Case report

# Primary amelanotic melanoma of anorectum – a rare case report with diagnostic challenge

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Anorectal melanoma is an exceptionally rare and aggressive form of cancer. One *per* cent of anorectal malignant tumours are anorectal malignant melanomas, which are exceedingly uncommon. We report a case of a 47-year-old woman who experienced painless rectal bleeding. On examination, an irregular lump was seen in the posterior rectal wall, measuring 4 × 3.7 cm. Biopsies were obtained under endoscopic guidance for histomorphology and immunohistochemistry. The biopsy examination showed nests of tumour mass in the lamina and muscularis mucosae. The tumour mass was composed of round to oval cells having enlarged nuclei, conspicuous nucleoli, and a scant amount of cytoplasm. No melanin pigmentation was noted in the tumour cells. HMB-45, S-100, and vimentin were all detected by immunohistochemistry. A definitive diagnosis of amelanotic malignant melanoma was rendered. The patient underwent abdominoperineal resection with a hysterectomy and bilateral salpingo-oophorectomy. Anorectal melanoma presents with bleeding *per rectum* and is often misdiagnosed as internal haemorrhoids or adenocarcinoma clinically. Amelanotic melanoma, which lacks melanin pigment, is difficult to diagnose. Patients who appear with rectal bleeding should have a malignant melanoma evaluation as a possible differential diagnosis, and suitable diagnostic procedures, such as a colonoscopy and a biopsy with immunohistochemistry, should be carried out to arrive at a conclusive diagnosis.

**Key words:** malignant melanoma, amelanotic melanoma, HMB45.

## Introduction

Anorectal melanoma is a malignant tumour that grows in the anal canal and arises from melanocyte cells [1]. Melanoma of the anorectum is an exceptionally rare and aggressive form of cancer. It makes up 1% of all anorectal malignant tumours, making it one of the rarest forms of the disease [2]. The peak incidence is seen among older adults, especially people in the sixth decade of life [3]. Because bleeding *per rectum* is the most frequent presenting symptom, the diagnosis is frequently mistaken for haemorrhoids. We report a case of a 47-year-old woman who experienced painless rectal bleeding.

## Case report

A woman who was 47 years old and had been experiencing rectal bleeding that was painless for the previous 6 months was taken to the hospital. There was no substantial medical history for the patient. Physical examination revealed that the patient had no abdominal distention and appeared pale. An irregular lump was seen in the *rectum* upon rectal examination. The lesion, which had dimensions 4 × 3.7 cm on the posterior rectal wall and was 7 cm away from the anal margin, was discovered during a colonoscopy. Biopsies were obtained under endoscopic guidance for histomorphology and immunohisto-

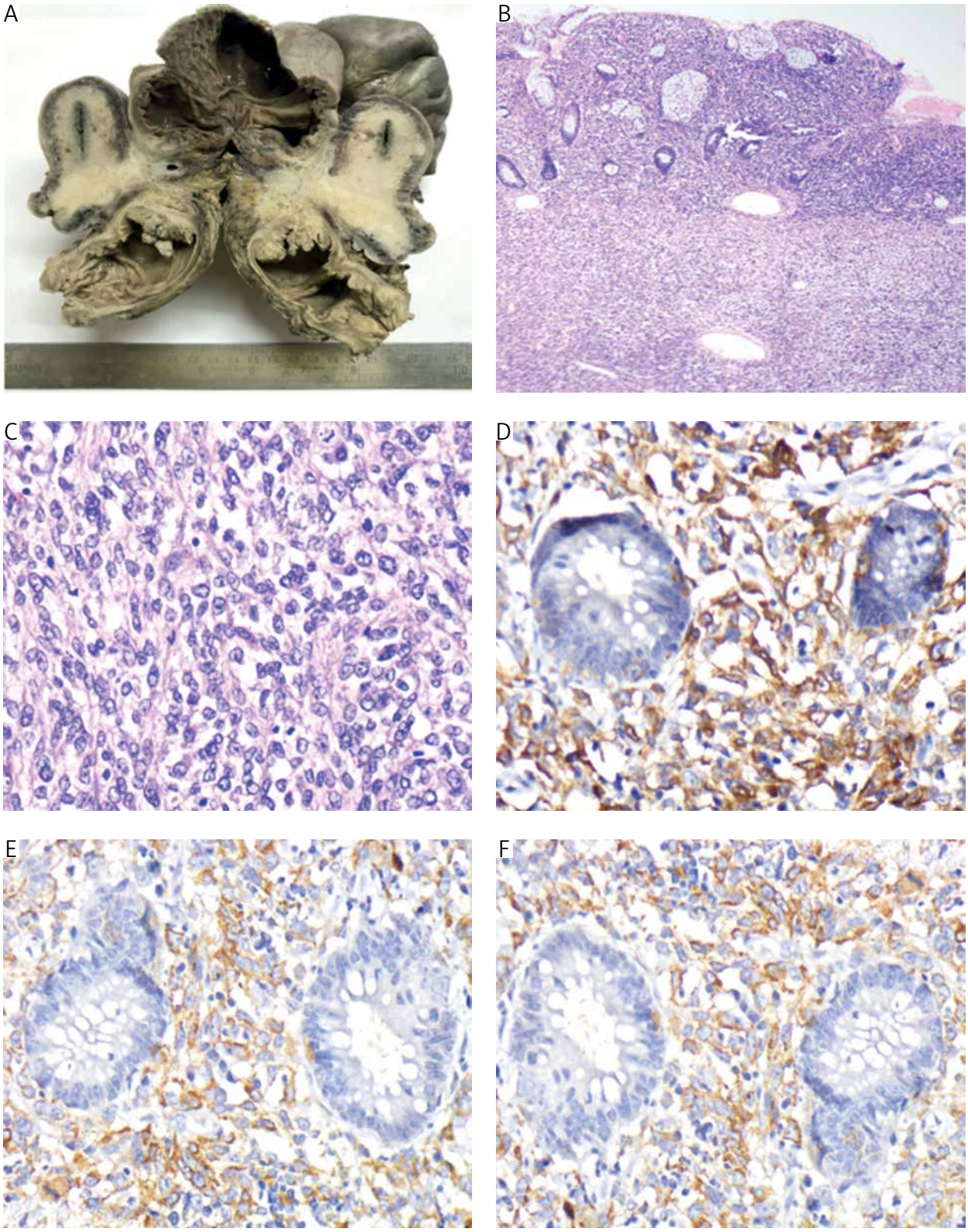


**Fig. 1.** A) Gross imaging of specimen showing a tumour in the posterior wall of the rectum adhering to the anterior uterine wall; B) histopathological examination showed nests of tumour mass in the lamina and muscularis mucosae (H&E section, 10×); C) the tumour mass is composed of round to oval cells having enlarged nuclei, conspicuous nucleoli, and scant amount of cytoplasm. No melanin pigmentation was noted in the tumour cells; H&E section, 40×; D, E, F) the tumour cells are immunoreactive for HMB-45, S-100, and vimentin, respectively

chemistry. Haemoglobin was 8.6 g/dl, total leucocyte count was $9.2 \times 10^3/\mu l$, and platelet count was $320 \times 10^3/\mu l$, according to a complete blood count. A tumour in the posterior wall of the *rectum* adhering to the anterior uterine wall was visible on a computer tomography scan. Both the ovaries showed well-circumscribed multilocular lesions, the largest of which was measured at 2.5 × 2.0 cm in the right ovary. There was no evidence of dissemination of the tumour to the lymph nodes, local dissemination, or distant metastases to liver and lung. The biopsy examination showed nests of tumour mass in the lamina and muscularis mucosae. The tumour mass was composed of round to oval cells with enlarged nuclei, conspicuous nucleoli, and a scant amount of cytoplasm. No melanin pigmentation was noted in the tumour cells (Fig. 1A, B). Based on clinical examination, radiology, and histopathology, the differential diagnosis of undifferentiated carcinoma, NHL, epithelioid GIST, and malignant melanoma was made. HMB-45, S-100, and vimentin were all detected by immunohistochemistry (Fig. 1C, D, E). The tumour cells were negative for Pan-CK, LCA, CD117, and DOG-1. A definitive diagnosis of amelanotic malignant melanoma was rendered. The patient underwent abdominoperineal resection (APR) with a hysterectomy and bilateral salpingo-oophorectomy. She was discharged on the 10$^{th}$ day after surgery with a good outcome. The sections from the APR specimen showed that tumour cells were infiltrating panmurally into the muscularis propria and perimuscular connective tissue. No invasion of circumferential resection margins or uterine wall was seen.

No mesenteric lymph nodes were positive for tumour metastasis. The sections from the uterus and bilateral ovaries showed atrophic endometrium and follicular cysts, respectively. Based on the American Joint Commission on Cancer classification, our tumour was classified as stage II with infiltration of the muscular layer. The patient was scheduled for adjuvant chemoradiotherapy. Three months after each of the patient's annual visits, there have been no recurrences since the operation.

## Discussion

Moore published the first account of anorectal melanoma in 1857. One *per* cent of anorectal malignant tumours are anorectal malignant melanomas, which are extremely uncommon but aggressive cancers that develop from melanocytes in the mucosa surrounding the anorectal junction [2]. These lesions can be polypoidal or sessile in form. While 30% of anorectal melanomas are amelanotic, about 70% of these lesions are pigmented. Immunochemistry and histology support the verification of the diagnosis. The histology markers HMB-45, S100, vimentin, vimentin, and Sox10 are frequently employed [4, 5]. In the present case, the tumour cells were positive for HMB-45, S100, and Melan-A. The differential diagnoses of malignant amelanotic melanoma includes undifferentiated carcinoma, gastrointestinal stromal tumour, and non-Hodgkin's lymphoma. Immunohistochemistry was required for a definite diagnosis [6] Bleeding from the *rectum* occurs in 53–89% of patients with anorectal melanoma. A mass in the anal region, altered bowel habit, and tenesmus are other clinical features. In our case study, the most prominent symptom was painless bleeding from the *rectum*. Malignant melanoma in the *rectum* is extremely unusual, and Kusuma *et al.* described 2 occurrences: one in a 60-year-old man and one in a 51-year-old man, both of whom were successfully treated by APR and local excision [7]. In their study, Kuka *et al.* described a case of primary rectal melanoma in a 58-year-old African woman who had lower abdomen pain with occasional rectal bleeding for 3 months before undergoing a colonoscopy, which revealed a rectal polypoid mass in the anorectal region [8].

There were 2 described staging procedures for anorectal melanoma:

1) A system based on the depth of the main tumour, the presence of lymph nodes, and distant metastasis, created by the AJCC.
2) Another disease-based staging method assigns the stages of local disease, regional lymph node disease, and metastatic illness to local disease, regional lymph node disease, and stage 3, respectively [9].

Lung and liver metastases are frequently observed. A case of a 72-year-old White male with complaints of bleeding *per rectum*, who had liver metastases from original rectal melanoma was presented by Ugonabo *et al.* [10]. In the present case, no regional lymph node or distant metastasis was seen.

Various modalities such as surgery, chemotherapy, and radiotherapy are implemented in the treatment of anorectal melanoma. Two types of surgery are offered as part of the treatment: wide local excision or radical excision as APR. Positron emission tomography/computed tomography is the most widely used method for locating perirectal lymph nodes and testing for distant metastases, to assess the patient's status for curative surgery alternatives [11]. The patient in the present case underwent APR with a hysterectomy, bilateral salpingo-oophorectomy, and adjuvant chemotherapy following surgery.

## Conclusions

Anorectal melanoma presents with bleeding *per rectum* and is often misdiagnosed as internal haemorrhoids or adenocarcinoma clinically. Patients frequently present with advanced disease. Amelanotic melanoma, which lacks melanin pigment, is difficult

to diagnose. Patients who appear with rectal bleeding should have a malignant melanoma evaluation as a possible differential diagnosis, and suitable diagnostic procedures, such as a colonoscopy and a biopsy with immunohistochemistry, should be carried out to arrive at a conclusive diagnosis.

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*The authors declare no conflict of interest.*

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