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Case Report

An amelanotic melanoma of the rectum: report of a rare aggressive primary tumor

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ABSTRACT

Anorectal melanoma is a rare and highly aggressive malignancy accounting for 0.4% to 1.6% of all melanoma and 0.5% of all anorectal cancer. About 80% cases are confused with haemorrhoids, rectal polyp, rectal ulcer or adenocarcinoma as they commonly present with rectal bleed. We herein reported a case of a 55-year male presenting with a 6-months history of vague abdominal pain, constipation, tenesmus, weight loss, and passage of blood in stool. Clinical examination and proctoscopy revealed a fungating mass. CT scan showed a mass involving lower rectum with periserosal infiltration. The patient underwent total abdominoperineal resection and specimen was sent for histopathological examination. Gross examination showed a fungating growth measuring 3.5×2.5×2 cm along with 8 lymph nodes. On microscopy rectal mucosa was normal, submucosa showed spindle shape tumor cells arranged in sheets & fascicles having pleomorphic nuclei, high N:C ratio with moderate amount of eosinophilic cytoplasm along with areas of necrosis and no evidence of lymphovascular emboli. 6/8 lymph nodes showed metastatic deposits along with large number of atypical mitotic figures. H and E examination suggested diagnosis of gastrointestinal stromal tumor (T2N1Mx). A panel of IHC markers was advised for definite diagnosis. The tumor cells were reactive for HMB45, S-100, Ki67 thus rendering a diagnosis of amelanotic melanoma. Due to absence/small numbers of melanin granules in H and E and diversity in morphology of neoplastic cells, misdiagnosis as lymphoma, gastrointestinal stromal tumor and sarcoma is very common. Hence IHC plays an indispensable role in the diagnosis of amelanotic melanoma of rectum.

Keywords: Amelanotic melanoma, Mutations, Immunohistochemistry, Total abdominoperineal resection

INTRODUCTION

Primary anorectal melanoma is a rare and highly aggressive malignancy with poor prognosis.[1] It accounts only 0.4% to 1.6% of all melanoma and 0.5% of all anorectal cancer whereas in these 30% are amelanotic.[2] Approximately 61% of anorectal melanoma are presented at time of distant metastasis so it is a very aggressive malignancy.[1] Typically, patients are presented in sixth to eighth decades of life with predominance in white, and female.[3] The aetiology of anorectal melanoma is different from that of cutaneous melanomas. The etiology of cutaneous melanoma is BRAF gene mutations associated with UV light are not seen in anorectal melanoma and c-kit gene mutations are more common.[4] About 80% cases are confused with hemorrhoids, rectal polyp, rectal ulcer or adenocarcinoma as they commonly present with rectal bleed.[5] Due to absence or small numbers of melanin granules misdiagnosis as lymphoma, carcinoma and sarcoma is common and immunohistochemistry (IHC) is required for confirming the diagnosis of amelanotic malignant melanoma.[6]

CASE REPORT

A 55-year-old man presented to the surgery outpatient department with a six-month history of vague abdominal

pain, worsening constipation, tenesmus, weight loss and passage of blood in stool. On clinical examination and proctoscopy, a fungating mass was seen externally which protruded out of the rectum. CT scan showed a mass involving lower rectum infiltrating the serosa. The patient underwent total abdominoperineal resection and the resected specimen was sent for histopathological examination to the pathology department. On gross examination a fungating growth measuring 3.5×2.5×2 cm was seen which was 2.5 cm away from the anal verge. Cut surface of growth showed solid white area along with hemorrhage and eight lymph nodes were also identified. Microscopy of the H and E sections revealed normal rectal mucosa, muscularis propria showed spindle shape neoplastic cells arranged in fascicles and sheets having pleomorphic nuclei having high N:C ratio with moderate amount of eosinophilic cytoplasm along with areas of necrosis with no evidence of lymphovascular emboli or perineural invasion. The large number of atypical mitotic figures (10/10 HPF) were also seen. Proximal and distal resected margins were negative for tumor. Six out of eight lymph nodes showed metastatic deposits. Many dilated and congested blood vessels were also seen. Based on microscopic examination, the possibility of gastrointestinal stromal tumor (T2N1Mx) was suggested and a panel of IHC markers was advised for a definite diagnosis, which included HMB 45, S 100, SOX 1, vimentin, Pan CK, CD 117, non-specific enolase (NSE), epithelial membrane antigen (EMA), Desmin and Ki67. The tumor cells were immunohistochemically reactive for HMB45, S 100, Ki67 and focal reactive for vimentin and NSE, whereas all other markers were nonreactive. Thus, a diagnosis of malignant amelanotic melanoma was rendered on immunohistochemistry.

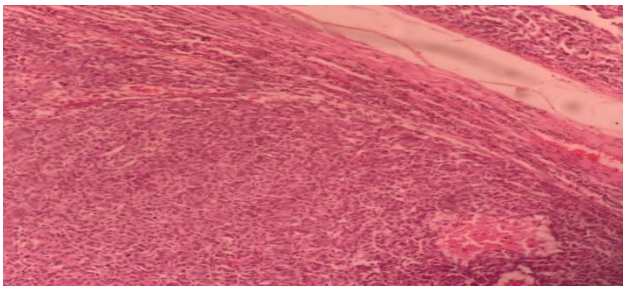

Figure 1: H and E section show spindle shape neoplastic cells (10×).

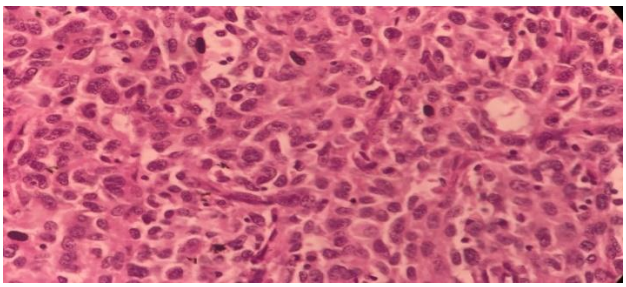

Figure 2: H and E section show spindle shape neoplastic cells (40×).

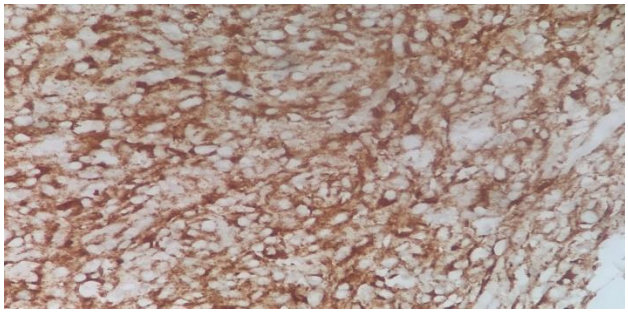

Figure 3: Positive HMB 45 staining (40×).

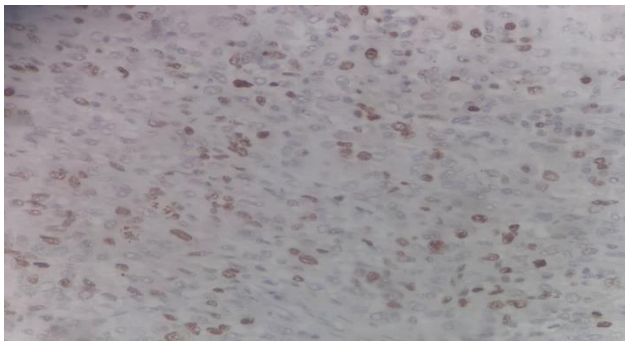

Figure 4: Positive HMB 45 staining (40×).

DISCUSSION

Anorectal melanoma was first described by Moore in 1857.[1] Melanomas can affect any anatomical structure where melanocytes are present e.g., epidermis, eyes, nasal cavity, oropharynx, urinary tract, rectum, anus and vagina. Most common site for melanoma is epidermis, it constitutes 91.2% of all melanoma cases. Mucosal melanomas are very rare it constitutes only 1.3% of all melanomas.[7] The main risk factor in case of cutaneous melanoma is BRAF gene mutation associated with UV rays but in mucosal melanomas c-Kit mutation is more common, oxidative stress and immunosuppression related to this.[8] Anorectal melanoma's patients presents with non-specific symptoms like altered bowel movement, rectal bleeding and pelvic pain, therefore can be mistaken as benign condition.[10] Many cases approximately 90%, diagnosed after metastasis to liver and lung via lymphatic spread hence it is an aggressive tumor.[10] The diagnosis of anorectal amelanotic melanoma is very complicated due its histological variability and can be misdiagnosed as lymphoma, carcinoma and sarcoma.[7] Kolosov et al in their study reviewed 17 cases of primary melanoma of the anorectal region and found that most of them were women (n=11, 64.71%) and the most common presenting symptom, in eight patients (47.06%), was bleeding per rectum, four complained of pain (23.53%) and three complained of altered bowel movement (17.65%). In their study, on H and E four types of tumor cell morphology were seen: epithelioid, spindle, lymphoma-like and pleomorphic type. These tumor cell types were in combinations: mainly with epithelioid cells and most commonly epithelioid with spindle cells. 41% cases did

not show any pigmentation in tumor cells but strong positive staining with HMB 45, Melan A, S100 and vimentin, confirming the diagnosis amelanotic melanoma.[7] Bell et al in their study described a case initially presented as inflamed hemorrhoids and on CT revealed possibility of malignancy. On H and E section of biopsy biphasic malignant tumor cells with no melanin pigment was identified and immunoprofile showed immunopositivity with S-100 and SOX10 for spindled component and immunopositivity with desmin, AE1/AE3 and EMA for epitheloid component. On the basis of these findings confirm the diagnosis of primary dedifferentiated amelanotic anorectal melanoma was made. Patient died 20 days after the diagnosis was rendered due to lung and liver metastasis. This showed it was a highly aggressive tumor with poor prognosis.[11] Sahoo et al in their study described a case of ulceroproliferative growth which was located seven cm away from the anal verge and on colonoscopy suspected undifferentiated adenocarcinoma. Histopathological examination of the specimen revealed clusters and nests of neoplastic cells invading the lamina propria and on immunohistochemistry tumor cells were immunoreactive for HMB-45 and S-100. Thus, a diagnosis of amelanotic melanoma was made. While the chemoradiation therapy, patient developed metastatic inguinal lymphadenopathy, it showed amelanotic melanoma as a highly aggressive tumor.[1] Magalhaes et al 2018 in their study described in anorectal mucosal melanoma melanocytes can present in several forms (pleomorphic, epithelioid, spindle cells, etc) and can misdiagnose as other tumors such as sarcomas, GIST and undifferentiated carcinomas. The definite diagnosis made with the help of immunohistochemistry S-100, HMB-45, Melanin A and Mart-1 antibodies.[9]

CONCLUSION

Anorectal melanoma is a rare aggressive disease. Diagnosis is difficult as it is easily mistaken for hemorrhoids, polyp and patients most often present with advanced stage. Clinicians and pathologists must be aware of its varied clinical and histomorphological spectrum and the diagnostic difficulties associated with it. Thus, IHC plays indispensable role in its diagnosis.

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