International Journal of Science and Research Archive
eISSN: 2582-8185
Cross Ref DOI: 10.30574/ijsra
Journal homepage: https://ijsra.net/

(CASE REPORT)

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Amelanotic anorectal melanoma: A case report

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International Journal of Science and Research Archive, 2024, 12(02), 1426–1429

Publication history: Received on 17 June 2024; revised on 28 July 2024; accepted on 30 July 2024

Article DOI: https://doi.org/10.30574/ijsra.2024.12.2.1365

Abstract

Anorectal melanomas represent less than 1% of all anorectal cancers and 0.3% of malignant melanomas [1]. Rectal localization is rare, accounting for 0.3% of all melanomas [2]. It is often secondary to infiltration of the rectal mucosa by an anal-derived process originating from melanocytes normally present in the squamous epithelium of the pectineal zone and in the transitional epithelium above the pectineal line [6-7]. Its prognosis is dreadful due to the early occurrence of metastases. We report an observation from the Department of Medicine B, Ibn Sina University Hospital, and Rabat.

Keywords: Amelanotic anorectal melanoma; Rare; Diagnosis; Histopathology; Metastases.

1. Introduction

Anorectal melanomas represent less than 1% of all anorectal cancers and 0.3% of malignant melanomas [1]. In addition to its rarity, this localization is characterized by the absence of standards for its management and by its poor prognosis, with a five-year survival rate of less than 20% [1-2]. Around 70% of these lesions are pigmented, but 30% of anorectal melanomas are amelanotic. Detection of melanoma immune markers by immunohistochemistry (IHC) is necessary to confirm the diagnosis of malignant amelanotic melanoma [3]. Early diagnosis can improve the prognosis of these patients. We report a case of amelanotic anorectal melanoma from our department.

2. Observation

A 65-year-old man reported the appearance of an anal mass accompanied by proctalgia proctalgia and rectal discharge, evolving in a context of altered general condition. Proctological examination revealed an irregular, painless, ulcerating-bourging process in the anal margin (Figure 1). Endoscopic examination revealed a hemi-circumferential ulcerating process extending to the lower rectum (Figure 2). Histologically, the colonic mucosa was infiltrated by a diffuse tumor proliferation consisting of pleomorphic cells with enlarged, irregularly contoured, anisokaryotic nuclei with prominent nucleoli and scant cytoplasm (Figure 3). Immunohistochemical staining supported the diagnosis of submucosal malignant melanoma, as the markers: anti PS 100 antibody positive and anti Melan A antibody positive (Figure 4). The patient underwent a thoraco-abdomino-pelvic CT scan to study locoregional and distant extension, finding a T3 process with secondary hepatic and pulmonary localization. The patient was referred to an oncology center to start his chemotherapy regimen.

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Figure 1 Ulcerative-bourgeous process at the anal margin

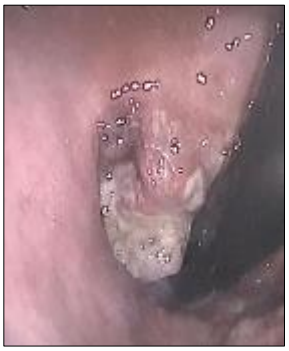

Figure 2 Endoscopic view of tumor infiltration of the lower rectum in retrovision

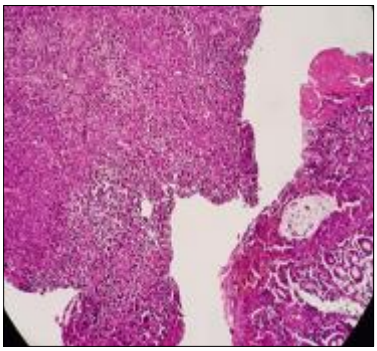

Figure 3 Poorly differentiated tumor process

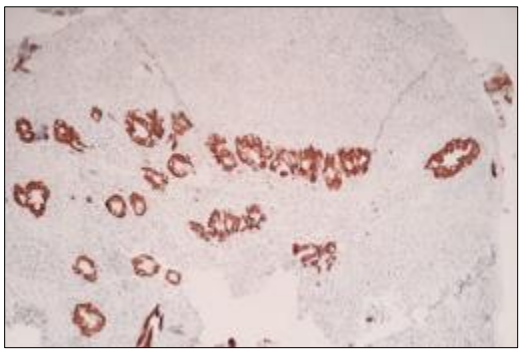

Figure 4 Immunohistochemistry with positive anti-Ps 100 and anti-Melan A antibodies in favor of MAR

3. Discussion

Anorectal melanoma (MAR) was first reported by Moore in 1857 [4]. It is an extremely rare and aggressive malignancy. Amelanotic MAR is even rarer, with the same epidemiological and clinical features as pigmentary MAR, occurring most often between the fifth and sixth decades, with a female predominance [5]. Rectal localization is rare, accounting for 0.3% of all melanomas [2]. It is often secondary to infiltration of the rectal mucosa by an anal-derived process originating from melanocytes normally present in the squamous epithelium of the pectineal zone and in the transitional epithelium above the pectineal line [6-7]. Anal melanomas have the same immunohistochemical and ultrastructural characteristics as their cutaneous counterparts [8]. Amelanotic MAR may present as a mass, manifest as proctalgia or bleeding, and/or may be amelanotic and can only be identified by histopathological examination. However, a diagnostic biopsy is not recommended for cutaneous malignant melanoma, unlike MAR, which is essential. While not all pigmented anorectal tumors are malignant melanomas, not all malignant melanomas are pigmented [9]. Histopathological findings are similar to those of melanomas from other sites; melanin identification and immunohistochemical staining for S100, HMB-45 and/or Vimentin confirm the diagnosis [10]. Differentiation between amelanotic MAR and other tumors such as Paget's disease, lymphoma, undifferentiated carcinoma and gastrointestinal stromal tumor (GIST) can be difficult on histological criteria alone. Seya et al [8] reported an amelanotic MAR that was misdiagnosed on endoscopic biopsy as neuroendocrine carcinoma or GIST due to reactivity with CD56, S100 and CD117, and malignant melanoma was excluded because HMB-45 was negative. However, study of the surgical specimen showed reactivity with HMB-45 and Mart-1 in both the biopsy and the surgical specimen [8]. In cases of suspected amelanotic MAR, the immunohistochemical panel should include S-100, vimentin and the two specific antibodies HMB-45 and Mart-1. Once the diagnosis has been established, the next step is to determine whether the tumor is primary or metastatic. The presence of junctional activity beneath the squamous epithelium is indicative of a primary malignant melanoma. Due to late diagnosis and rapid progression, amelanotic MAR, like pigmentary melanoma, is accompanied by distant metastases in 60% of patients at the time of final diagnosis [11]. After histological diagnosis, a full staging work-up with CT scan of the abdomen and thorax or magnetic resonance imaging is required to rule out metastatic disease.

Amelanotic MAR requires multidisciplinary management, similar to that of pigmented MAR. Surgical resection is the mainstay of treatment [12], ranging from wide local excision to perineal abdominal amputation.

The role of adjuvant chemotherapy has not been established. The prognosis is poor, whatever the therapy used, and the most important predictive factors are disease stage, duration of symptoms, tumor size and lymph node status [12].

4. Conclusion

Amelanotic anorectal melanoma is a rare aggressive tumor. There is as yet no standardized staging system in the literature, nor any standard medical or surgical treatment. Early detection is therefore essential to reduce mortality. Early diagnosis is the key to improving the survival rate of patients with these unusual melanoma variants.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed. All authors contributed to the conduct of this work. All authors also declare that they have read and approved the final version of the manuscript.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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