 74-year-old man presented with a one-week history of "flecks of blood on the toilet tissue" after defecation without bowel complaints, history of cutaneous melanoma or current skin complaints. Physical examination including vital signs, skin and abdominal exams, was normal. However, on rectal exam, a firm nodule was detected just inside the anal verge and interpreted as an internal hemorrhoid. (**Figure 1**) Rectal exam at presentation was negative for occult blood. Hemoglobin was normal, but bleeding persisted. After one month, colonoscopy was performed. A 2 cm, non-pigmented, anal lesion was biopsied, revealing melanoma on immunohistology.

Figure 1. A firm, amelanotic anorectal melanoma.



DISCUSSION

This rare malignancy arises from melanocytes, most commonly found in the skin, but also in ocular or mucosal sites.[5] Mucosal and cutaneous melanomas are distinct diseases with discrete genetic features. Mucosal melanoma is characterized by a distinct pattern of chromosomal aberrations.[6] Any mucosal site may be affected, but most arise from the vulvovaginal and head and neck mucosa; the anorectum is the 3rd most common site for mucosal melanoma, occurring nearly equally in the anal canal, rectum, or anorectum.[7] Mucosal melanoma has been documented rarely in the mucosa of other tissues including the male urethra, gallbladder, esophagus and large and small bowel.[7] Often discovered at an advanced stage, it is usually found in older patients with chronic disease; half present at age 70 or older. The 5-year survival rate is as low as 6%.[4,8]

Risk factors for cutaneous melanoma, including light skin, exposure to ultraviolet radiation, family history of melanoma,

are not as well documented for mucosal anorectal melanoma. Cutaneous melanoma is 5 to 20 times more frequent in Caucasians whereas anorectal melanoma is only twice as common in Caucasians.[9] In the United States, fewer than 3% of skin melanomas occur in African Americans and dark-skinned Latinos; however, these groups represent as many as 9% of individuals diagnosed with mucosal melanoma.[1]

Additionally, recent data support an association of anorectal melanoma with HIV in men who have sex with men.[10] A recent study reported a family history of cutaneous melanoma which was associated with the development of a mucosal melanoma.[11]

Anorectal melanoma is commonly asymptomatic until a late stage or manifests as non-specific, sometimes vague symptoms. The most common clinical presentation, as in our patient, is rectal bleeding. Other initial complaints may be rectal pain, sensation of rectal fullness or a mass, constipation or diarrhea, tenesmus, anal pruritis, or vague, non-specific complaints ignored by both patient and physician. Incontinence and iron deficiency are rare, but reported. Fatigue and weight loss may be reported by individuals with widespread metastatic disease.[3]

Dangerous misdiagnosis and delayed treatment occur when benign diseases, including hemorrhoids, skin tags or small, benign polyps, are suspected rather than malignant melanoma or another malignancy. Other factors delaying diagnosis include: (1) Rarity of anorectal melanoma which is not associated with prior or contemporaneous skin malignancy so that diagnostic suspicion is low; (2) Amelanotic lesions, as in our patient, occur infrequently, although exact data does not exist[4]; (3) Although indicated at clinical presentaton, careful anorectal digital exam and anoscopy are commonly not performed; (4) Frequent atypical macroscopic appearance ranging from a firm polypoid lesion to hemorrhoid-like, pigmented or amelanotic ulceration.[2] Misdiagnosis of anal melanoma as more common malignancies, such as carcinoma, lymphoma, or angiosarcoma, would be evaluated urgently, revealing the unsuspected anorectal melanoma in a timely manner.

Zhang et al. reported misdiagnosis at presentation in more than half of their patients, 46 of 79 (58 %).[2] In this study, the maximal diameter of tumor in the misdiagnosed group was longer than in the not-misdiagnosed group (4.06 vs. 3.45 cm). Distant metastasis at diagnosis in the delayed diagnosis group was more common than in the not-delayed group (30.4% vs. 24.2%). Of incorrect diagnoses, half were misdiagnosed as carcinoma, half as benign lesions. The danger is that benign anorectal diseases do not generally require urgent evaluation and are prone to being neglected by both patient and doctor. Pessau_and colleagues reported a mean delay in diagnosis of 6 months in this context.[12]

Our discussion has a specific limitation. Anorectal melanoma is an extremely rare sub-type of melanoma. The majority of reports in the medical literature describe a single case report or a very small case series. Thus, some rates and percentages may be imprecise due to small sample size studies or different methodology and varied nomenclature defining tumor localization in the few large sample size database publications.

CONCLUSION

Although rare, clinicians must be aware of the diverse clinical spectrum and diagnostic difficulties associated with anorectal melanoma, especially when amelanotic. This tumor can mimic refractory hemorrhoids or other benign perianal disorders. Primary mucosal anorectal melanoma should be considered in selected patients when an anal or anorectal mass is discovered. Bowel or rectal complaints of any type in adults with a current or past history of melanoma should prompt evaluation of possible metastatic disease. Despite its rarity, anorectal melanoma should be considered when unusual or unclear anorectal lesions are detected.

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