

**CASE REPORT**DOI: <https://doi.org/10.3329/mediscope.v9i1.58527>**Rectal melanoma in a young man with symptoms mimicking spinal cord compression****SFK Ismail¹, *J Haron², AT Musa³, SET Sharif⁴, Z Kamaludin⁵, MAMM Rasidi⁶****Abstract**

Rectal melanoma is a rare type of melanoma. Less than 3% of all forms of malignant melanomas with mucosal melanoma in the first place have been recorded. Low survival rates with poor prognosis are generally seen. Presentation of non-specific gastrointestinal symptoms results in delayed diagnosis and treatment. Rectal melanoma identified in a young man with signs that resemble spinal cord compression is not widely encountered.

Keywords: Rectal mass, melanoma, rectal melanoma.

Introduction

Rectal melanoma is rare among all types of malignant melanomas that account for about 1% of all primary melanoma. Besides being rare, this is also an aggressive form of malignancy.¹ This malignancy is more prevalent in women. The late presentation may be due to non-specific intestinal symptoms such as altered bowel habits, anal pain and rectal bleeding that mimic lower gastrointestinal tract disease. Diagnosis of rectal melanoma is achieved by tissue biopsy or postoperative histopathology.² Treatment for this

malignancy is controversial because it is strongly resistant to radiotherapy and far less to chemotherapy. Surgically, wide local excision (WLE) or abdominal resection is one of the choices.

Due to the aggressiveness of the tumour, rectal melanoma has a poor five-year survival prognosis of less than 15% following diagnosis.³ We present a case of low rectal melanoma in a young man with symptoms that resemble spinal cord compression. The tumour was detected accidentally during lumbosacral MRI imaging.

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Case history

A 39-year-old man presented to the emergency department with progressive lower limb weakness over the past two months. Despite the symptoms, he had no history of trauma. The symptoms have intensified for the last two months, causing difficulties in sitting and walking. He has had a history of rectal swelling combined with painful defecation and rectal bleeding for the past five months. He denied any family history of malignancy or skin disease.

Physical examination revealed enlarged left inguinal lymph nodes but no palpable mass in the abdomen and no abnormal skin lesions. The power of the lower limbs was 3/5 with hypotonia and areflexia. The initial assessment of orthopaedic surgeon was spinal cord compression. The lumbosacral MRI showed degenerative disease with no evidence of spinal cord compression. However, an incidental finding of eccentric thickening of the posterior lateral wall of the rectum and a large presacral mass that has a poor fat plane with S3-S5 sacrum were noted (Figure 01).

The digital rectal examination revealed multiple lobulated lower rectal mass at 12-7 o'clock position. Flexible sigmoidoscopy showed a fungating anorectal mass with contact bleeding. Lesion starting at about 2 cm from the anal verge of about 8 cm in length (Figure 02). A biopsy was taken and sent for histopathology (HPE) examination.

The gross pathology of a specimen labelled as a lower rectal tumour consisted of multiple greyish, brownish tissues with an aggregate diameter of 8 mm. Microscopic findings show a tumour fragment mixed with an adjacent normal colonic mucosa that is consistent with malignant melanoma. Tumour cells arranged in a round, oval vesicular nucleus sheets with eosinophilic macronucleoli (Figure 03A). The cytoplasm was eosinophilic and moderate to abundant, with some melanin pigments within. Tumour cells are immunopositivity to S100, HMB-45 stains (Figure 03B & C), vimentin and negative for CKAE1/AE3 and LCA.

Based on the final HPE, the patient expected to

undergo an immediate CT scan in order to stage the disease before deciding on clinical management. Unfortunately, two months after the diagnosis, he died at home before the appointment.

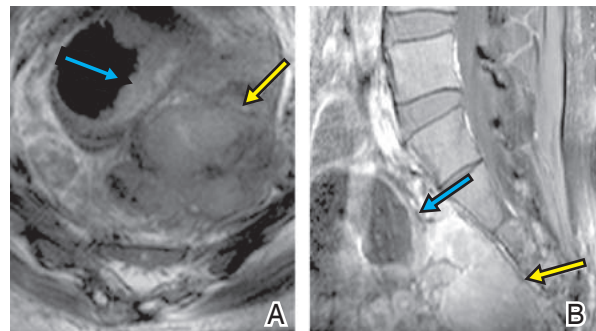


Figure 01: MRI of lumbosacral spine on axial T2W (A) and sagittal T1W (B) post-Gado shows a presacral mass (yellow arrow) and rectal wall thickening (blue arrow)

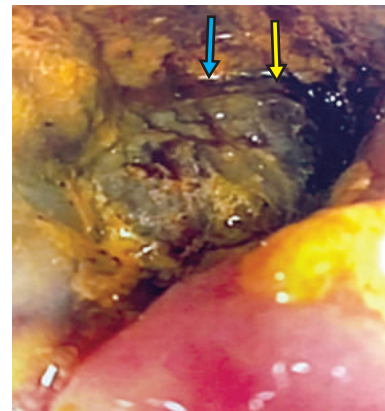


Figure 02: Image from the flexible sigmoidoscopy shows an anorectal growth (yellow arrow).

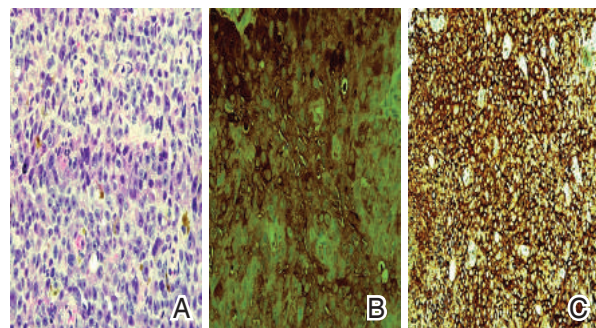


Figure 03: (A) The neoplastic cells are pleomorphic, and exhibit enlarged vesicular nuclei with macro nucleoli (H&E 400X). (B and C) The tumour cells are immunopositivity to S100 and HMB-45 stains 400X.

Discussion

Melanoma is categorised as both cutaneous and non-cutaneous. The type of non-cutaneous melanoma includes mucosal and ocular melanoma. The mucosal type is the lowest number of patients per year among all types of melanomas. Varieties of mucosal origin include the head, neck, female genitalia, anal, rectal and urinary tract.⁴

Anorectal melanoma is a rare occurrence of malignant melanoma. Of all melanomas, anorectal melanoma is only 0.4-1.6%, and anorectal malignancy is 1.0%. It is predominantly female and is over 50 years of age. Our patient is a male who presented to us at a relatively young age. There are various theories about anorectal melanoma pathogenesis. One pathogenesis is the oxidative stress of the immunosuppressive state.²

Melanocytes in the gastrointestinal tract induced production of mucin in the Goblet cells and decreased development of defensin in the Paneth cells.⁵ Rectal malignant melanoma derived from melanocyte cells originating from neuro-ectodermal multipotent neural crest cells.² It is often seen in the anorectal region at the junction of the squamous zone or transitional zone.⁵

Most patients with rectal melanoma had typical abdominal or lower gastrointestinal signs and symptoms such as abdominal pain, rectal bleeding, rectal tenesmus and later obstruction.² However, our patient had a lower limb weakness that mimics the compression of the spinal cord. His symptoms of rectal bleeding were suggestive of haemorrhoids, adenocarcinoma polyps and rectal cancer. This non-specific clinical presentation is a diagnostic challenge. There is often a delay in the management of the patient.⁵

HPE is the gold standard test for diagnosis. The tissue sample can be taken by sigmoid colonoscopy as the location is between the anus and the rectum. For histopathology staining, tumour cells are immunopositivity to S100,

HMB-45 stains, vimentin and negative to CKAE1/AE3 and LCA.⁶

Radiology imaging offers a non-invasive diagnostic method. Patient choices were endoscopic endorectal ultrasound, contrast-enhanced computed tomography (CECT) and magnetic resonance imaging (MRI). The thickness of the tumour and the surrounding nodule was assessed by endoscopic endorectal ultrasound. Meanwhile, the characteristics and extension of the tumour were evaluated by CECT and MRI. In CECT of the abdominal and pelvic, rectal melanoma was seen as an intraluminal rectal mass. Sometimes enlarged paraaortic, pelvic and inguinal lymph nodes are seen which are suggestive of lymphatic spread. Metastases to other organs such as liver and lung can be seen on CECT study.⁷

Delineation of the tumour margin and extension is best seen in the MRI study before surgical intervention. On the MRI, the tumour has a high signal intensity in the T1 weighted due to paramagnetic feature and a mixed signal in the T2 weighted that is unique to the rectal melanoma. Other rectal masses will have a low signal intensity in T1 weighted sequence.⁸

It is controversial to treat patients with radiotherapy and chemotherapy because they are less responsive and therefore, surgery is the best management option for rectal melanoma. Abdomino-peritoneal resection is the most favourable type of operation. However, WLE is the alternative to less extensive disease. Although radiotherapy and chemotherapy are less responsive, some patients underwent these post-surgical procedures to control tumour extension.⁹

Besides being a rare entity, rectal melanoma is regarded as an aggressive disease with poor prognosis, with just 3-22% of patients having five years of survival - less than ten months of survival seen in patients with metastatic or recurrent disease.¹⁰ In our situation, the patient died two months after the diagnosis.

Conclusion

Anorectal melanoma is a rare, aggressive type of malignancy. Most patients have a poor prognosis, and delayed diagnosis contributes to a more unfortunate outcome. Treatment for this type of melanoma is challenging due to the lack of response to radiotherapy and chemotherapy. A surgical approach is not curative.

Conflict of interest: There is no potential conflict of interest relevant to this article.

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