Amelanotic Melanoma of the Rectum: A Rare Case Report
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Abstract: Malignant melanoma accounts for 15% of the primary malignant tumors of the anorectal region. However, up to one-third of the anorectal melanomas are amelanotic and cause diagnostic challenges. They are often mistaken for benign anorectal disease. Anorectal melanoma is uncommon and aggressive cancer with an unfavourable prognosis. The main differential diagnosis is from Lymphoma, Undifferentiated carcinoma of the rectum, poorly differentiated squamous carcinoma of the anal canal & small cell carcinoma. A 60 year old female presented with bleeding per rectum, change in bowel habits and weight loss. Sigmoidoscopy revealed a pedunculated polypoidal mass near the anal verge. Histopathological examination was inconclusive and immunohistochemistry was essential to clinch the diagnosis of amelanotic malignant melanoma.

Keywords: Amelanotic Melanoma, Rectum, Pectinate line, Polypoid

INTRODUCTION
Primary rectal melanoma constitutes 0.1 - 4.6% of anorectal malignancies with a female preponderance [1]. Majority of the cases occur in the 5th and 6th decades [2]. Rectal bleeding is the commonest symptom [3]. They usually present as polypoid pigmented masses [4].

Most cases of melanoma located in the lower rectum represent upward extensions or metastasis of tumors originating in the anal canal [5].

The tumor is usually in or close to the pectinate line, from which it tends to grow towards the rectal ampulla. Sometimes it extends proximally a long way along the submucosa & merges through the mucosa at a high point. Thus simulates a primary rectal tumor. Rectal bleeding, palpable mass and pain are the most common complaints [6].

Anorectal melanoma is an uncommon and aggressive cancer with an unfavourable prognosis [7-10]. Between 0.4 and 1.6% of all melanomas arise in the anorectal region and the anorectal region is the most frequent site of melanoma after the skin and retina [10, 11]. The first case of anorectal melanoma was reported by Moore in 1857 and approximately 500 cases have been reported so far in the literature [10, 11].

CASE REPORT
A 60 year old female presented with history of bleeding per rectum for 15 days, change in bowel habits and weight loss since 2 months. Per-rectal examination revealed no abnormality. Proctoscopy revealed a friable mass around 10 cms from the anal verge. Sigmoidoscopy revealed a pedunculated , polypoid lesion measuring 6 x 6 cms. Biopsy taken from the lesion was sent for HPE.

DISCUSSION
Histopathology
Histopathology revealed a diffusely infiltrating neoplasm composed of cells exhibiting vesicular nucleus, clumped chromatin, prominent nucleolus, increased N: C ratio and cytoplasmic vacuolation. Scattered multinucleated giant cells are seen. The tumor is well vascularized and showed areas of haemorrhage. With these features a differential diagnosis of poorly differentiated carcinoma, malignant melanoma and lymphoma was made.
Immunohistochemistry

Blocks were sent to Tata Memorial Hospital, Mumbai for immunohistochemistry which revealed focal positivity for S100, negative for CD 56, synaptophysin, Chromogranin, CK 20, LCA, CD 20, CD3, ALK- 1, EMA and c-kit. Additional immunohistochemistry revealed positivity for Melan A and HMB 45. Special stain for melanin – Masson’s Fontana silver stain was negative.

With this immunohistochemical profile, a final diagnosis of amelanotic malignant melanoma of the rectum was made. It is important to distinguish metastatic melanoma from primary cutaneous melanoma deposits. Primary melanomas are positive for DMBT1.

The prognosis after abdominoperineal resection is poor [12, 13] with a median survival of 12-24 months [14, 15]. Duration of more than 3 months, perineural invasion, presence of inguinal lymph nodes and amelanotic melanoma have poorer prognosis [15, 16].

Malignant melanoma reported to account for upto 15% of anal primary malignant tumors [17, 18]. It presents as a large, protuberant or ulcerating mass which usually extends to the lower rectum [17]. The tumor originates from the melanocytes, usually found in the transitional zone above the dentate line [17, 18].

Malignant melanoma of anus is highly invasive spreading into the perianal and perirectal tissues, involving haemorrhoidal lymph nodes and early haematogenous spread into the liver and lungs. There are two major histologic subtypes, i.e., epithelioid and spindle cell melanoma. The main differential diagnosis is from lymphoma, undifferentiated carcinoma of the rectum, poorly differentiated squamous carcinoma and small cell carcinoma. Often the tumors lack melanin pigment but if searched carefully, melanin is reported to be found in 50% of the cases. The presence of junctional change in the adjacent mucosa should be considered but this is often destroyed by ulceration [19].

CONCLUSION

Malignant melanoma accounts for upto 15% of anal primary malignant tumors; presents as a large, protuberant or ulcerating mass usually extending into the lower rectum. In this case a final diagnosis of amelanotic malignant melanoma of the rectum was
made by immunohistochemical profile, histopathological examination was inconclusive.

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Abbreviations
HPE- Histopathological Examination, CD - Cluster of Differentiation, CK – Cytokeratin, LCA - Leucocyte Common Antigen, ALK - Anaplastic Lymphoma Kinase, EMA - Epithelial Membrane Antigen, HMB-45 - Human Melanoma Black – 45, DMBT-1 – Deleted in Malignant Brain Tumors -1

REFERENCES