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Anorectal Melanoma: A Case Report

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ABSTRACT

Anorectal melanoma is a rare and aggressive form of mucosal melanoma that is difficult to diagnose early due to nonspecific symptoms like rectal/anal bleeding, anal pain, or the presence of an anal mass. Its prognosis is generally poor, with most cases already metastasized at diagnosis. A case report is presented of a patient initially presenting with anaemia, painful defecation and rectal bleeding, later diagnosed with stage IV anorectal melanoma. There is limited literature on this disease, leading to a lack of comprehensive understanding. Surgical resection is the main treatment, but over 80% of patients die within five years due to distant metastasis. The 5-year survival rate is estimated to be between 6% and 22%.

Keywords: Anorectal, Melanoma, mucosa

INTRODUCTION

Skin cancers, including melanoma, is the most common type of cancer, with invasive melanoma accounting for about 1% of all cancers. Melanomas can occur on the skin (cutaneous melanoma) or in mucous membranes, such as the sinuses, nasal passages, oral cavity, vagina, anus and rectum. Very few primary malignant mucosal melanoma cases are reported within the oesophagus and anus, but the rectum is the rarest area of involvement [1,2] Mucosal melanoma make up about 1.4% of all melanomas with half originating in the head and neck region. Anorectal melanoma, accounting 0.4% to 1.6% of all malignant melanomas and 4% of anal malignancies, is increasing in the incidence [3.4]

The incidence of melanoma has been rising, with rates varying among different demographics. In 2023, American Cancer Society estimated that there would be approximately 186,680 new cases of melanoma diagnosed in the United States, with 7,990 deaths [5].

Melanoma develops when melanocytes (pigment-producing cells) undergo mutations, often due to UV light exposure primarily from sunlight, according to the American Cancer Society [6]. Other risk factors include family history, atypical moles, fair skin, freckling, light hair, being male, and immunocompromised states.

Treatment for melanoma depends on the stage of the disease, with surgery being the preferred first-line therapy. Other options include immunotherapy, targeted therapy, radiation therapy, and chemotherapy, with treatment evolving rapidly due to improved understanding of the disease.

This case report is meant to further bring into light a highly morbid type of melanoma in the hopes of promoting earlier detection and consequently better prognosis.

CASE PRESENTATION

The patient is a 70-year-old male with a medical history of hypertension, Diabetes mellitus type II. He initially presented with anaemia, painful defecation and blood per

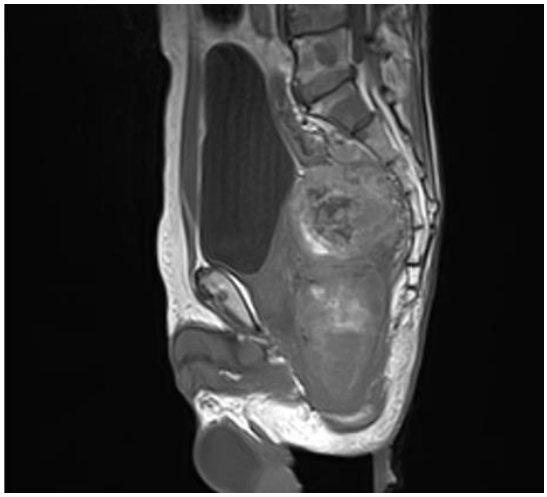
rectum while passing stools for the last 2 months. During workup, his haemoglobin was found to be 11gm/dL. On physical examination, a PR examination revealed a nodular growth in the anal canal and a large soft, polypoidal ulcerated lesion was visible on proctoscopy. There is no history of inflammatory bowel disease or any prior anorectal procedures.

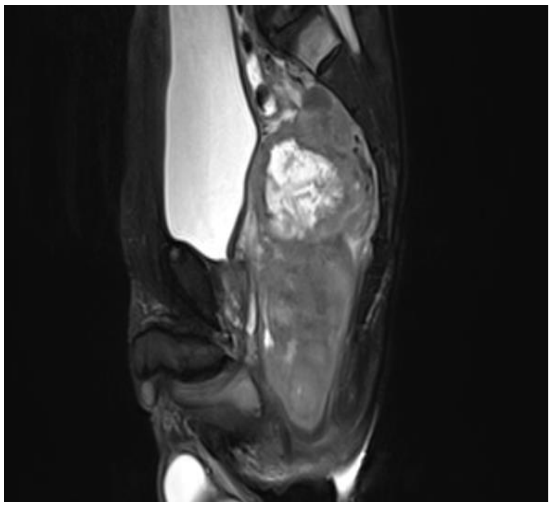
The patient was advised for an MRI pelvis, which revealed an ill-defined, lobulated eccentric mass lesion with an associated exophytic component extending from the anal canal involving the anorectal junction, rectum, extending upto the distal sigmoid colon. It measured approximately 8 cm x 8.5 cm x 15.5 cm (AP X TR X CC). The lesion was seen breaching the serosa of the left lateral wall of the rectum and extending into the left pararectal space. The lesion was heterogeneously iso to hyperintense on T1WI and T2WI with multiple foci of T1 shortening within. The lesion showed foci of hyperintensity on DWI which showed significant restriction of diffusion on ADC maps. Anteriorly the lesion was noted displacing the bladder anteriorly with loss of fat planes at places. Posteriorly the lesion was noted reaching the sacral vertebrae without any direct invasion of the vertebrae. Laterally the lesion was noted reaching upto the left obturator internus with loss of fat planes. Inferiorly the lesion was infiltrating the prostate. On post contrast images the lesion showed moderate heterogenous enhancement with few non- enhancing areas within-likely necrotic areas. A few heterogeneously enhancing sub centimetric sized lymph nodes were noted in the bilateral inguinal regions. Multiple areas of altered signal intensity appearing hyperintense on both T1W and T2W images with heterogenous post contrast enhancement were noted in the visualised axial and appendicular skeleton, likely bony metastatic lesions.

Screening of the whole spine was done which showed similar lesions in multiple cervical, dorsal and lumbar vertebrae with involvement of the posterior elements. Screening of the abdomen revealed multiple T2W hyperintense lesions in both the lobes of the liver, largest of them measuring around 17 mm in its largest diameter in segment VII of the liver. A large paraaortic lymph node measuring around 25 mm (MSAD) was noted. These liver lesions and the retroperitoneal lymph node showed restriction of diffusion. Thoracic screening revealed a pleural based lesion involving the right sided ribs with a small collection along the lateral chest wall in the visualised FOV of the chest.

The patient underwent trans-anal biopsy of the anal canal lesion. The patient was placed in a prone position. The perianal area was infiltrated with 20 cc of lignocaine for local anaesthesia. The lesion in the anal canal was identified, and a full-thickness biopsy was performed., The patient remained stable throughout the procedure and the procedure was tolerated well by the patient.

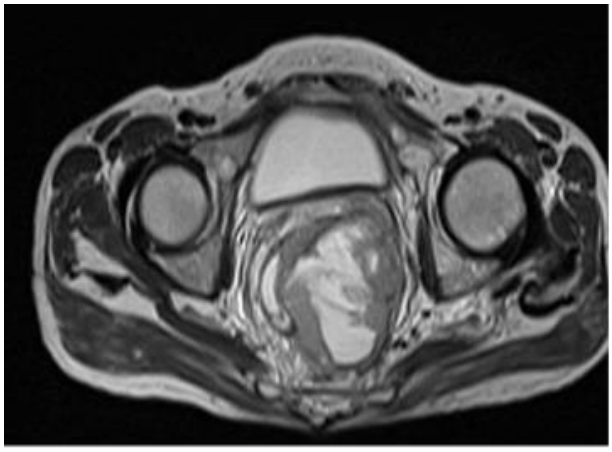
Pathology results revealed invasive malignant melanoma of the anal canal. The tumour exhibited ulceration and lympho vascular invasion, and the distal margin was positive. It involved the anal mucosa, extended into the columnar colonic mucosa, and had a high mitotic rate of 15/10 high-power fields. Immunohistochemical markers HMB45, S100 positivity supported the diagnosis. At this stage the patient was classified as Stage IV disease and further workup with a melanoma panel was sent.



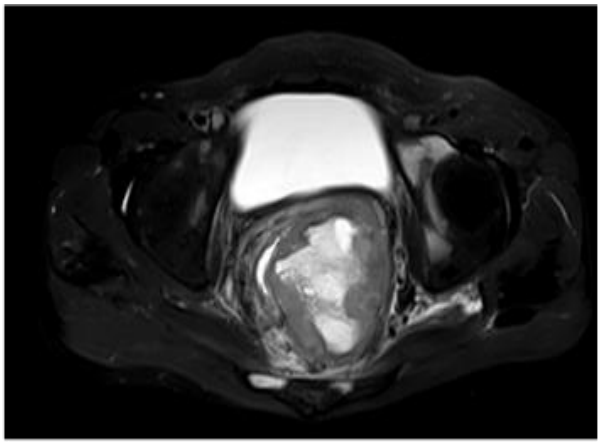


Sagittal plane images T1W sequence

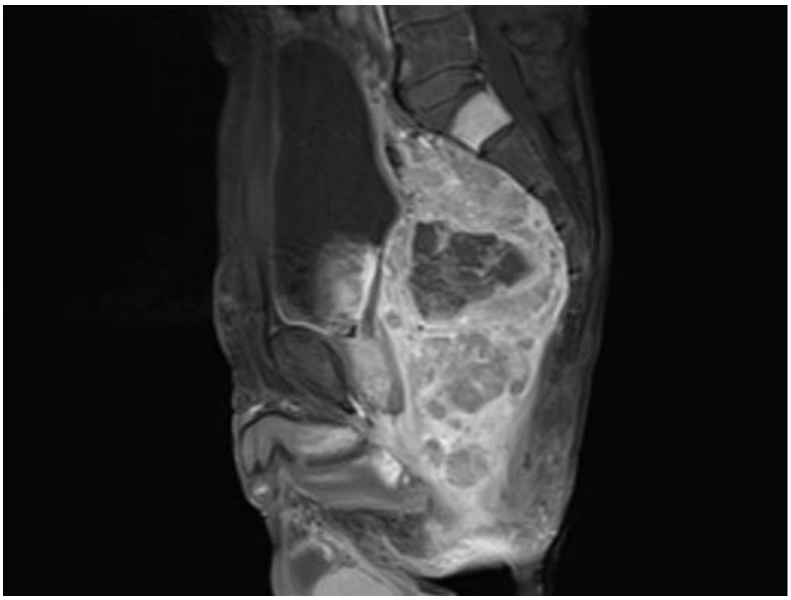
Sagittal plane images T2W sequence



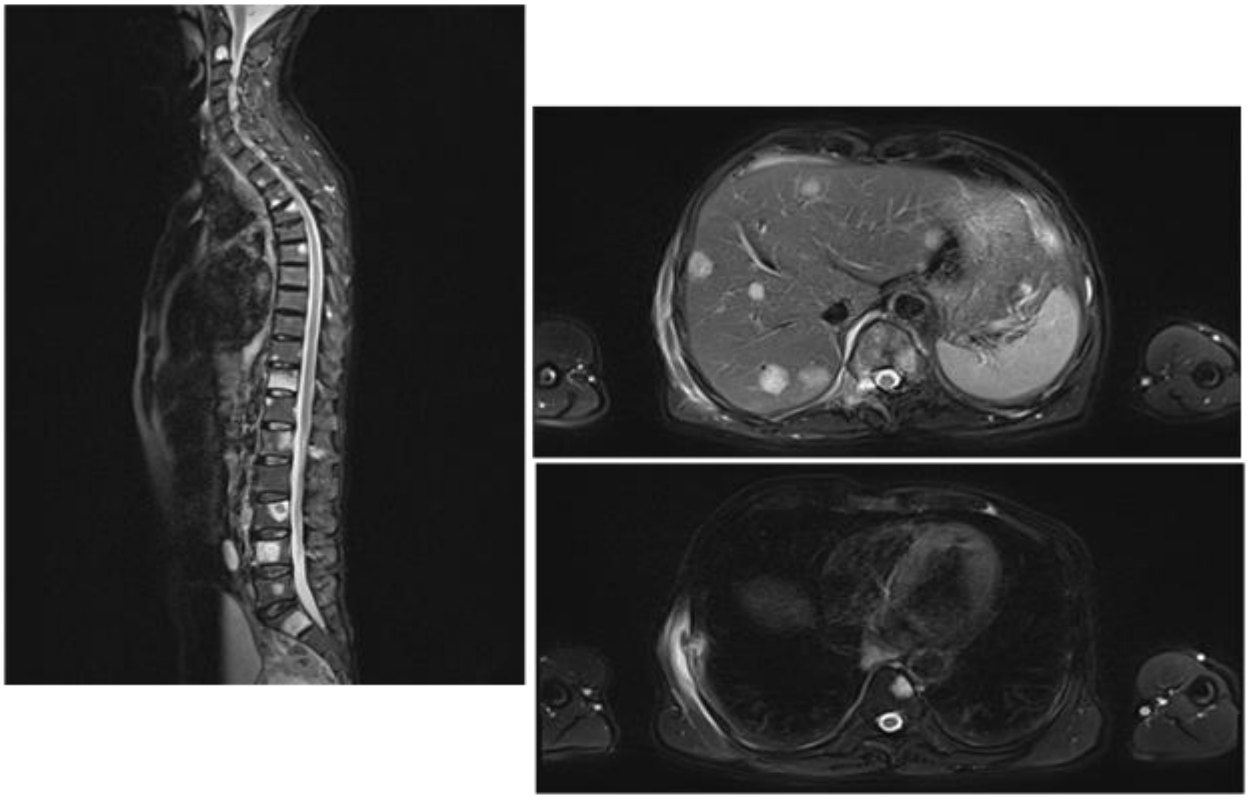
Axial Plane image T1W sequence



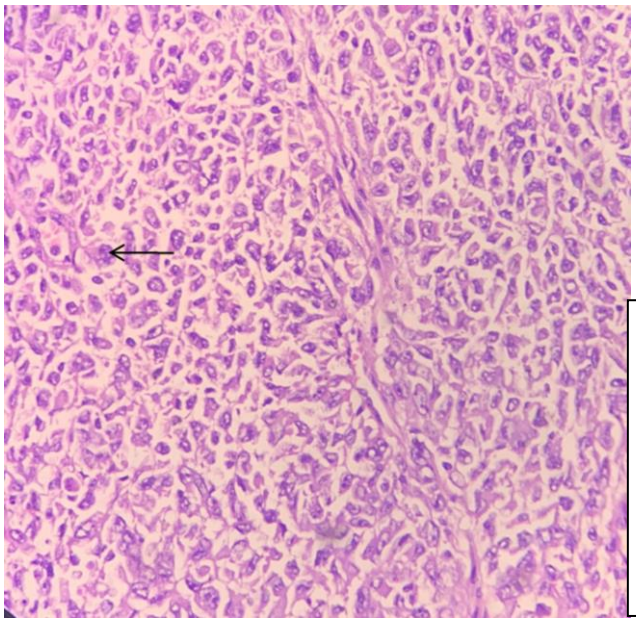
Axial Plane image T2 FS sequence



Sagittal Plane image T1 FS post contrast sequence



Screening of whole spine, abdomen, chest showing multiple bony metastatic lesions, multiple liver metastase and right pleural based lesion with small chest wall collection on STIRT2W images.



A histopathology section of the tissue showing the atypical cells arranged in 'sheets'. These cells show a high nuceo-cytoplasmic ratio, high degree of pleomorphism and prominent nucleoli(arrow) along with moderate amount of cytoplasm.

DISCUSSION

Anorectal melanoma is often diagnosed late due to its nonspecific symptoms, with 41% already regionally spread and 22% metastasized at diagnosis [7]. The lungs, liver, brain, and bone are the typical sites of metastasis [8]. Symptoms include painless rectal bleeding, which can be mistaken for other conditions like hemorrhoids. Melanomas are typically pigmented, but if a dark-colored mass is observed at the anal verge, it can be mistaken for a thrombosed

hemorrhoid. Additionally, anorectal melanomas can present as amelanotic melanomas, lacking pigmentation and requiring histopathological evaluation for diagnosis [9].

Melanoma is a type of cancer originating from melanocytes, the skin's pigment-producing cells. These cells, located in the basal layer of the epidermis, produce melanin, which gives skin its color and provides protection against UV radiation. In mucosal membranes, melanocytes contribute to antimicrobial defense and immune responses [10]. The pathogenesis of anorectal melanoma is not fully understood, with theories suggesting a link to oxidative stress, immunosuppression, derivation from Schwannian neuroblastic cells, or cells of the amine-precursor uptake and decarboxylation system of the gut [10,11]. The KIT receptor tyrosine kinase has been implicated in anorectal melanoma development, with mutations in genes like KIT, BRAF, and NRAS associated with melanoma development overall [3].

Due to the rarity of anorectal melanoma, there is a lack of randomized trials and standardized treatment plans. However, surgical interventions are commonly employed [10]. Wide Local Excision (WLE) or Abdominoperineal Resection (APR) are the preferred surgical approaches, with similar outcomes. Considering the morbidity associated with APR, WLE is generally recommended [12]. Intermediate thickness mucosal melanoma may require sentinel lymph node biopsy [13].

Various adjuvant therapies have been used in treating anorectal melanoma. Alfa-interferon immunotherapy has been effective in improving survival and reducing recurrence rates in patients with positive lymph nodes. Other treatments, such as 117-Caesium brachytherapy and chemotherapy regimens containing dacarbazine, vincristine, and Nimustine hydrochloride, have shown varied success rates [14]. Treatment decisions should take into account the patient's quality of life and comorbidities, especially for metastatic cases. Immunotherapy, particularly agents like Ipilimumab, Nivolumab, and Pembrolizumab, which target T-cell-mediated antitumor immune responses, has shown promising outcomes [10,15]. Imatinib mesylate has shown encouraging results in patients with KIT-mutated rectal melanoma, and combination therapies like Nivolumab and Ipilimumab have been effective in advanced melanoma [16,17]. Sunitinib has led to complete remission in some patients with KIT-mutated melanoma. While radiation therapy has not been extensively studied, it has been used for hemostasis in certain cases. Early diagnosis and staging are crucial factors influencing the prognosis of anorectal melanoma.

Anorectal Melanoma (AMM) typically has a poor prognosis, with an estimated 5-year survival rate of around 20% regardless of the treatment used. The main goal of surgery is to enhance the patient's quality of life, as many AMM patients already have widespread metastasis when diagnosed. Perineural invasion, as detected on histopathology, is the most critical factor influencing prognosis and the chance of recurrence.

CONCLUSION

Anorectal malignant melanoma is a rare and highly aggressive disease, often challenging to detect early due to its nonspecific initial symptoms. Surgical resection is currently the preferred treatment, but comprehensive research on the disease is lacking. Our objective is to contribute to the existing case studies, raise awareness about its high morbidity and mortality rates, and help develop management guidelines.

Diagnosing skin cancer, including anorectal melanoma, heavily relies on patient detection, which can lead to missed lesions, particularly with the obscurity of anorectal melanoma. To address this, implementing a full-body skin surveys as part of annual physical examinations. Since skin cancers make up about one-third of reported cancers worldwide, regular skin exams can aid in early detection. Additionally, we

recommend annual anoscopy with special attention to the dentate line due to the aggressive nature of anorectal melanomas.

Declaration by Authors

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Conflict of Interest: The authors declare no conflict of interest.

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