Primary amelanotic melanoma of the rectum mimicking adenocarcinoma

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Patient: Male, 55
Final Diagnosis: Melanoma
Symptoms: Worsening constipation • tenesmus • weight loss
Medication: —
Clinical Procedure: Chemoradiation therapy
Specialty: Oncology
Objective: Challenging differential diagnosis
Background: Malignant melanoma is usually readily diagnosed by the presence of melanin granules. Although amelanotic melanoma contains a few melanin granules, it is often difficult to differentiate from non-epithelial malignant tumors. Immunohistochemical staining may be needed to diagnose the condition.
Case Report: This report describes a case of amelanotic melanoma of the rectum, which was originally suspected to be an adenocarcinoma, but was subsequently correctly diagnosed by immunohistochemical staining with HMB-45 antibody and by the presence of S-100 protein. A pinkish-red ulceroproliferative growth was located about 7 cm from the anal verge. The patient was treated by laparoscopic low anterior resection.
Conclusions: Very few cases of amelanotic melanoma of rectum have been reported in the literature and there is only limited clinical experience with this disease. It appears to be a highly lethal tumor and may need much more aggressive treatment than that used for carcinoma of the rectum.
Key words: anorectal melanoma • immunohistochemistry • low anterior resection • laparoscopy

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Background

Primary anorectal melanoma is a rare and highly lethal neoplasm with poor prognosis [1–6], which was first reported in 1857 by Moore [4]. Mucosal melanomas account for approximately 1.2% of all melanomas, and anorectal melanomas account for fewer than 25% of all mucosal melanomas. Anorectal melanomas are exceedingly rare, accounting for only 0.3% of melanomas and 0.8% of anorectal malignancies [7]. Furthermore, approximately 30% of anorectal melanomas are amelanotic and can endoscopically resemble benign polyloid lesions. Owing to its rarity and histological variability, misdiagnosis as lymphoma, carcinoma, or sarcoma is common [5–8]. Histological evaluation with immunohistochemical stains like HMB-45 (Human Melanoma Black-45), S-100 (Soluble 100%), and Melan A (melanoma-associated protein A) is often required for definitive diagnosis. Because 61% of patients with anorectal melanomas already have distant metastases at the time of diagnosis, the prognosis is very poor, with a median post-treatment survival time of 12–20 months and a 5-year survival rate of 6–22% [7,9–14]. To date, approximately 500 cases of anorectal melanoma have been reported in the literature [15], including fewer than 15 cases of amelanotic melanoma. Due to its rarity, amelanotic melanoma treatment is not standardized and it still remains a highly aggressive tumor. We present a case of amelanotic melanoma of the rectum, originally suspected to be a poorly differentiated adenocarcinoma, but subsequently correctly diagnosed by HMB-45 and S-100 protein immunohistochemistry.

Case Report

A 55-year-old man presented to the outpatient department with a 3-month history of worsening constipation, tenesmus, 6-kg weight loss, and passage of scant blood in his stool. Digital rectal examination revealed an irregular firm mass along the anterior wall of the rectum, and the patient was referred for a colonoscopy. Routine blood test reports were normal except for mild anemia and CEA of 31.15 ng/ml. Colonoscopy showed an ulceroproliferative growth involving the lower part of the rectum (Figure 1) with lower border about 7 cm from the anal verge. There was no pigmentation of the tumor. Colonoscopic biopsy indicated an undifferentiated adenocarcinoma. CT scan showed a mass involving the lower rectum with periserosal infiltration. No lymphadenopathy or liver metastasis were noted. The patient underwent laparoscopic low anterior resection and the specimen was sent for histopathological examination. Histological examination of the biopsy specimen revealed a cluster and nests of neoplastic cells having pleomorphic nuclei invading the lamina propria with evidence of lymphovascular emboli (Figure 2). Tumor margins were negative. Immunohistochemical analysis revealed that the tumor cells were positive for HMB-45 antibody and S-100 protein (Figures 3 and 4). These findings supported the diagnosis of an amelanotic melanoma. While receiving chemoradiation therapy, the patient developed metastatic inguinal lymphadenopathy. This signifies the highly aggressive nature of amelanotic melanoma.

Discussion

Melanomas are malignancies that can affect any anatomic region where melanocytes exist (eg, the epidermis, eyes, nasal cavity, oropharynx, vagina, urinary tract, rectum, and anus). The most common form of melanoma involves the epidermis and constitutes 91.2% of melanoma cases, whereas ocular and mucosal forms account for 5.3% and 1.3% of cases, respectively. The remaining 2.2% of cases are from unknown primary sites [7]. Anorectal melanoma is an exceedingly rare mucosal melanocytic malignancy, constituting only 0.3% of melanomas.
and 0.8% of anorectal malignancies [7]. The median age at diagnosis is 66 years, with a 60% female predominance [16,17].

The diagnosis of malignant melanoma is readily made if melanin pigment is present. Malignant melanoma usually presents as a black or brown lesion. It is readily diagnosed by conventional histochemical staining; however, amelanotic melanoma, which is a unique variant of malignant melanoma, can be misdiagnosed as a carcinoma or sarcoma because of the lack of pigmentation. It has been recently reported that immunohistochemical staining with HMB-45 is useful for the cytological and histological diagnosis of amelanotic melanoma. The HMB-45 antibody stains a 10-kDa cytoplasmic glycoprotein thought to be part of the premelanosome complex. HMB-45 can be important in the evaluation of undifferentiated neoplastic lesions that are suspected to be melanomas [18].

Conclusions

Amelanotic melanomas can be misdiagnosed as carcinomas or sarcomas because of the small number of melanin granules.

Immunohistochemical staining with HMB-45 and S-100 is useful for the cytological and histological diagnosis of amelanotic melanoma.

Amelanotic melanoma is a highly aggressive tumor and, since the reported cases in literature are very few, the treatment is not standardized.

In our case, despite negative tumor margins and good lymph node clearance, the patient developed recurrence in the form of inguinal lymphadenopathy.

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