Melanoma masquerading as a rectal polyp

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Case Report

Abstract
Anal melanoma is a rare and an aggressive malignancy. Unlike cutaneous melanoma, anal melanoma has no known risk factors or precursor lesions. Anal melanoma constitutes only 0.5 to 2% of all anorectal malignancies and less than 2% of all melanomas. Desmoplastic amelanotic melanoma is an unusual variant of malignant melanoma. We report a case of a 34 year old male with complaints of bleeding per rectum and a polypoidal mass protruding through the anus. Histopathological findings along with immunohistochemistry findings were consistent with the diagnosis of desmoplastic amelanotic melanoma. This case is being presented with a brief discussion along with differential diagnoses.

Keywords: Rectal Polyp; Amelanotic Melanoma; Anorectal Malignancy

Introduction
Anorectal melanomas are very rare tumors with a poor prognosis. The diagnosis is difficult and the disease is often incorrectly labeled clinically as hemorrhoids or rectal polyps in about 80% of the cases, causing a delay in diagnosis hence explaining why the patients present with locally advanced disease and metastasis in 60-70%. Amelanotic melanoma represents 25-50% of anorectal melanomas. These show considerable histological variability, of which desmoplastic variant is unusual. We report a case of desmoplastic amelanotic melanoma presenting as a rectal polyp.

Case Presentation
A 34-year-old gentleman came to the hospital with complaints of bleeding per rectum during defecation and a mass protruding from the anus. Proctoscopic findings showed hemorrhoids at 3°, 7° and 11° clock positions. An incidental finding was a polypoidal lesion at 10° clock position which was excised and sent for histopathologic examination.

Pathologic Findings
On gross examination the tissue was a grey white ulcerated polypoidal mass measuring 1.5 × 1 cms. The cut surface was grey white and homogenous. Histopathology examination revealed focally ulcerated stratified squamous epithelium with underlying tumor tissue. The tumor tissue was seen extending up to the resected margin. Tumor cells were arranged in small round to irregular nests and showed fascicular arrangement with intervening collagenous stroma (Figure 1). At places, these tumor cells were in continuity with overlying basal cell layer of surface epithelium and were large with scanty cytoplasm, round to elongated nuclei showing marked pleomorphism and hyperchromatism.

A few of them showed prominent eosinophilic macro nuclei (Figure 2). Mitotic figures of 8-10/10HPF were observed in cellular areas. With the above histopathological findings, differential diagnoses of desmoplastic amelanotic melanoma, anorectal gastrointestinal stromal tumor (GIST), leiomyosarcoma and undifferentiated epithelial malignant tumor were considered.

The superficial parts of the tumor were also positive. The superficial parts of the tumor were also positive. The superficial parts of the tumor were also positive. The superficial parts of the tumor were also positive.

**Immunohistochemistry**

S 100 - STRONG POSITIVE
VIMENTIN - STRONG POSITIVE
HMB 45 - +/- (ESSENTIALLY NEGATIVE)
Microphthalmia transcription factor [Mitf]-POSITIVE
Melan-A and Tyrosinase - POSITIVE
CD 117 - POSITIVE
DESMIN - NEGATIVE
EMA - NEGATIVE
SMA - NEGATIVE

**Prognosis**

Delays in diagnosis and the aggressive nature of the disease cause anal melanoma to frequently present at an advanced stage.

The staging of anal melanoma differs from that of cutaneous melanoma, which is based primarily on thickness in millimeters (Breslow classification). Standard guidelines for adequate staging in mucosal melanoma are not well defined. Anal melanoma can be staged on a clinical basis, focusing on loco regional and distant spread. Stage I is local disease only, stage II is a local disease with regional lymph nodes, and stage III is distant metastatic disease.

The most common sites for metastases include inguinal lymph nodes, mesenteric lymph nodes, hypogastric lymph nodes, para-aortic lymph nodes, liver, lung, skin, and brain.

**TABLE 1: Differential diagnosis (based on immunohistochemistry)**

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<tr>
<th></th>
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<th>Leiomyosarcoma</th>
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<tbody>
<tr>
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**Discussion**

Anorectal melanoma is a rare malignant tumor associated with late detection and a poor prognosis. The first case of the disease was reported by Moore in 1857, and so far approximately 500 cases have been reported in the literature.

Anorectal melanoma is a neuroectodermal neoplasm arising from melanoblastic cells of the mucosal surface. Mucosal melanoma is the third most common melanoma after the cutaneous and ocular varieties (1.3%). Anal canal is the most frequent site for primary GI Melanomas and is 23.8% of all the mucosal melanomas. A timely diagnosis of anal melanoma is difficult as 80% of lesions lack obvious pigmentation and up to 20% of tumors are even histologically amelanotic.

The median age at presentation is 55 years, although the range is wide, 29 to 91 years.

**Clinical features of rectal amelanotic melanoma**

The patient may present with bleeding per rectum, pain, mass protruding through anus, constipation, diarrhea, weight loss or tenesmus. It presents as an indurated, non-ulcerated, and slightly elevated plaque or nodule. It may be flesh-colored rather than melanotic. Pigmentation, when present, is either sparse or irregularly distributed.

**Histopathologic features**

The tumor is composed of elongated hyperchromatic spindle cells distributed singly or in bundles, fascicles, or nests, between variably increased collagen fibers of the papillary and the reticular dermis. The superficial parts of the tumor show epithelioid cells with the cells retaining fibroblast-like features in the deeper areas. Fascicles of hyperchromatic spindle cells aligned at different angles in relation to the long axis of the epidermis are characteristic. A significant percentage of cases show involvement of local neurovascular bundles with infiltration of the adventitia, perineurium or endoneurium and may have a prognostic implication.

Anorectal melanomas display a variety of cytomorphological features with architectural patterns like fasciculation, whorling, nesting, trabeculation, angiocentric, pseudoglandular, pseudopapillary, pseudofollicular and pseudorosetting. Stromal changes include myxoid, desmoplastic changes and very rarely pseudoangiosarcomatous change, granulomatous inflammation or osteoclastic giant cell response.

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Conclusion

To summarize, desmoplastic amelanotic melanoma of the anal canal is a rare neoplasm with poor prognosis. Awareness of its presentation and histopathological variations can help in early detection and management.

Conflict of interest

The authors declare that they have no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

References