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Case Report
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Amelanotic Melanoma Presenting as Rectal Polyps

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Abstract

Anorectal melanoma is an aggressive mucosal melanocytic malignancy. It is rare, and approximately one third of them are amelanotic. They can resemble benign polypoid lesions of ano-rectum. Symptoms are non-specific and include anal bleeding, anal mass, and pain. The pre-operative diagnosis remains difficult as it is not suspected due to rarity of the disease. Immunohistochemical studies on biopsies help in diagnosis. Anorectal melanoma carries a poor prognosis as it is often diagnosed late. Optimal therapy is still evolving and often surgical resection remains the therapy of choice.

Keywords: Amelanotic melanoma; Rectum; Polyp

Introduction

Most of the melanomas originate in the squamous epithelium and mucosal melanomas are rare, accounting for approximately 1.2% of all melanomas. Primary malignant melanoma of anus and rectum is a rare but an aggressive neoplasm. It accounts for less than 1% of all melanomas and approximately 4% of anorectal tumors other than adenocarcinoma [1]. Nearly one third of anorectal melanomas are amelanotic and can be mistaken for benign polypoid lesions or even hemorrhoids [2]. Women in the fifth or sixth decade are affected commonly, and usual symptoms are bleeding per rectum and obstructive defecation. It can mimic benign conditions like hemorrhoids or rectal polyp. The optimal surgical procedure for primary anorectal melanoma is evolving; options include wide local excision or endoscopic mucosal resection or an abdominoperineal resection.

We present our case of a 78-year-old lady with amelanotic melanoma of ano-rectum.

Case Presentation

A seventy-eight-year-old lady presented with complaints of passing mucus per rectally for six weeks with a history of sense of incomplete evacuation and flatulence. No history of abdominal pain, passing blood in stools or loose motions. No vomiting. She felt weak and felt that she lost some weight, but past weight records were not available. She was hypertensive and diabetic and was receiving medications for the same. In view of her age and symptoms she was advised colonoscopy which revealed anorectal polypoidal lesion (Figure 1) and internal hemorrhoids. Rest of the entire colon was normal. It was difficult to take biopsies due to slimy surface and soft nature of the tumor but suction biopsy technique yielded good chunk of tissue. She was also advised a CT scan of abdomen and pelvis which revealed distal rectal polyp between 2 to 4 o' clock position with tiny few, lymph nodes in anorectal fossa on either side.

Histopathology of the biopsy revealed tumor mass comprising of round to oval cells arranged in sheets, exhibiting abundant eosinophilic cytoplasm with vesicular nuclei, clumped chromatin, prominent eosinophilic nucleoli and high Nuclear: Cytoplasmic (N:C) ratio (Figure 2). The tumor was vascular and showed areas of hemorrhage. No pigment or mucin was seen. Neither anal nor rectal lining epithelium was seen. Possibilities of poorly differentiated carcinoma or metastatic deposits or anorectal melanoma were considered. The blocks were subjected for immunohistochemistry which showed tumor cells to be immunoreactive to Vimentin, HMB 45 (Figure 3) and Melan A (Figure 4) and immune-negative for Pan CK, p63 and CK 5/6 confirming the lesion to be amelanotic melanoma. The patient was referred to an onco-surgeon and Abdominoperineal (AP) resection with permanent colostomy was done. The histopathology of the resected specimen revealed it to be mucosal amelanotic melanoma of anal canal, restricted to mucosa, no lympho-vascular or neural invasion seen and none of seventeen pericolic nodes showed metastatic deposits. She made a good recovery and is being followed up.

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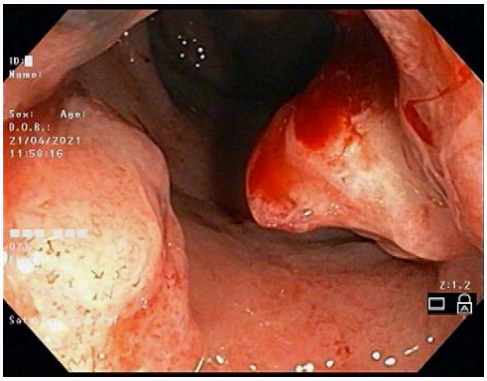

Figure 1: Colonoscopic appearance.

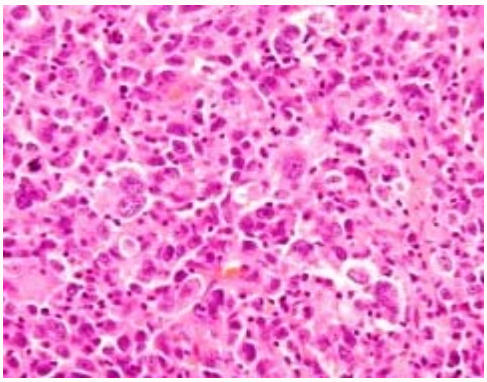

Figure 2: Shows tumor mass consisting of round to oval cells arranged in sheets, exhibiting abundant eosinophilic cytoplasm with vesicular nuclei, clumped chromatin, prominent eosinophilic nucleoli, high N:C ratio. (H&E 40x × 10x).

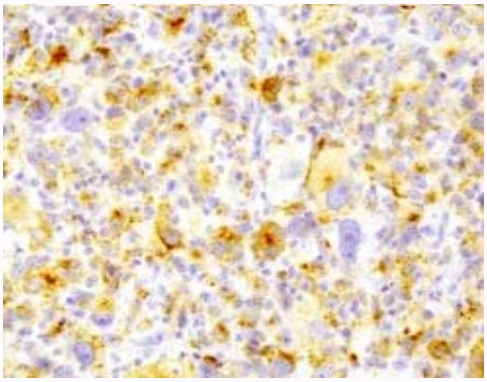

Figure 3: Tumor cells immunoreactive to HMB 45.

Discussion

Anorectal melanoma is a neuroectodermal neoplasm originating from melanoblastic cells of mucosal surface [3]. It was first described in 1857 by Moore and to date nearly 500 cases have been reported in literature [2]. It is a rare tumor with poor prognosis because of lympho-vascular invasion [4]. Mucosal melanomas are rare and account for nearly 1% of all melanomas. Anorectal melanomas account for fewer than 25% of all mucosal melanomas. Nearly one third of anorectal melanomas are amelanotic and can be mistaken for benign polypoid lesions of ano-rectum [2]. It is common in fifth or sixth decade of life especially in women. Exact diagnosis requires immunohistochemistry otherwise it can be missed. More than sixty

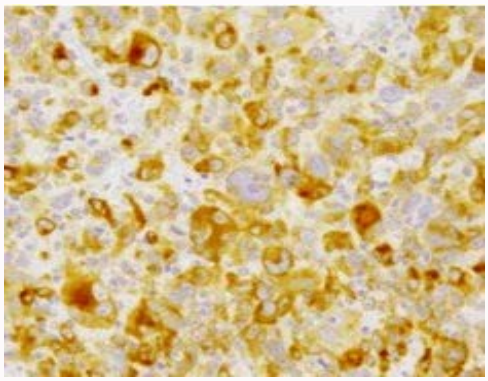

Figure 4: Tumor cells immunoreactive to Melan A.

percent of patients of anorectal melanomas have either lymph node involvement or distant metastases due to hematogenous spread, making it an advanced disease at diagnosis, resulting in poor outcome [4,5].

The knowledge about the pathogenesis of anorectal melanomas is limited and obscure. Two theories have been proposed for origin of melanomas in the gastrointestinal tract. According to first hypothesis, neural crest derivatives such as APUD cells in gastrointestinal tract may de-differentiate into melanocytes and subsequently undergo malignant transformation [6]. According to second hypothesis, ectopic migration of melanocytes in gastrointestinal tract melanocytes was suggested as the mechanism and this was confirmed by identifying benign melanocytes in cases of esophageal carcinoma and esophagitis [7-9].

Symptoms of anorectal melanoma are like those caused by hemorrhoids, polyps, rectal cancer and include:

A. Passing mucus and blood per rectum

B. Anal pain or discomfort, tenesmus

C. Feeling of rectal fullness or incomplete evacuation

D. Externalization of tumor and changes in bowel habits

E. Pruritus Ani

F. Inguinal masses [10].

Usually, the lesion starts on or close to the pectinate line, from which it tends to grow proximally. It can extend proximally in the submucosa & simulate a submucosal rectal tumor [11]. Colonoscopy is required for obtaining a tissue biopsy. Endoscopic ultrasound can detect tumor thickness and surrounding nodal status. Distant spread can be detected by Contrast-enhanced CT scan, PET scan or MRI. CT scan appearances, of primary rectal malignant melanomas can vary from intraluminal mass in the distal rectum or usually without luminal obstruction. Often there is perirectal infiltration and enlarged lymph nodes [3]. Histologically the differential diagnosis includes lymphoma, undifferentiated carcinoma of the rectum, poorly differentiated squamous carcinoma of the anal canal & small cell carcinoma. Immunohistochemical stains like HMB-45, S-100, and Melan A are required for definitive diagnosis [2].

Knowledge about the prognostic parameters, staging and treatment protocols is limited and evolving. Generally, prognosis is poor due to the late diagnosis, aggressive behavior of the tumor because of the rich vascular and lymphatic supply of the ano-rectal

mucosa [10]. Zhang et al. [12] reported misdiagnosis at presentation in more than half of their patients, 46 of 79 (58%). Half of these patients were misdiagnosed as carcinoma and half as benign lesions. Better understanding of the disease biology and immunology can help in optimizing the treatment of the disease. For this, identification of molecular determinants of response to target therapies and immunotherapy are being studied.

Conclusion

Only few hundred cases of amelanotic melanoma of rectum have been reported in the literature so far. Thorough examination, coupled with a high index of suspicion, will prevent delayed diagnosis. Despite its rarity, anorectal melanoma should be considered when unusual or unclear anorectal lesions are detected. Immunohistochemistry is essential for histological diagnosis of amelanotic melanoma. We also required immunohistochemical profile for final diagnosis of amelanotic malignant melanoma as routine H and E stain was inconclusive.

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