

Primary Rectal Malignant Melanoma: Case Report of a Rare Entity

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ABSTRACT

Malignant melanoma of the rectum is a very rare and aggressive disease. It represents up to 4% of all anorectal malignancies. Rectal melanoma most common presents in the fifth to sixth decades as rectal bleeding or anal pain. Rectal melanoma is often misdiagnosed as hemorrhoids or rectal polyp, leading to a delay in diagnosis and hence treatment. Timely, diagnosis and prompt treatment are extremely important as such cases carry a poor prognosis. We report this case because of its rarity.

KEY WORDS: Anorectum, hemorrhoid, malignant melanoma, rectal polyp, rectum.

Introduction

Malignant melanoma of the rectum is a rare entity with an incidence of 0.5-4% of all anorectal malignancies and <1% of all melanomas.^[1,2] The first case of melanoma of anus and rectum was reported by Moore in 1857.^[3] Anorectal area is the third most common primary site for melanoma after skin and retina, however only 0.4-1.6% of all primary melanomas arise here.^[4] The majority of the patients are females in their fifth or sixth decades presenting with rectal bleeding, which is often mistaken for bleeding associated with hemorrhoids.^[5,6] Prognosis is very poor with a median survival of 24 months and a 5 years survival of 10-15%.^[4,5]

Case Report

A 65-year-old female, presented with altered bowel habits and on and off painless bleeding per anum since 4 months. She was undertaken for colonoscopy which revealed a sessile polyp measuring 2 cm × 1 cm at the anorectal angle. A biopsy was taken from the polyp and sent for histopathological examination. Grossly, the excised polyp was non-pigmented. On microscopy, the sections from the biopsy showed sheets and nests of round to

polygonal cells (Figure 1) with moderate amount of eosinophilic cytoplasm, pleomorphic nuclei with irregular margins and coarse chromatin and prominent eosinophilic nucleoli (Figure 2). Some of the cells showed melanin pigment in the cytoplasm (Figures 2 and 3). A provisional diagnosis of rectal melanoma was made and immunohistochemistry was done for the tissue sections, which showed a positive reaction for HMB45 and Melan A (Figure 4) and negative for Pan CK, CD3, CD20, and CD45.

Following this, the patient was subjected to further investigations to rule out any other focus of primary tumor or any metastases. Dermatological and ophthalmological examinations did not reveal any primary tumor. Computed tomography of the thorax, abdomen and pelvis did not reveal any metastases.

A diagnosis of primary rectal melanoma was made and the patient underwent wide local excision (WLE). She made an uncomplicated recovery and is now under follow-up.

Discussion

All types of melanomas, irrespective of their origins (cutaneous or mucosal), originate from melanocytes, which are cells derived from the embryological neural crest. In fetal life, these cells migrate to many sites throughout the body, most abundantly to the skin and some to the eyes and mucosal surfaces. Thus, cutaneous melanoma is the most common comprising >90% of all melanomas. Of the remaining, i.e., <10%, ocular melanoma accounts for 5%, melanoma of unknown

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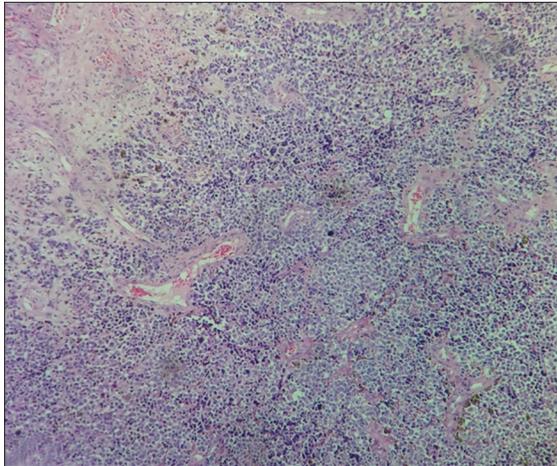


Figure 1: Sheets and nests of round to polygonal cells (H and E, ×40)

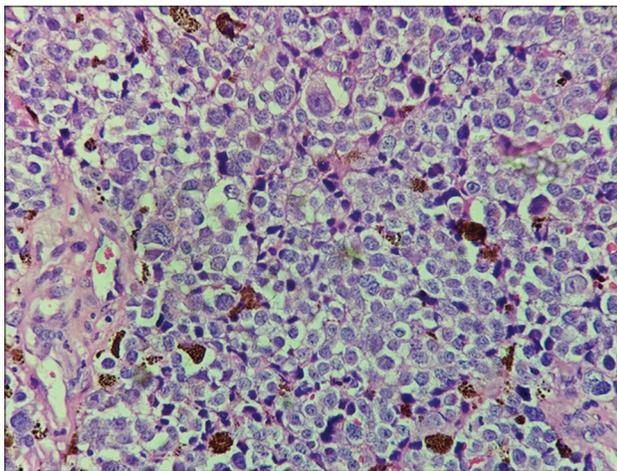


Figure 2: Round to polygonal cells with moderate eosinophilic cytoplasm, pleomorphic nuclei with irregular margins and prominent nucleoli. Few cells show melanin pigment in cytoplasm (H and E, ×400)

origin accounts for 2%, and mucosal melanoma accounts for 1%.^[1,2]

Melanocytes may undergo malignant transformation on exposure to ultraviolet light. However, this does not hold true for rectal melanomas.^[7] The development of melanoma in the rectum may be attributed to immune mechanisms as a higher incidence of rectal melanoma is seen in patients with human papillomavirus and human immunodeficiency virus infections.^[7,8]

Rectal melanoma predominantly affects women and is typically seen in the sixth to seventh decade of life.^[5,9] Our patient was a 65-year-old female which

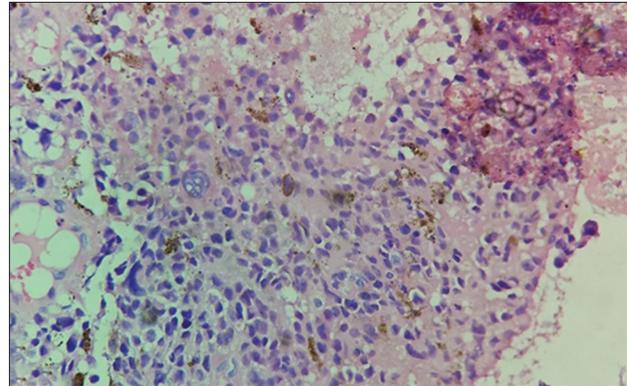


Figure 3: Cells with melanin pigment in cytoplasm (H and E, ×400)

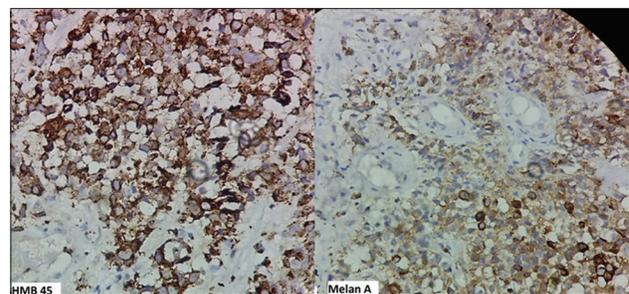


Figure 4: Cells with positive immunoreactivity for HMB 45 and Melan A (×400)

fairly complies with the age and sex distribution of rectal melanoma.

The most common presenting symptom in rectal melanoma is rectal bleeding along with altered bowel habits, local pain, discomfort, feeling of mass, pruritus, and tenesmus.^[4-6,9] Similar symptoms were seen in our patient. Such non-specific symptoms usually lead to a delayed diagnosis and hence a poor prognosis.

Gross appearance of rectal melanomas can also be confusing leading to a delayed diagnosis or misdiagnosis. Melanomas are often misdiagnosed as hemorrhoids, rectal polyps or prolapse.^[4-6,9] The majority of melanomas are pigmented, however, amelanotic forms are also seen^[4] as was seen in our case.

Treatment modalities in rectal melanoma have been a topic of discussion. Some authors support abdominoperineal resection while others prefer WLE, and then there are others who propose there is no advantage to either approach as treatment of the primary tumor does not influence the systemic

course of the disease.^[4] However, all the studies indicate that despite the extent of resection, 75% of patients have a recurrence of tumor with distant and lethal relapse.^[4]

Conclusion

Rectal melanoma is a rare lethal tumor with a median survival of 24 months and a 5 years survival of 10-15%. The presence of non-specific symptoms and absence of obvious gross pigmentation usually lead to delayed diagnosis or misdiagnosis. Hence, malignant melanoma of the rectum should always be kept in mind as an important differential diagnosis in patients presenting with rectal bleeding.

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Financial Support: None; **Conflict of Interest:** None

How to cite this article: Dey A, Riti TKS. Primary rectal malignant melanoma: Case report of a rare entity. *J Med Sci Health* 2015;1(3):21-23.

Date of submission: 06-08-2015
Date of peer review: 05-09-2015
Date of acceptance: 09-09-2015
Date of publishing: 10-10-2015