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Anorectal Melanomas: Case Series

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INTRODUCTION

Melanoma is the most common tumor to metastasize to the gastrointestinal (GI) tract. 60% of patients who die with malignant melanoma have GI involvement at autopsy¹. Symptomatic disease presenting during life is unusual. Anorectal melanoma, a rare tumor that represents 1% of all melanomas and less than 4% of all anal malignancies, manifests with a range of non-specific symptoms and physical exam findings². In fact, about one-third of lesions may be amelanotic^{3,4}. Initial misdiagnosis as a benign lesion such as hemorrhoid is common.

We report 3 cases of anorectal melanoma presenting as primary GI complaints all without known cutaneous malignancy. Physicians must be aware of the clinical spectrum, anatomic distribution, and diagnostic difficulties associated with anorectal melanoma.

CASE PRESENTATION

Case 1

A 48 year-old woman presented with 1 week of bright red blood per rectum described as small amounts of blood surrounding otherwise brown stool. Physical exam was normal except for a firm mass on rectal exam. Hemoglobin was normal. Colonoscopy showed a 2-3 cm black mass inside the rectum at the dentate line. Histology was consistent with melanoma. A transanal excision documented a 4-5 mm thick melanoma invading the muscularis mucosa with vascular invasion. Surveillance chest radiograph 18 months later revealed lung metastasis. She had resection of the right lower lobe and right middle lobe. No cutaneous lesion was ever noted on her skin exam.

Case 2

An 84 year-old woman presented with several weeks of intermittent painless rectal bleeding. Initial rectal exam showed a hemorrhoid-like lesion. Hemoglobin was normal. She was treated symptomatically with hydrocortisone cream. Because her bleeding persisted after 3 weeks of treatment, further evaluation was performed. A non-pigmented tumor inside the rectum was discovered on retroflexion. Immunohistochemical staining of the lesion was strongly positive for S-100 and focally positive for HMB-45, both markers sensitive for melanocytic tumors. Transanal excision was performed. After a symptom-free interval of 2 years, intracerebral metastases were discovered. No cutaneous lesion was ever noted on skin exam.

Case 3

A 74 year-old woman presented with worsening fecal incontinence over several months complicated by sharp rectal pain, 25-pound weight loss, anorexia, constipation, tenesmus, and intermittent rectal bleeding. She described bleeding as flecks of blood on toilet tissue after defecation without gross blood in the stool. Family history was significant for head and neck cancer, breast cancer, and lung cancer. A non-pigmented 3 cm anorectal mass was found at colonoscopy. Biopsy was positive for melanoma. She underwent an abdominoperineal resection with colostomy. Distant spread of tumor was not identified in the initial post-op evaluation; skin exam was negative for melanoma both prior to and at the time of diagnosis.

DISCUSSION

Compared to cutaneous melanoma, non-cutaneous melanoma is rare and located in ocular sites or sites such as anorectal, vaginal, and nasal mucosa. The anorectum is the third most frequent mucosal site for melanoma, after head and neck and the female genital tract. Mucosal melanoma occurs predominantly in older individuals; 49% present at age 70 or older⁵. A recent report, however, by Ellis and co-workers described cases of an 11-year-old and a 19-year-old patient with anorectal melanoma⁶.

Race, ethnicity, and gender differences exist. In the U.S., fewer than 3% of cutaneous melanomas are diagnosed in African Americans and Hispanics; however, these minority groups represent approximately 9% of individuals diagnosed with mucosal melanoma⁵. Women are more likely than men to be diagnosed with anorectal melanoma (male: female ratio 3:4). Sun exposure is not a risk factor for mucosal melanoma². Collected data suggest an increased risk in HIV-positive patients⁷.

Presenting complaints are non-specific and mimic common non-neoplastic disorders such as hemorrhoids. As described in each of our cases, rectal bleeding was the most frequent complaint followed by mass sensation and tenesmus. Other symptoms include anal pruritus, changes in bowel habits, and rectal pain or prolapse. Prior to histology, other possible malignant diagnoses include epidermoid carcinoma, basal cell carcinoma, adenocarcinoma, and more unusual tumors – anaplastic sarcoma, lymphoma, and undifferentiated cylindrical cellular carcinoma. The macroscopic appearance of anorectal melanoma ranges from a polypoid mass to a pigmented or non-pigmented ulceration^{4,8}.

Diagnosis is often delayed because most symptoms are non-specific or thorough anorectal examinations, including anoscopy, are not performed. Identifiable lesions are pigmented in about two-thirds of cases^{3,4}. Commonly, as in each of our three cases, no skin lesions are noted. One case series suggests that a benign anorectal condition was the initial diagnosis in 46 of 79 patients⁹. Cases misdiagnosed as hemorrhoids had a statistically significant decrease in survival with the average duration of symptoms before diagnosis being between 4 and 6 months. In a review of mucosal melanoma, one study found that anorectal melanomas had been misdiagnosed in about two thirds of patients – most commonly as hemorrhoids, adenocarcinoma, or polyp¹⁰.

Terminology varies depending on which system is used. One popular classification uses the following criteria: melanoma is termed by location – anal if below dentate line, rectal if above, and anorectal if around. No one schema is accepted or used universally. Purely rectal melanoma generally represent less than 10% of cases¹¹.

Diagnosing anorectal melanoma is challenging. Complete skin examination and review of previous skin biopsies are important¹². Physical examination must include visual inspection of the anus, digital examination, and anoscopy and/or proctoscopy (retroflexion of endoscope in the rectum looking for evidence of thrombosis or related pathology^{12,13}.

Anorectal melanoma is highly lethal. In many patients, this malignancy presents as localized, resectable disease; however, initially undetected metastases are common; thus, long-term survival is unusual. As many as a third of patients have metastatic disease at presentation and more than 80% of patients die of distant metastatic disease within 5 years¹⁵. Location also plays a role in survival. A small retrospective study using the classification described above found that two-thirds of rectal and anorectal melanomas recurred systemically whereas anal melanoma tended to recur first within sentinel lymph nodes¹⁶.

KEY POINTS

Any patient with a history of melanoma presenting with digestive symptoms, especially anorectal, should have a careful diagnostic evaluation. This malignancy can masquerade as refractory hemorrhoid disease as in our second case. Diagnosis of melanoma should be considered in hemorrhoids that enlarge; are pigmented, ulcerated or indurated; and persist despite conservative therapy. Anorectal melanoma may present (as in our third case) as

a non-pigmented mass, and be associated with small amounts of rectal bleeding, pain, itching, or incontinence. Bowel complaints of any magnitude in adults with a known melanoma should prompt consideration of metastatic GI involvement in patients with a known melanoma. Immunohistochemical staining is useful in diagnosing a questionable lesion. Despite its rarity, due to its significant mortality, anorectal melanoma should remain on the differential in unusual anorectal lesions.

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