

CLINICAL CASE REPORT

## A case report: Anorectal melanoma and review literature

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### ABSTRACT

Anorectal melanoma (ARM) is an extremely rare case. It is an aggressive malignancy. ARM is difficult to diagnose because of its non-specific symptoms. The clinical sign is rectal bleeding, tenesmus, an anorectal lump and change in bowel habits and it is like another disease, for example hemorrhoid, anorectal polypoid and cancer...Diagnose this disease based on clinical, endoscopic and MRI examination and especially biopsy. We present a case of male 79-year-old in Viettiep hospital with perineal pain and rectal bleeding. He was admitted and diagnosed ARM by MRI and Endoscopy and postoperative histopathology. The patient was undergone surgery. The result of Histo-pathology is ARM. We emphasize the role of MRI in diagnosis and evaluation stage of disease that can help for treatment. The hypo or mixed intense on T2, hyperintense on T1, hyperintense in Diff and low in ADC, hyperenhancement after gadolinium injection are factors for positive and differential diagnosis between other anorectal tumors. In conclusion, ARM is a rare lesion but it can be suggested for diagnosis with MRI so we suggest that MRI should be examination for diagnostic and treatment purpose.

**Keywords:** Anorectal melanoma, abdominal–pelvic MRI

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Received: October 03, 2025

Reviewed: October 05, 2025

Accepted: December 02, 2025

### 1. Introduction

Melanoma can originate from the mucous membrane or the skin. For mucosal melanoma, which is a rare cancer with invasion and biological origin, the clinical manifestations are different from those of the skin. Asians and people with dark skin have a higher incidence [2]. To date, the mechanism of melanoma lesions in the mucosa is still unclear. Of which, malignant melanoma in the mucosa accounts for about 1/3 in the anorectum, after the head-neck and genitals in women. It accounts for less than 1% of melanomas, 0.1% of rectal cancers and about 4% of anal cancers. Clinical symptoms are not specific and therefore it is difficult to diagnose based on clinical symptoms alone. Its prognosis is also very poor, the 5-year survival rate is only less than 20% (15-17%) compared to 80% of this type of skin tumor [1], [2][7]. We would like to present a clinical case that was diagnosed and treated at Viet Tiep Friendship Hospital.

### 2. Clinical cases

Male patient 79 years old. Admitted to the hospital on May 20, 2024 with an initial diagnosis of anal fistula, perianal abscess.

*Medical history:* The patient has a history of type 2 diabetes and AFB (+) pulmonary tuberculosis, being treated for 4 months, had gallstone surgery 18 years ago. According to the

patient, the patient has had pain in the anal area for many days and has been treated at Thuy Nguyen District Hospital. The patient was admitted to the hospital because of rectal bleeding.

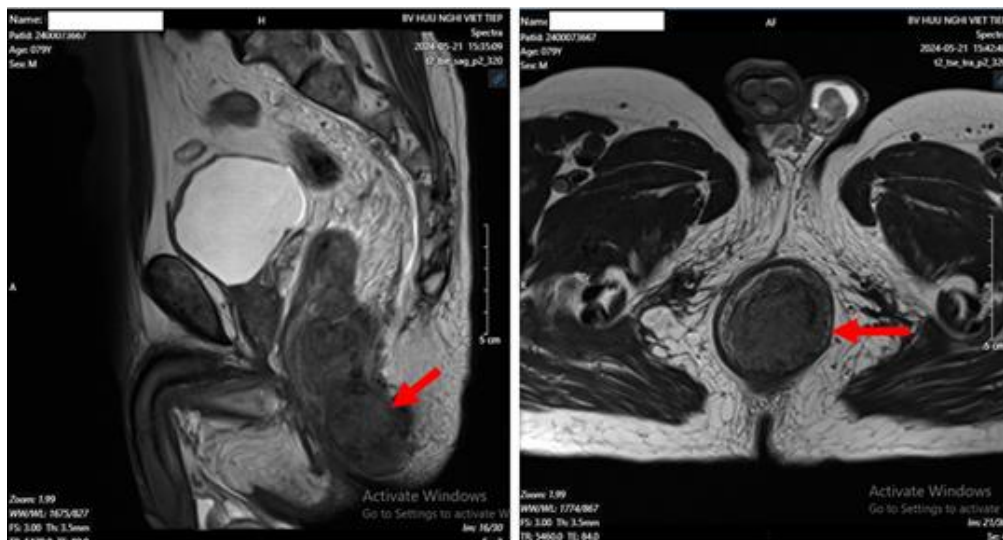
*Examination:* There is a mass about 3 o'clock from the edge of the anus with a firm density.

The patient has no signs of intestinal obstruction.

*Existing tests:* Blood count shows a slight increase in white blood cells, 10.8G/l. Other tests were unremarkable.

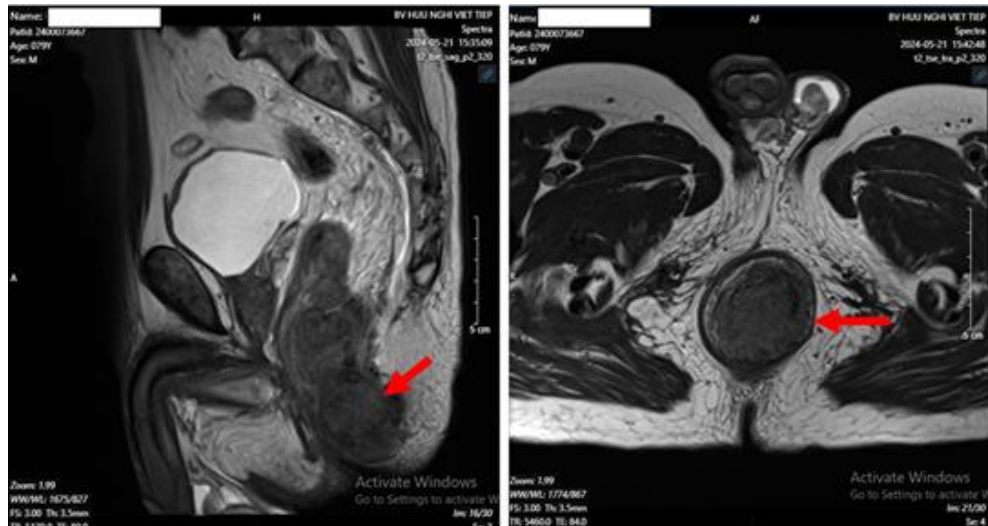
*Colonoscopy:* the result was an image of many polyps, diverticula of the ascending colon, including a polyp about 12mm, stained with epithelial proliferation. Biopsy of this polyp was performed and an image of many colon polyps was obtained. Next to the anus at the 12 o'clock position, there was a mass about 4x6cm, dark black, ulcerated and had a pseudomembrane, painful to the touch and easy to bleed. Internal hemorrhoids grade 1. Conclusion of the endoscopy: suspected hemangioma abscess/ Many colon polyps and diverticula.

MRI of the pelvis with **Siemens 3.0T SPECTRA** scanner with the pelvis protocol. The images obtained are as follows:



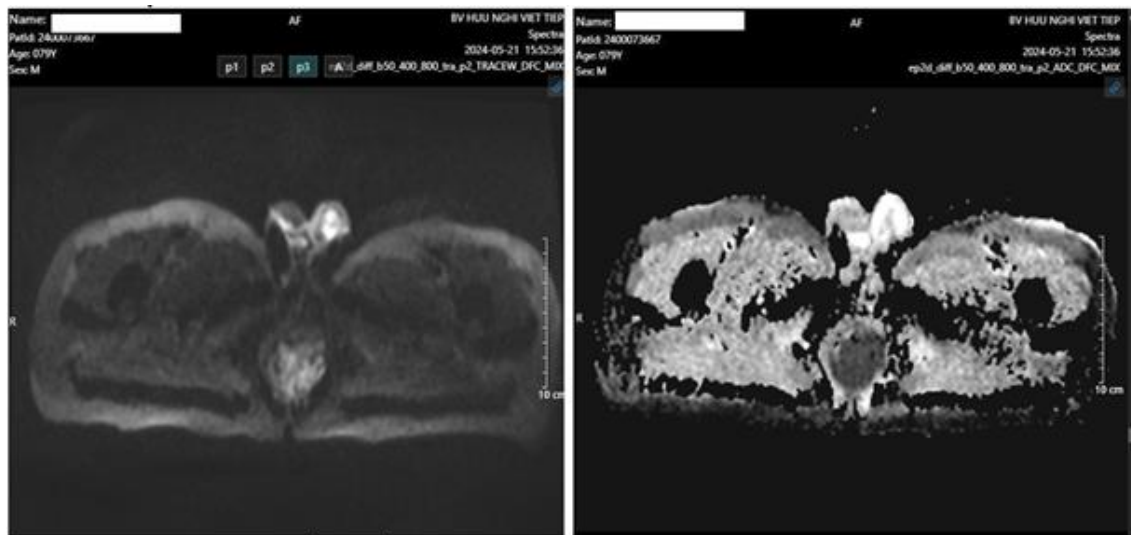
**Figure 1.** T2W (sagittal, coronal and axial) images

Image of anorectal mass occupying most of the rectal and anal lumen, hypointense on T2W (increased compared to the muscle), no disruption of the rectal wall architecture (arrow), size about 4x6cm.



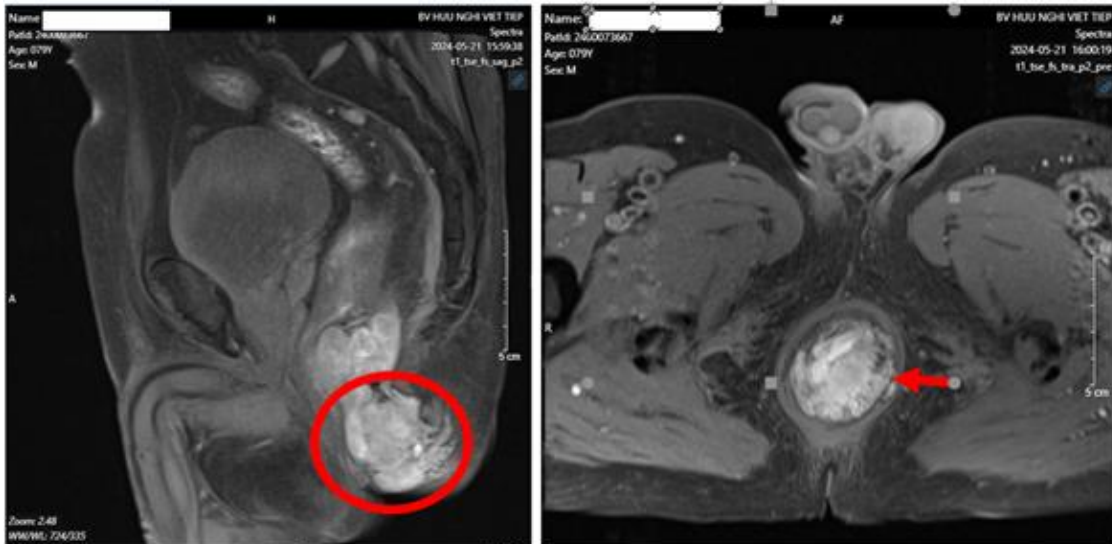
*Figure 2. T2W fat-suppressed image*

The mass in the anorectum does not invade the fat layer around the mass, and the hypointense on T2W fat-suppressed image.



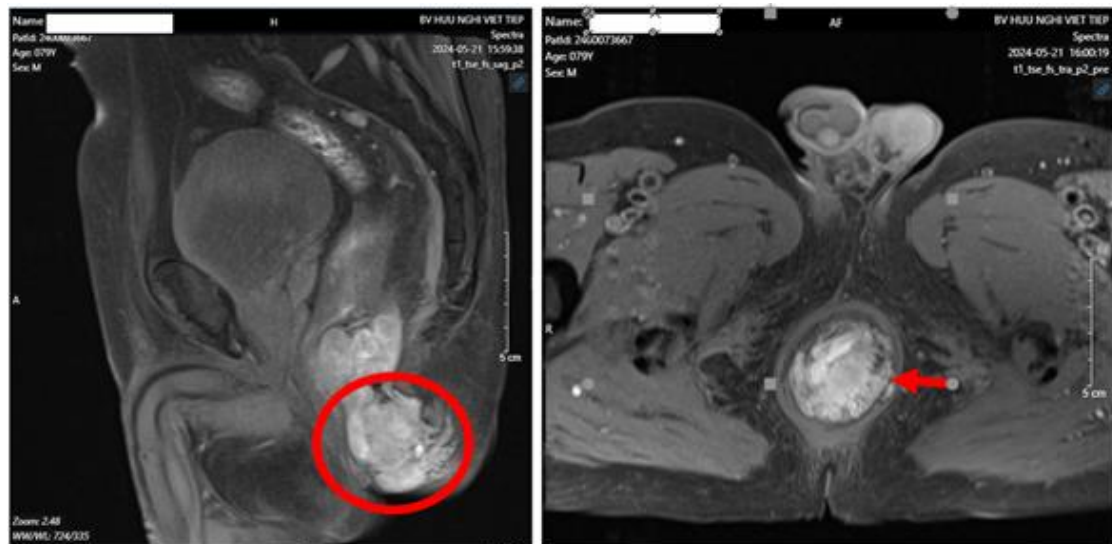
*Figure 3. Diffusion-weighted imaging (DWI) sequence*

High signal on DWI with corresponding low signal on ADC map, consistent with restricted diffusion.



*Figure 4. Fat-suppressed T1*

Mass hyperintense compared to muscle (sagittal and axial)

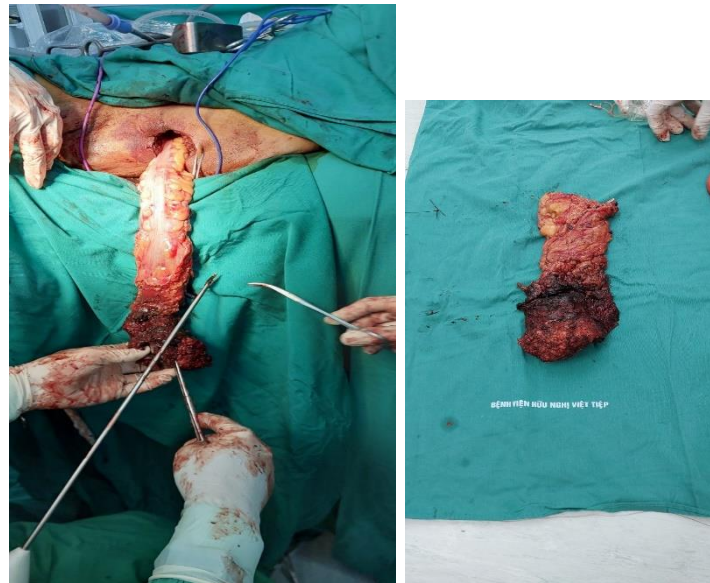


*Figure 5. Fat-suppressed T1 with CE*

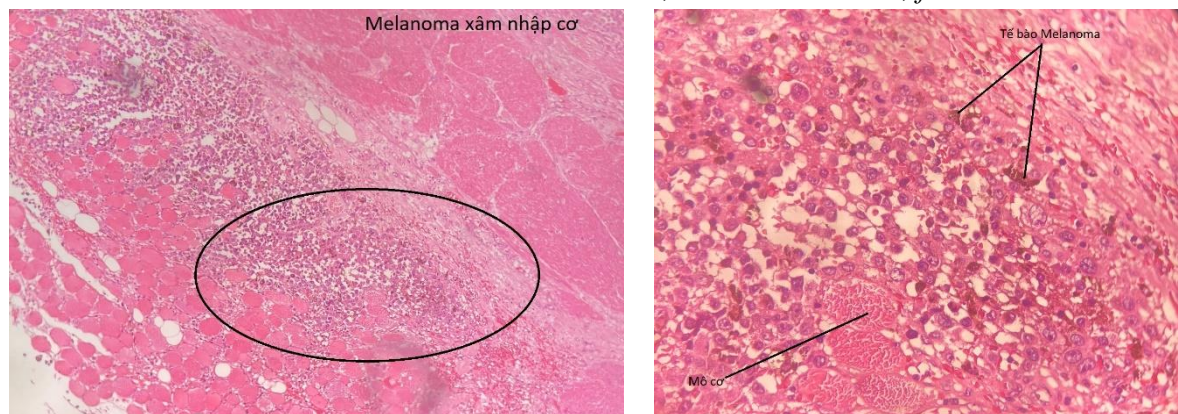
Contrast-enhanced mass (sagittal and axial)

**\*MRI results:** The mass in the anorectal region has hyperintense on T1 and T1 fat-erasing, hypointense on T2 and T2 fat-suppressed, restricted diffusion, contrast enhancement and some lymph nodes near the lesion have mass-like properties. The mass has an image that does not break the architecture of the anorectal wall, does not show surrounding fat infiltration, and does not see contraction in the rectal mesentery. The upper rectal region has an image of some small, thick-walled masses, with contrast enhancement. The lesion has exceeded the dentate line.

**\*The patient to undergo surgical treatment and post-operative antomo-pathology for the mass in the anorectum.** The surgical results showed that the mass in the anorectum was about 6cm in size, hard, purple-black, with ulcers on the surface, no infiltration to the surrounding area, and some lymph nodes near the mass. On microscopic it was a melanoma.



**Figure 6.** Surgery to remove the tumor-bearing rectal segment, gross appearance: dark brown tumor located in the rectum, about 5cm in size, firm.



**Figure 7.** Microscopic

Melanocytes are scattered and invaded into muscle layer.

### 3. Discussion

Melanocytes are cells originating from the neural crest, during the embryonic period. They tend to migrate to many locations in the body but mainly the skin. Therefore, cutaneous melanoma is the most common, accounting for more than 90% of all melanomas. In addition, melanoma can occur in the retina, uvea or mucosal surfaces... Anorectal melanoma is very rare, accounting for less than 1% of all melanomas. [1].

Mucosal melanomas are not directly related to cutaneous melanomas. Cutaneous melanomas are often associated with ultraviolet radiation while malignant mucosal melanomas, the mechanism is still unclear, are thought to be related to KIT gene mutations rather than recessive BRAF and NRAS genes [2]. Approximately 20% of anorectal melanomas are asymptomatic, so endoscopically they resemble benign rectal polyps and are therefore often misdiagnosed as hemorrhoids, polyps, or rectal adenocarcinomas.

On colonoscopy, it appears in the area below the dentate line and the transitional zone of the dentate line although it may extend throughout the anus. The mass may be small like a

polyp with or without ulceration and often causes luminal narrowing and sometimes it can be cause of intestinal obstruction. In comparison, adenocarcinoma of the rectum which usually appears as an ulcerated mass with a narrowing of the rectal lumen leading to intestinal obstruction. In addition, not all anorectal melanomas show the presence of pigment so the diagnosis can be misleading and is often confused with thrombosed hemorrhoids.

In terms of imaging, most authors believe that magnetic resonance imaging (MRI) plays a very important role in diagnosis, helping to confirm the diagnosis and especially to assess the spread and invasion of the tumor. On magnetic resonance imaging, the important sign to evaluate is the increased signal on T1W and decreased signal on T2W. However, not all melanomas contain pigment, so this feature can be seen on magnetic resonance imaging and lead to difficulty in distinguishing from other mass lesions in the anorectal region. According to Park et al <sup>[1]</sup> in a retrospective study of 12 cases confirmed to be malignant anorectal melanoma, the majority of lesions showed increased signal intensity on T1W or mixed signal intensity on T2W. All lesions had restricted diffusion on Diffusion images and contrast enhancement. Meanwhile, another study by Li et al <sup>[3]</sup> also on 12 cases diagnosed with malignant melanoma of the anorectum, it was found that 11 cases had increased signal on T1W seen in both lesions and metastatic lesions of malignant melanoma of the anorectum. Zeyan Xu et al studied in 37 patient ARM and 98 low rectal cancer and showed that hyperintensity on T1 and hypointensity on T2 can help differential between ARM and other low rectal cancer <sup>[6]</sup>. However, there is a point that distinguishing the increased signal on T1W of the tumor and bleeding is difficult, so we think that we should do more Gradient echo (T2\*) pulses to differentiate. Author Li et al <sup>[3]</sup> also recommended additional CT scans to differentiate between these two lesions and in addition, CT scans also help evaluate distant metastases, especially in late stages of the tumor, such as evaluating lesions in the liver, lungs, bones, etc.

For PET-CT, malignant melanoma lesions of the anorectum are hypermetabolic and strongly radiosensitive, so it is also suggested for staging and treatment response assessment.

However, the gold standard should be biopsy, histopathology and also immunohistochemistry because there may be non-pigmented melanomas. The morphology of the tumor cells can vary in size and shape and it is completely different from the cells of adenocarcinoma, squamous cell carcinoma.

In general, the authors believe that immunohistochemical analysis is very important and relatively specific for malignant melanoma and that molecular biological evaluation is extremely important for KIT, NRAS, BRAF gene alterations <sup>[3]</sup>. Studying this genetic variation will help in targeted therapy such as with Imatinib.

In the rectum, melanocytes are located in the transition zone, so most anorectal melanomas originate from the dentate line, then spread along the anorectal canal (65% are located in the anal canal or at the anal verge). Clinical symptoms are often nonspecific: rectal bleeding, pain, and changes in bowel habits. Itching, tenesmus, and diarrhea may occur...

In our clinical case, the imaging description is consistent with other authors' assessment of MRI morphology, which is increased signal on T1W, decreased signal on T2W, with limited diffusion and contrast enhancement after injection. In addition, the surrounding invasion was also assessed and showed that it was suitable for surgery, that is, no surrounding infiltration. This image is also consistent with other authors in the medical literature that the lesion tends to

grow toward the anorectal lumen and rarely invades the peripheral fat layer. The image is also consistent with the surgical lesion that there are some nearby lymph nodes and on pathological anatomy there are 8 lymph node-like structures and all have metastatic lesions. In addition, this tumor grows along the axis of the rectum-anus so it usually does not cause signs of intestinal obstruction, which are also points to help differentiate it from cancer of the glandular or squamous epithelium in the anorectal region.

The choice of treatment for anorectal melanoma remains controversial, with total surgical resection being the preferred treatment, WLE (wide excision) being recommended in early stage patients, and APR (complete resection) being recommended in advanced, invasive patients [4],[5].

The limitation that we noticed through this case is that it is necessary to take additional Gradient echo pulses to assess whether there is bleeding or not and also need to take CT scan to screen for lesions as well as differentiate from bleeding.

Another limitation is that immunohistochemistry and molecular biology are not performed to determine the possibility of targeted therapy and subsequent treatment after surgery.

#### 4. Conclusion

Anorectal melanoma is an extremely rare malignancy, with rapid progression and difficulty in diagnosis, which should be considered when a mass is observed on magnetic resonance imaging in the rectal segment, with increased signal on T1W, iso- or hypointense on T2W, with limited diffusion and contrast enhancement. The lesion has minimal invasion of the surrounding area. The gold standard is still histopathology.

#### 5. Recommendations

We also recommend that patients diagnosed with anorectal melanoma should undergo additional computed tomography (in addition to magnetic resonance imaging) for screening and differentiation. Patient should be examination with immunohistochemistry and molecular biology to be able to diagnose accurately and help with better treatment.

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