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Case Report

Anorectal Melanoma: Unexpected tumor in unexpected population

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ABSTRACT

Melanoma is a rare tumor in Saudi population. Therefore, diagnosing melanoma among the Saudi population in unexpected location is difficult without a high index of suspicion and the help of laboratory investigations, including ancillary immunohistochemical (IHC) studies. Here, we present the first reported case of anorectal melanoma in the central region of Saudi Arabia and the second reported case in the whole country. This is a report of a 59-year-old Saudi female patient who showed an abdominal large rectal mass. Urgent colonoscopy examination revealed a large fungating rectal mass with the ulcerated surface. The microscopic examination of the colonoscopy biopsies showed colonic mucosa with a lamina propria infiltrated by pleomorphic atypical round/oval neoplastic cells arranged in sheets, clusters, and trabecular patterns. The IHC stains showed that the neoplastic cells were positive for S100 and Melan-A, but negative for CKpan, CK7, CK20, CDX2, CD45, DOG1, chromogranin, and synaptophysin. The overall workup revealed a diagnosis of the primary anorectal melanoma.

Keywords: Anorectal melanoma, anorectal tumors, melanoma, Saudi Arabia

Introduction

Melanoma is a malignant melanocytic neoplasm.[1] It arises mainly in the skin, but it can arise rarely as a primary neoplasm in other locations such as uvula of the eye, nervous system, gastrointestinal tract (GIT), and genitourinary tract. All types of melanoma are considered rare in Saudi Arabia since they represent 0.2–0.3% of all the reported malignancies in the country, according to "*Cancer Incidence Report 2016*" issued by Saudi Health Council.

Anorectal melanoma represents only 1.3% of all melanomas in general[2] and <1% of all colorectal malignancies.[1] It is a highly aggressive tumor with poor prognosis since its 5-year survival rate is <10%.[2] In Saudi Arabia, anorectal melanoma is extremely rare, and only one case had been reported in Makkah, which is located in the western region of Saudi Arabia,[3] making this case the first reported case in the central region specifically and the second reported one in Saudi Arabia in general.

Case Report

A 59-year-old Saudi female patient presented to the emergency room complaining of bleeding per rectum for the past 2 weeks. On physical examination, she was thin and pale. Her vital signs were HR 98 beat/min, RR 16 breath/min, BP 131/98 mmHg, and temperature 36.8°C. Her hemoglobin and complete blood count report revealed microcytic hypochromic anemia. Per rectal examination revealed large internal hemorrhoids. An abdominal computed tomography scan showed a large rectal mass. Urgent colonoscopy examination performed and revealed a large fungating rectal mass with the ulcerated surface. Biopsies from this mass were taken for histopathological examination. The microscopic examination of these biopsies showed colonic mucosa with a lamina propria infiltrated by pleomorphic atypical round/oval neoplastic cells arranged in sheets, clusters, and trabecular patterns. The neoplastic cells have scanty to moderate amount of cytoplasm [Figure 1] with scanty brown pigmentation and oval nuclei without prominent nucleoli. Frequent mitotic figures and areas of necrosis were noticed. A panel of immunohistochemical (IHC) stains was

done and showed that the neoplastic cells were positive for S100 [Figure 2] and Melan-A [Figure 3] and negative for CKpan, CK7, CK20, CDX2, CD45, DOG1, chromogranin, and synaptophysin. Depending on these histopathological and IHC results, a diagnosis of melanoma was made.

After that, the patient was re-examined again from head-to-toe looking for any suspicious primary skin melanomatous lesion, but no suspected lesion was found. Additional radiological investigations did not reveal any internal suspicious mass other than the rectal one. Based on that, the diagnosis of the primary anorectal melanoma was made and the patient had been referred to a specialized oncology center for additional management and follow-up.

Discussion

Anorectal melanoma is an extremely aggressive tumor. In fact, it is a fatal disease since the patients have 2-year period and 6-month period mean survival without and with the presence of metastasis, respectively, regardless of the treatment.[1,3] In most cases, the cause of death is mostly due to the distant metastasis.[1]

The exact etiopathogenesis of anorectal melanoma is still unclear.[2] Most of the patients are elderly females above 60 years of age.[1] In contrast to our case, this disease is commonly affecting white Caucasian peoples.[1]

Similar to what happens in our case clinically, most of the anorectal melanoma cases are presented with bleeding per rectum.[1,4] Other signs and symptoms include anorectal pain, tenesmus, pruritus, changes in bowel habits, and sings and symptoms related to the metastasis.[1,2,4] Since anorectal melanoma share the same signs and symptoms of some common inflammatory and neoplastic anorectal lesions,[1,2,4] the diagnosis of such disease is extremely difficult and needs a high index of suspicion.[1,5]

Although it is a helpful tool, colonoscopy alone cannot be used to confirm the diagnosis of anorectal melanoma.[6] The gold standard for diagnose of melanoma, in general, is hematoxylin and eosin plus IHC stains of sections prepared from a lesional biopsy; however, the diagnosis of anorectal melanoma is a clinicopathological one with seven specific criteria, including (a) single lesion, (b) no other currently primary sites for melanoma, (c) no history of previous melanoma or atypical melanocytic proliferation in other sites, (d) no enlargement of the draining lymph nodes, (e) presence of anorectal lesion that is proved by the biopsy to be melanoma, (f) presence of intramucosal lesional melanocytes either overlying or adjacent epithelium, and (g) presence of lymphocytic infiltration at the periphery of the mass.[2]

Radiological studies are helpful tools in evaluating the original neoplasm, the presence of possible metastasis, and the

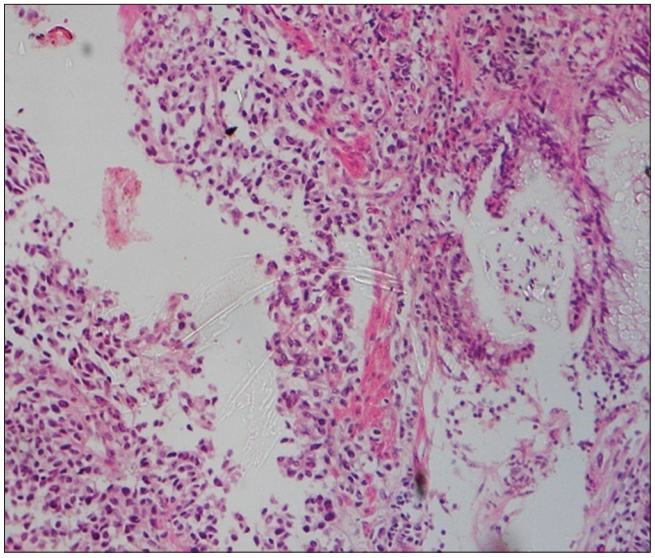

Figure 1: H and E stain show that neoplastic cells have scanty to moderate amount of cytoplasm and oval nuclei without prominent nucleoli (20×)

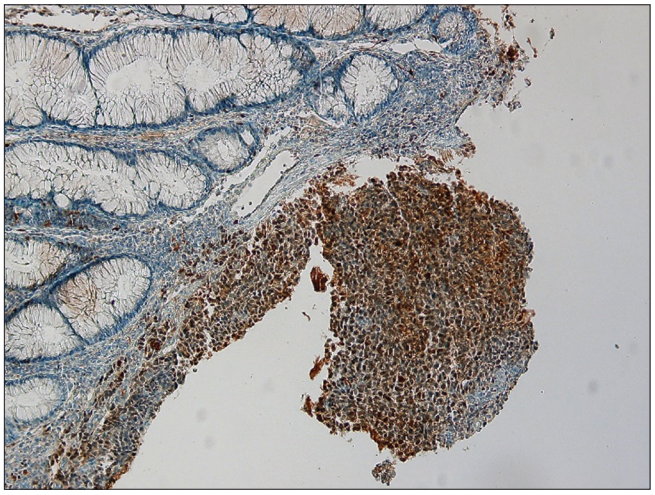

Figure 2: S100 IHC stain is highlighting the neoplastic cells (×10)

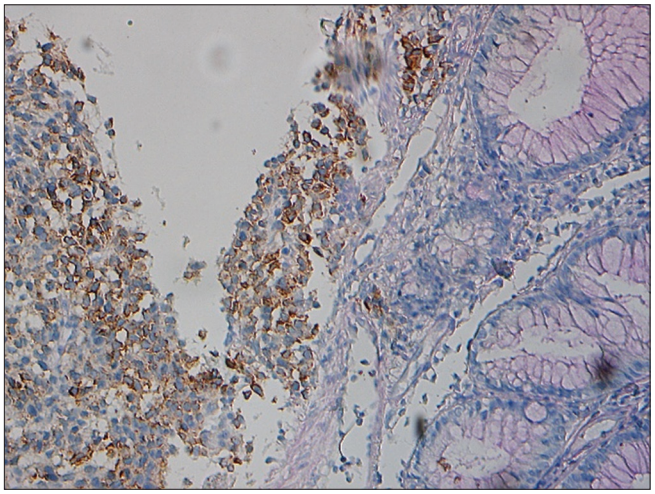

Figure 3: Melan-A IHC stain is positive in the neoplastic cells (20×)

therapeutic response of the neoplasm to the treatment; however, it cannot be used to confirm the diagnosis.[2]

Endoscopically, most of the cases arise at the transitional zone between the rectum and the anal canal[1] and there is no major difference between those originate from the rectum, and those originate from the anal canal regarding the survival rate.[7] Anorectal melanomas are usually arising as polypoidal masses that can be easily mistaken as benign lesions, [1] unlike our case, which presented as fungated ulcerated mass. The size of the anorectal melanomas varies from few millimeters to several centimeters.[1]

Anorectal melanoma is similar to its cutaneous counterpart microscopically, immunohistochemically, and genetically.[1] Therefore, metastatic cutaneous cases can be differentiated by the clinicopathological correlation only. The major histopathological differential diagnoses of anorectal melanoma are (a) adenocarcinoma, (b) squamous cell carcinoma, (c) neuroendocrine tumors, (d) lymphoma, gastrointestinal stromal tumor, and (e) melanosis coli.[1] Here, the morphology and the ancillary studies will be helpful in differentiating those tumors from anorectal melanoma. Although the differential diagnosis includes adenocarcinoma, these two tumors can be found together in the same patient as collision tumors.[5,8-10]

Anorectal melanoma can metastasis through (a) lymphatic vessels to the regional lymph nodes, (b) blood vessels to the lungs and breasts, and (c) the peritoneum.[2,11-13] The later may result in peritoneal carcinomatosis.[11]

The management plan is different from case to case since there is no specific guideline for those conditions due to their rarity.[2] Different treatment modalities were used in those patients, including surgery (both wide local excision and abdominoperineal resection), chemotherapy, radiotherapy, and targeted therapy, but none of them shows improvement regarding to the survival of the patients.[1,2] They are done only to improve patients' morbidity.[1]

Conclusion

In summary, although anorectal melanoma is rare in Saudi Arabia, the disease does exist, and doctors should think about it, especially if the histopathological features of GIT biopsy do not show the classical picture of the common GIT tumors. With the help of laboratory ancillary studies, these tumors will not be missed.

Availability of Data and Material

The data used in this study are available and will be provided by the corresponding author on a reasonable request.

Patient Consent

Written informed consent was taken from the involved patient.

Competing Interests

The authors declare no conflicts of interest.

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