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Case Report

Pathology Section

Rectal Melanocarcinoma in an Elderly: A Case Report

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ABSTRACT

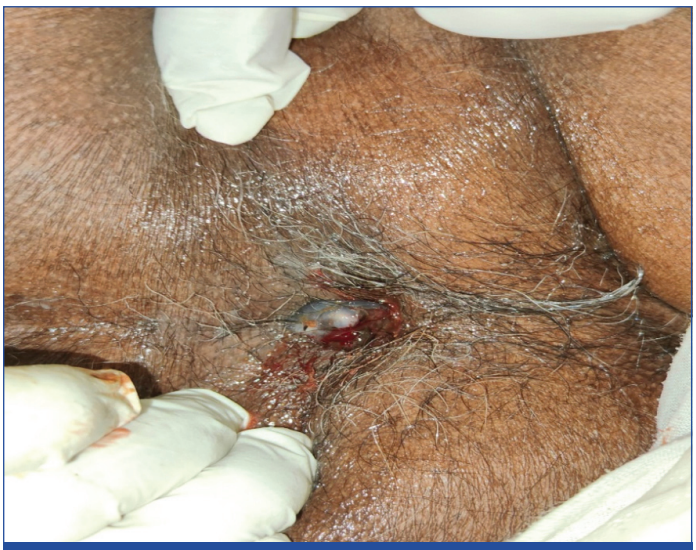
Rectal melanocarcinoma is a rare and aggressive malignancy often misdiagnosed due to non specific presentation. Hereby the authors present a case of a 71-year-old male who presented with mild anal pain and occasional bleeding, along with a 3 cm protruding mass from the anal canal. Clinical examination identified thrombosed external haemorrhoids and a suspicious 2 cm mass. Routine blood tests and Echocardiography (ECG) were unremarkable, but proctoscopic examination confirmed the presence of the mass, which was surgically excised. Histopathological analysis of the excised tissue revealed spindle and polygonal cells with prominent macronucleoli and melanin pigment in the cytoplasm, particularly at the squamocolumnar junction. Microscopic examination at various magnifications supported the diagnosis of rectal melanocarcinoma. Despite surgical intervention, the prognosis for rectal melanocarcinoma remains poor due to its aggressive nature and high potential for metastasis. The present case underscores the critical importance of early recognition and thorough histopathological evaluation of atypical anorectal lesions to avoid misdiagnosis.

Keywords: Anorectal mass, Histopathology, Malignant melanoma, Rare malignancy

CASE REPORT

A 71-year-old male presented with mild anal pain and occasional bleeding, having reported a 3 cm mass protruding from his anal canal, which was associated with discomfort during bowel movements. Clinical examination revealed thrombosed external haemorrhoids and a suspicious 2 cm mass near the squamocolumnar junction. The patient's medical history was unremarkable and he denied any significant weight loss, changes in bowel habits, or family history of malignancy.

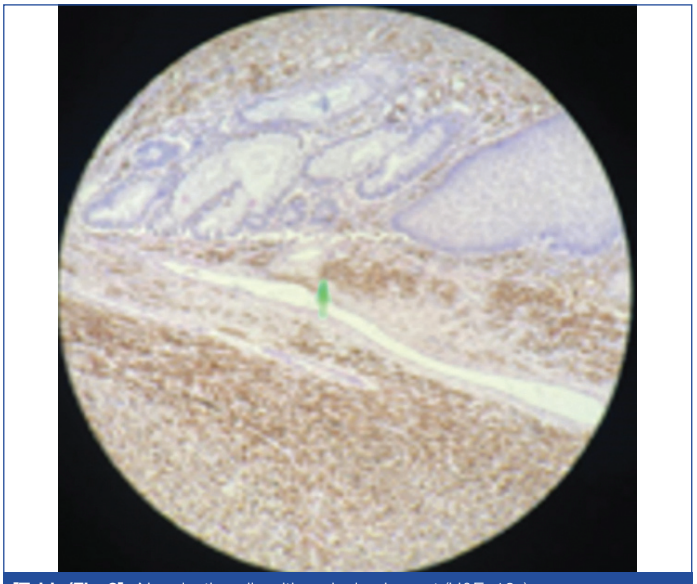
Routine laboratory investigations, including a Complete Blood Count (CBC), renal function tests (urea, creatinine and electrolytes), liver function tests {Alanine Transaminase (ALT), Aspartate Aminotransferase (AST), Alkaline Phosphatase (ALP) and total bilirubin} and coagulation profile, were performed and found to be within normal limits. ECG findings showed no abnormalities. A proctoscopic examination revealed a suspicious anorectal mass, prompting surgical excision. The excised tissue consisted of two parts: thrombosed external haemorrhoids and a brownish-black mass measuring 2.1×1.0×0.5 cm [Table/Fig-1].



[Table/Fig-1]: Rectal melanocarcinoma anorectal lesion.

Histopathological evaluation of the excised mass provided definitive evidence for the diagnosis of rectal melanocarcinoma. The gross morphology of the excised tissue revealed a distinct brownish-black appearance, a hallmark characteristic of melanocytic anorectal lesions. This pigmentation is indicative of melanin production, a defining feature of melanoma. The striking colouration of the mass raised a strong suspicion of melanocytic origin, which was further corroborated by microscopic findings that confirmed the malignant nature of the anorectal lesion.

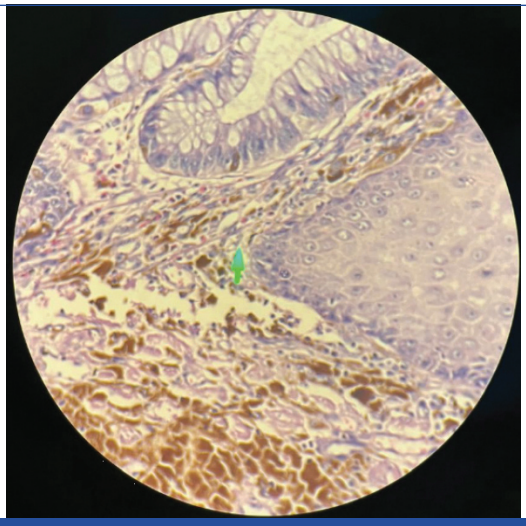
Microscopic examination of the excised tissue provided critical insights into the diagnosis of rectal melanocarcinoma. At 10x magnification, neoplastic cells containing melanin pigment were observed. These cells exhibited characteristic features that confirmed the melanocytic origin of the anorectal lesion, a hallmark of malignant melanoma [Table/Fig-2].



[Table/Fig-2]: Neoplastic cells with melanin pigment (H&E, 10x).

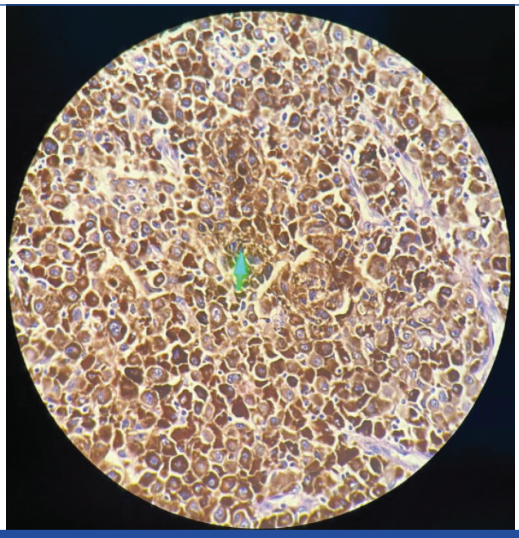
At 40x magnification, spindle-shaped neoplastic cells were prominently visible. These cells demonstrated abundant melanin pigment within their cytoplasm, further supporting the diagnosis of melanocarcinoma

[Table/Fig-3]. The spindle-shaped morphology is a typical feature of melanoma, particularly in its more aggressive forms.



[Table/Fig-3]: Neoplastic spindle cells with melanin pigment (H&E, 40x).

At 100x magnification, polygonal neoplastic cells with distinct macronucleoli were evident. These cells exhibited significant melanin deposits in their cytoplasm, providing further diagnostic confirmation. The prominent nuclear features and melanin pigmentation highlighted the malignancy's aggressive nature and established the diagnosis of the anorectal lesion conclusively [Table/Fig-4].



[Table/Fig-4]: Neoplastic polygonal cells with melanin in the cytoplasm (H&E, 100x).

The findings at these varying magnifications collectively demonstrated the presence of malignant melanoma in the rectal tissue, reinforcing the importance of detailed histopathological analysis for such rare and challenging cases. The histological examination revealed clusters of spindle and polygonal cells with clumped chromatin along the nuclear membrane. Melanin pigment was abundant in the cytoplasm, particularly at the squamocolumnar junction, providing conclusive evidence of malignant melanoma. Based on these findings, the final diagnosis of rectal melanocarcinoma was established.

The patient underwent surgical excision of the tumour with negative margins, which remains the cornerstone of treatment for this condition. Postoperative follow-up included routine imaging {abdominal and pelvic Computed Tomography (CT) scans} and serum tumour markers to monitor for recurrence or metastasis. Despite the aggressive nature of rectal melanocarcinoma, the patient demonstrated no evidence of disease progression during the initial six-month follow-up period.

Adjuvant therapies such as immunotherapy or targeted therapy were discussed but deferred due to the absence of metastatic lesions and the patient's stable clinical condition. The present case highlights the critical need for ongoing surveillance and a multidisciplinary approach to management.

DISCUSSION

Rectal melanocarcinoma is a rare and aggressive malignancy, constituting less than 1% of rectal cancers and less than 1% of all melanomas. The present case highlights the critical importance of early detection and accurate histopathological evaluation in managing this unusual disease. Below, the authors compared the present case findings with previously published cases to provide a broader perspective on the clinical and pathological characteristics, management approaches and outcomes associated with rectal melanocarcinoma.

The present case patient presented with mild anal pain and occasional bleeding, consistent with symptoms reported in several studies of rectal melanocarcinoma. Morlino A et al., noted that rectal melanomas often present with non-specific symptoms such as anal discomfort or rectal bleeding, contributing to delayed diagnosis [1]. Similarly, Tomioka K et al., reported cases initially misdiagnosed as poorly differentiated adenocarcinoma, highlighting the clinical ambiguity associated with this malignancy [2]. In the present case, the initial clinical examination suggested thrombosed haemorrhoids, but further investigation revealed a suspicious anorectal mass, underscoring the importance of maintaining a high index of suspicion for anorectal malignancies when symptoms persist.

Histopathological evaluation in the present case revealed characteristic brownish-black pigmentation due to melanin, along with spindle and polygonal cells with prominent macronucleoli. This aligns with findings from Gavriilidis P et al., who emphasised melanin pigmentation as a hallmark of anorectal melanoma [3]. However, immunohistochemical variability has been observed across cases, as noted by Seya T et al., where KIT and HMB-45 expression differed between biopsy and resected specimens [4]. In the present case, exhibited consistent melanin pigmentation across different magnifications, aiding in the definitive diagnosis. These comparisons highlight the diagnostic challenges posed by histopathological and immunohistochemical heterogeneity in rectal melanocarcinoma.

The present case demonstrated neoplastic cells with abundant melanin pigment, including spindle-shaped cells at 40x magnification and polygonal cells with macronucleoli at 100x magnification. This morphological presentation is consistent with aggressive forms of melanoma, as reported by Zhang F et al., who identified tumour size and cellular morphology as key prognostic indicators [5]. In comparison, the cases reported by Tomioka K et al., showed similar pigmentation and spindle cell morphology but lacked the polygonal cell configuration seen in the present case patient [2]. Furthermore, the present case findings corroborate the observations of Tse JY et al., who described atypical hyperplastic melanocytes within the anorectal epithelium, suggesting a possible origin from melanocytes in the rectal columnar epithelium [6].

Immunohistochemical analysis plays a crucial role in differentiating anorectal melanoma from other malignancies. The present case was diagnosed using histopathological features without the need for advanced immunohistochemistry. However, studies such as those by Khatoon A et al., and Deng T et al., emphasised the diagnostic value of immunohistochemical markers like HMB-45, S-100 and Melan-A [7,8]. The present case, in the absence of such markers suggests that histopathological examination alone, particularly the distinct melanin pigmentation, can be sufficient for diagnosis when characteristic cellular morphology is observed.

Surgical excision with negative margins was performed for the present patient, consistent with the treatment approach recommended by multiple studies. Early-stage diagnosis and surgical resection are crucial for improved outcomes, as highlighted by Youssi Z et al., [9]. In the current case, postoperative follow-up showed no disease

progression over six months, similar to the favorable outcomes reported in cases with complete surgical resection [9]. In contrast, Erdas E et al., documented a recurrence within six months despite surgical intervention, emphasising the aggressive nature of rectal melanocarcinoma [10].

Adjuvant therapies, including immunotherapy and targeted therapy, were considered but deferred in the present case due to the absence of metastatic lesions. This decision aligns with the management strategy discussed by Zhang F et al., who identified surgical resection as the primary treatment modality, with adjuvant therapy reserved for advanced or metastatic cases [5].

The six-month disease-free follow-up in the present case is encouraging, given the typically poor prognosis associated with rectal melanocarcinoma. Zhang F et al., reported a median overall survival of 12 months for rectal melanoma patients, emphasising the importance of early diagnosis and surgical intervention [5]. In comparison, Morlino A et al., noted that aggressive surgical approaches did not significantly improve survival, underscoring the need for comprehensive follow-up and consideration of adjuvant therapies [1]. The present case demonstrates that negative surgical margins and regular imaging can effectively monitor disease progression in early-stage rectal melanocarcinoma.

CONCLUSION(S)

Rectal melanocarcinoma is a rare and aggressive malignancy that is often misdiagnosed due to its non-specific presentation. Early recognition, thorough histopathological evaluation and timely surgical intervention are critical for improving outcomes. Despite advancements in therapy, the prognosis remains poor, emphasising the need for heightened clinical vigilance and awareness to identify and manage this challenging condition effectively.

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