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Case Report

Surgery Section

Anal Nodular Melanoma Masquerading as Thrombosed Haemorrhoid: A Case Report

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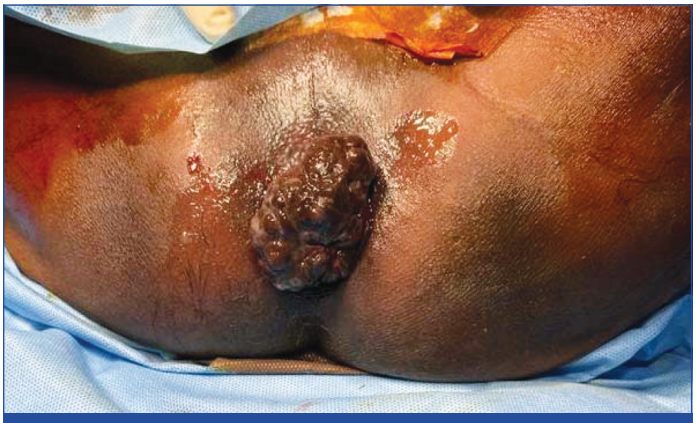
ABSTRACT

Anorectal Melanoma (AM) is an aggressive and rare malignancy with a poor prognosis. It is more commonly observed in women over the age of 50 years. Due to its rarity, AM is often undetected at the time of diagnosis or has already metastasised. This case describes a 67-year-old male who presented with a mass descending per annum for two years. This was associated with pain and blood in the stools for two weeks. History of constipation present. No known comorbidities and no past surgical history. The mass was lobular and soft in consistency. It was suspected to be thrombosed haemorrhoids, and he was taken up for surgery. The mass was excised in toto, and on histopathological examination, it was diagnosed to be invasive melanoma- Nodular melanoma of the anal canal with no evidence of lymph vascular emboli in the section. Immunohistochemistry (IHC) markers showed pan-Cytokeratin (CK)-negative, S-100-positive, HMB-45-positive and MELAN A-positive. No signs of recurrence were noted for six months, and the patient is currently on follow-up. Anal melanomas are often mistaken for a thrombosed pile mass, and improper surgery might result in recurrence. Careful planning and evaluation before proceeding with excision is advised.

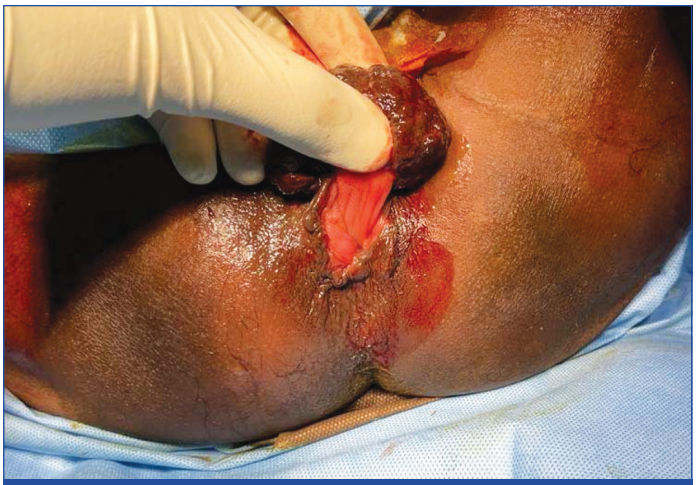
Keywords: Anorectal mass, Mucosal melanomas, Non-cutaneous melanoma, Pan-cytokeratin, Thrombosed mass

CASE REPORT

A 67-year-old male presented with a mass descending per annum for two years. He complained of pain while passing stools for the past two weeks and reported intermittent incidents of blood in stools over the last two years. He had a history of constipation but denied any history of fever, melena, weight loss, or loss of appetite. The patient had no known history of Type 2 Diabetes Mellitus, systemic hypertension, bronchial asthma, or tuberculosis. There was also no family history of malignancies or prior surgical interventions. On examination, the patient was conscious and fully oriented to time, place, and person. No signs of pallor, icterus, cyanosis, or lymphadenopathy were observed. Local examination of the anal region revealed a lobular swelling measuring 6×4 cm, arising from the 7 o'clock region. His sphincter tone was increased. The mass did not bleed on touch, and there was no warmth, tenderness, or discharge [Table/Fig-1]. On systemic examination, the abdomen was soft, non-distended, with no warmth or tenderness. Bowel sounds were present. Hernial orifices were free, and the external genitalia appeared normal. A differential diagnosis of a perianal mass was considered, with the possibility of a thrombosed pile mass. The patient was planned for surgical intervention. Under spinal anaesthesia, the mass was excised in toto and sent for biopsy [Table/Fig-2]. The postoperative period was uneventful.



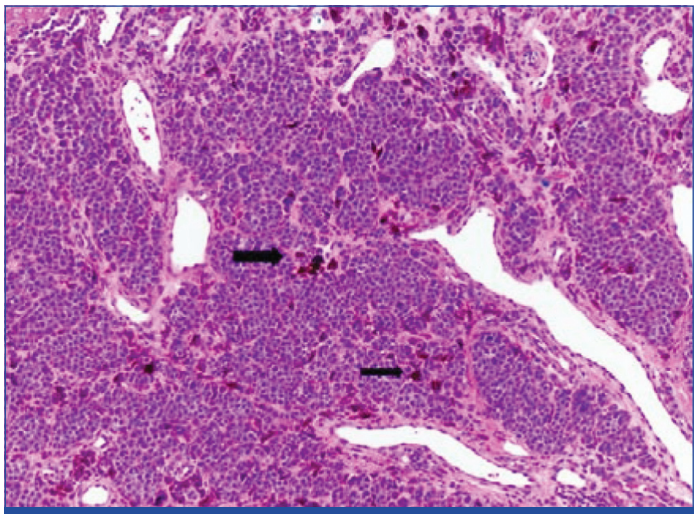
[Table/Fig-1]: Lobular swelling present in the anal region.



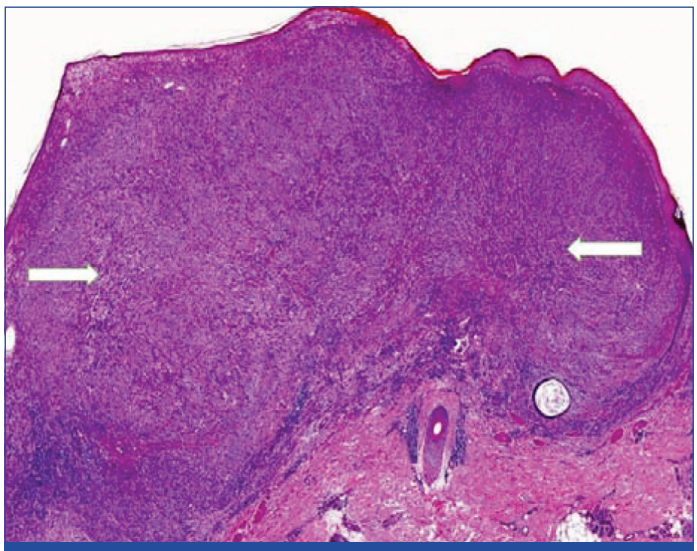
[Table/Fig-2]: Shows intraoperative image of mass.

Macroscopic examination revealed an irregular nodular mass measuring 5×2.5×1.5 cm. The skin surface of all specimens appeared grey-brown and congested, while the underlying surface ranged from grey-white to grey-brown. The cut surface was similarly grey-white to grey-brown. Microscopy demonstrated a polypoidal lesion lined focally by anal mucosa, with an adjacent infiltrating malignant tumour composed of sheets and anastomosing nests of large polygonal cells [Table/Fig-3]. These cells had round, moderately pleomorphic nuclei, prominent eosinophilic nucleoli, and extensive cytoplasmic coarsely granular melanin pigment [Table/Fig-4]. The surfaces were covered with necrotic and suppurative material throughout. Mitotic activity was recorded at 16-18 per High-Power Field (HPF). Areas of tumour necrosis, aggregates of melanin pigment, and regions of fibrosis were observed elsewhere. The tumour did not involve the anal mucosa and was located 0.3 cm away from it. No lymph vascular emboli were identified. However, the tumour was found to be infiltrating the muscularis propria. According to the TNM Stage of Mucosal Melanoma of Anorectum, this case belongs to stage I disease (localised disease) [1]. The results indicated invasive melanoma, specifically nodular melanoma of the anal canal. Immunohistochemistry markers Pan-Cytokeratin (Pan CK) were

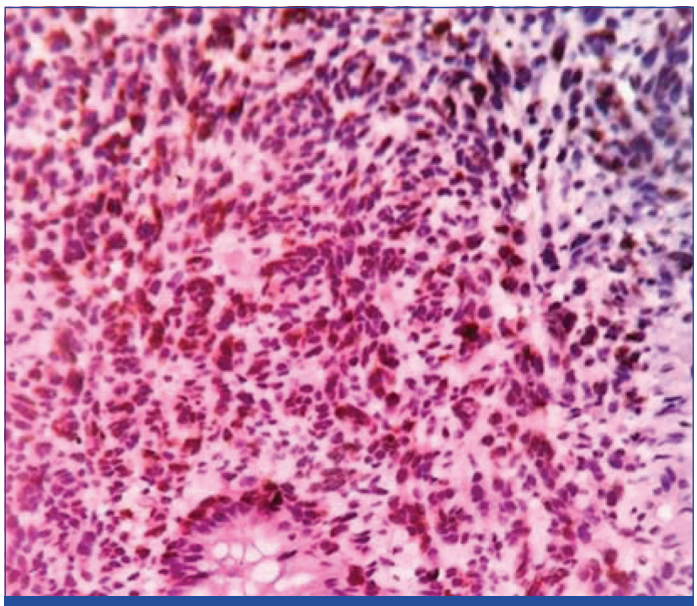
negative, while S-100 was positive, HMB-45 was positive, as shown in [Table/Fig-5]. MELAN A (MART-1)-positive was noted in tumour cells [Table/Fig-6]. The medical oncologist's opinion was obtained, and the patient was advised to maintain regular follow-ups every six months. The patient has been on consistent follow-up for a year and a half, with no evidence of recurrence noted during this period.



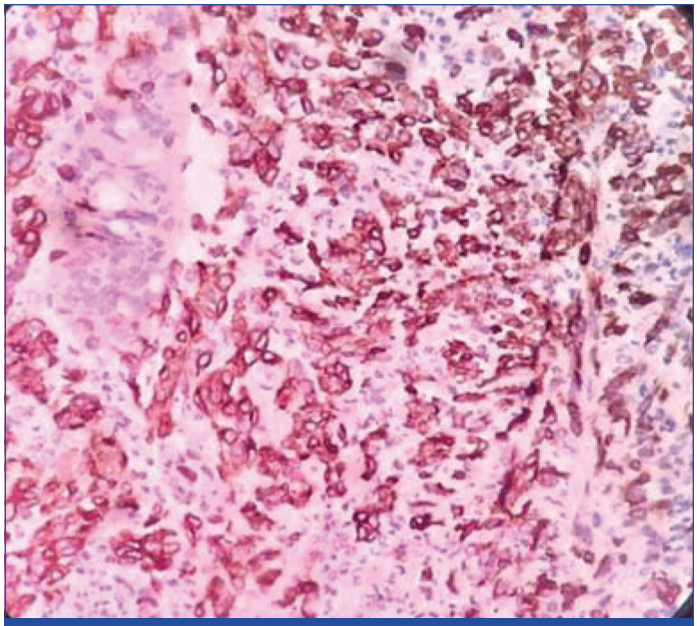
[Table/Fig-3]: Malignant tumour cells arranged in sheets and nests with aggregates of melanin pigment. The black arrow indicates nodular proliferation of atypical melanocytes (H&E, 40x magnification).



[Table/Fig-4]: Nodular dermal proliferation of atypical melanocytes. The white arrow indicates melanin pigment (H&E, 10x magnification).



[Table/Fig-5]: IHC, HMB-45 positive.



[Table/Fig-6]: IHC, MELAN A (MART-1) positive in tumour cells.

DISCUSSION

The AM is a rare and often challenging malignancy to diagnose due to its concealed location. The diagnosis is frequently made too late because of its rarity and vague symptoms, which adds to the poor prognosis. AM is frequently misidentified as colorectal cancer, haemorrhoids, or polyps [2]. The gold standard for treating AM is still surgical resection. Both Abdominal Perineal Resection (APR) and Wide Local Excision (WLE) are viable choices; however, the optimal method is still disputable [2]. This malignancy exhibits highly aggressive behaviour, often resulting in a poor prognosis. The delayed diagnosis significantly contributes to this outcome, as symptoms may be subtle or mistaken for benign anorectal conditions [3]. Anorectal Mucosal Melanoma (AMM) accounts for approximately 0.4-1.6% of all malignant melanomas and 4% of anal malignancies. The incidence of melanoma has been steadily increasing over the past few decades [4]. Gastrointestinal melanomas most commonly occur in the anorectal region, with fewer cases in the stomach, small intestine, and colon. Rectal melanoma is more prevalent than anal melanoma. Mucosal melanomas are rarer in individuals with darker skin, possibly because melanin acts as an antioxidant rather than just a UV shield [5]. The melanocytes in this region originate from the neural crest or mucocutaneous junctions, migrating to the skin. In mucosal tissues, they contribute to immune responses via antioxidant properties. Malignant changes may be linked to immunosuppression or oxidative stress [5]. AM can also arise from Schwannian neuroblastic cells within intestinal autonomic innervation or the amine precursor uptake and decarboxylation system cells [5]. Due to its hidden location, AM is often diagnosed late, making the detection of pigmented lesions challenging. Common symptoms include altered bowel habits, obstruction, rectal bleeding, anal pain, and tenesmus, often accompanied by a protruding mass. The melanoma typically appears as an irregular, ulcerated polypoid lesion with brown or black pigmentation [6]. Pan-CK tests are typically negative in AM cases. AM has a poor overall survival rate, with a 5-year survival range of 10-20% [7]. No conclusive evidence supports that one surgical method is superior to others, and a study by Bleicher J et al., compared WLE and APR, highlighting WLE's advantages in avoiding colostomies and reducing morbidity [8]. In a trial including 49 patients, WLE was deemed safe, with only minor complications- three cases of mild infections requiring antibiotics and one case of postoperative bleeding needing a second surgery. No major complications were reported [8]. Given its lack of clear advantages, surgical management should prioritise minimising morbidity [8]. There is ongoing debate about the optimal systemic therapy for this type of melanoma, emphasising the need

for newer approaches like targeted treatments and immunotherapy. Unlike cutaneous melanomas, anorectal and other mucosal melanomas have yet to show significant immunogenicity [9]. Anal melanoma treatment often involves radiation, chemotherapy, and immunotherapy, mainly as adjuvant options unless surgery is unfeasible. While these therapies have improved survival in cutaneous melanoma, their effectiveness in anal melanoma remains uncertain [10]. Targeted medicines and immune checkpoint inhibitor use are likewise debatable, and there is insufficient data to support decisions. The role of checkpoint inhibitors, tyrosine kinase inhibitors, and BRAF/MEK inhibitors in treating AM remains unclear, despite their proven success in cutaneous melanoma over the past decade. Immunotherapy with checkpoint inhibitors has shown limited and inconsistent efficacy in ARM, likely due to the distinct biological characteristics of mucosal melanomas compared to their cutaneous counterparts [10]. Further research and clinical trials are needed to determine the most effective therapeutic strategies for ARM, potentially integrating novel targeted approaches to improve patient outcomes.

CONCLUSION(S)

The AM is an infrequent but highly aggressive malignancy considered by rapid progression and frequent late-stage diagnoses. The absence of standardised staging and treatment protocols, largely due to its biological variability and rarity, complicates its management. Early identification is crucial, as AM is often mistaken for benign anorectal conditions. Given this risk, all haemorrhoidal specimens should undergo histopathological evaluation to rule out anal melanoma, ensuring timely and accurate diagnosis. Once confirmed, appropriate treatment strategies, including surgical excision and possible adjuvant therapies, can be tailored based on individual patient factors to optimise outcomes.

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