Letter

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Primary rectal malignant melanoma with schistosomiasis

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SUMMARY: Primary rectal malignant melanoma with schistosomiasis is extremely rare. To date, only a few cases have been reported in the literature. Due to its high mortality rate, most patients with rectal malignant melanoma die within five years of diagnosis. However, the etiology and optimal treatment strategies remain controversial. A 79-year-old female patient presented with intermittent hematochezia for 2 months. Digital rectal examination, computed tomography (CT) scan, and colonoscopy revealed a fleshy mass measuring 3 cm in diameter in the rectum. A biopsy confirmed a preoperative diagnosis of malignant melanoma of the rectum, and a radical rectal resection was performed. Histopathological examination of the surgical specimen confirmed malignant melanoma, and numerous *Schistosoma japonicum* organisms were identified within the tumor. The patient subsequently received Dabrafenib and Trametinib therapy and remained disease-free for 5 years postoperatively, with no evidence of recurrence. This case highlights the potential treatment strategies for this rare carcinoma and underscores the need for further investigation into the relationship between schistosomiasis and melanoma.

Keywords: primary rectal malignant melanoma, schistosomiasis, *Schistosoma japonicum*, rectal cancer, rare tumor, targeted therapy

Rectal malignant melanoma is a rare and aggressive neoplasm that primarily arises in melanocyte-rich tissues such as the skin, eyes, and meninges. However, it can also occur in the gastrointestinal tract. Melanomas account for 1-3% of gastrointestinal malignancies, and the majority are metastases from other primary sites (*1*). Therefore, the diagnosis of primary malignant melanoma should be made only when no other suspicious primary lesions are found. Primary rectal malignant melanoma can be easily misdiagnosed based solely on clinical or radiological examinations (*2*). Consequently, histological examination is essential to confirm the diagnosis. Schistosomiasis is caused by parasitic flatworms called schistosomes, which typically infect the intestinal tract, urinary tract, and liver (*3*). In this case, *Schistosoma japonicum* was identified within the tumor, and several studies have suggested that intestinal schistosomiasis might contribute to the development of intestinal tumors (*4*). However, whether schistosomiasis infection could be a potential risk factor for malignant melanoma remains unclear.

A 79-year-old female patient presented with intermittent hematochezia for 2 months. She reported no nausea, vomiting, abdominal pain, diarrhea, or weight loss. Her appetite and food intake were normal. Physical examination revealed no abnormal pigmentation of the skin, oral mucosa, or ocular regions. However, digital rectal examination identified a fleshy mass approximately 4 cm above the anal verge, occupying nearly one-third of the rectal lumen. Blood was observed on the examining glove following the procedure. Abdominal computed tomography (CT) scan (Figure 1A) and colonoscopy (Figure 1B) were performed, revealing a lesion originating in the rectum and extending proximally for approximately 3 cm. A biopsy was performed, and the tumor exhibited contact-induced bleeding. Histological examination of endoscopic biopsy samples was highly suggestive of malignant melanoma of the rectum. Following multidisciplinary discussion in the gastrointestinal (GI) division, a radical anterior resection with total mesorectal excision and sigmoid-rectal anastomosis was performed. Written informed consent was obtained from the patient for publication of this case report and the accompanying images. Microscopically, the tumor was composed of epithelioid melanoma cells with prominent nucleoli (original magnification: 400×, Figure 2A). Notably, a large number of *Schistosoma japonicum* organisms were identified within the tumor (Figure 2B). Immunohistochemical analysis showed tumor cell positivity for Melan-A (original magnification:

400×, Figure 2A), S100 protein (Figure 2B), HMB-45 (Figure 2C), and vimentin. Genetic analysis revealed a V600 mutation in the BRAF gene. Consequently, postoperative treatment included Dabrafenib and Trametinib, along with praziquantel for anti-schistosomal therapy. The patient recovered well after surgery and remained alive and disease-free 5 years postoperatively, with no evidence of recurrence.

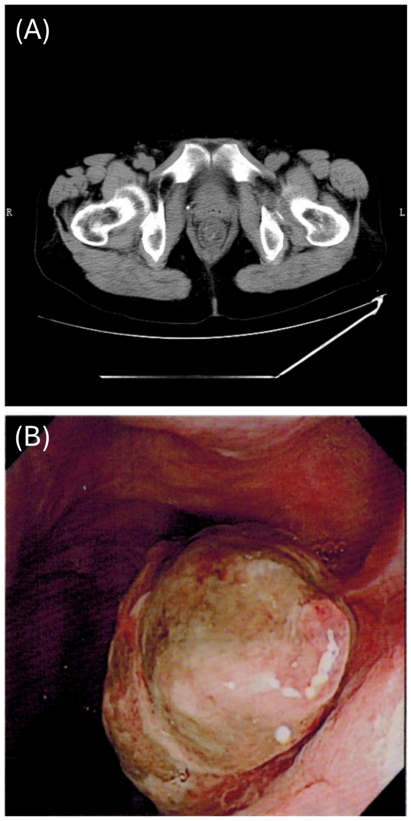

Figure 1. (A) Abdominal computed tomography scan showing a malignant rectal lesion, and (B) Colonoscopic view of the rectal tumor.

Currently, surgical resection remains the most effective treatment for this malignancy. However, due to the abundant vascular and lymphatic supply of the gastrointestinal tract, which facilitates early metastasis, the tumor is often not amenable to complete resection at the time of diagnosis, resulting in a high mortality rate from widespread metastases (*5*). The prognosis of primary malignant melanoma of the rectum remains extremely poor, with a 5-year survival rate of only 10-30% (*6*). Prognostic factors include patient age, disease stage, tumor location, lymph node involvement, depth of invasion, and the timeliness of diagnosis. Some studies have reported favorable outcomes when the depth of tumor invasion is less than 4 mm (*7*). Although melanomas are generally considered resistant to radiotherapy and conventional chemotherapy, approximately 10-20% of patients respond to certain chemotherapeutic agents, including dacarbazine, temozolomide, and cisplatin. For patients with mutations in the c-KIT gene or the BRAF V600 mutation, targeted therapies are indicated. These include c-KIT inhibitors such as imatinib, and BRAF inhibitors such as vemurafenib, dabrafenib, and trametinib (*8*).

Although the etiology of primary rectal malignant melanoma remains unclear, numerous studies have demonstrated that intestinal schistosomiasis can contribute to the development of colorectal cancer. In a study by Mingchai (*9*), 289 out of 454 colorectal cancer patients (63.7%) from schistosomiasis-endemic regions were found to have concomitant schistosomiasis.

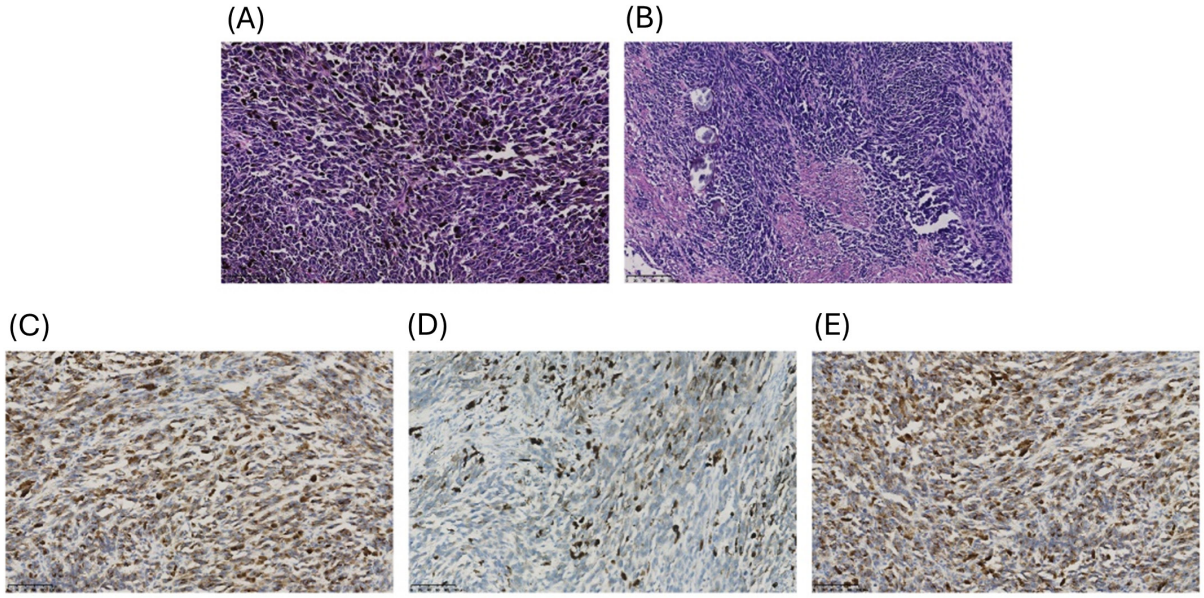

Figure 2. (A-B) H&E staining of tumor tissue at 400× magnification, highlighting epithelioid melanoma cells with prominent nucleoli (A) and the presence of *Schistosoma japonicum* within the tumor (B); (C-E) Immunohistochemical staining of tumor cells at 400× magnification, demonstrating expression of (C) Melan-A, (D) S100, and (E) HMB-45.

This suggests a potential association between schistosomiasis and colorectal carcinoma in endemic areas. Other studies have also proposed Schistosoma as a significant risk factor for colorectal cancer. Wang Z *et al*. (*4*) reported that schistosomiasis may promote colorectal tumorigenesis by influencing the polarization of tumor-associated macrophages toward the M2 phenotype, which plays a key role in tumor progression. Schistosoma eggs embedded in the submucosa can trigger chronic inflammation, polyp formation, mucosal atypical hyperplasia, and ultimately carcinoma. Interestingly, inflammatory features have been observed in melanoma tissues across all clinical stages (*10*). In this case, *Schistosoma japonicum* was identified within the rectal malignant melanoma. This raises the hypothesis that schistosomiasi may contribute to the tumor microenvironment in rectal melanoma. However, further studies are required to clarify this potential relationship.

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