**Abstract**

We present the case of a 60 year old patient suffering pain and the sensation of a foreign body in the anal region associated with traces of blood in stools. Digital rectal exam (DRE) revealed a hardened lesion located on the wall of the anal canal. Colonoscopy revealed a raised proliferating lesion with a blackish color which was about 2 inches in diameter. This was compatible with an anal canal malignancy. We proceeded to a biopsy and immunohistochemistry study which tested positive for S-100 and negative for HMB-45. A multi-slice helical chest, abdominal and pelvic CAT scan ruled out metastatic tumors and lymphadenopathy. The patient underwent local transanal excision of the partially pigmented tumor. Post-surgical histopathological results confirmed the diagnosis of malignant anal amelanotic melanoma positive for S-100. The sample tested weakly positive for Melan-A and positive for KI-67. The favorable outcome of the procedure led to the patient’s discharge 3 days after surgery.

**Keywords**

Anal neoplasms, melanoma amelanotic, Peru.

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**INTRODUCTION**

Anal tumors are rare neoplasms of the digestive tract. Anal cancer accounts for 4% of all malignancies of the lower gastrointestinal tract. (1, 2) Approximately 80% are of squamous cell origin, 10% are adenocarcinoma and the remainder consists of other types of malignant tumors such as sarcoma, lymphoma, and melanoma. (3) In this group with the lowest percentage, each type of tumor has specific characteristics: lymphoma is a lymphoid tissue tumor, sarcoma has possible mesenchymal origins, (4) and melanoma originates in the melanocytes. (5) Melanoma may or may not be pigmented and can be found in any part of the body. It is frequently located on the skin, followed by the eye and the anus. (6) Although the anus is the third location and is the most common location for gastrointestinal primary melanoma, its presentation is rare. (7)
Although the perianal region was found to be normal during physical examination, a rectal examination showed a hardened 2 cm in diameter lesion located in the anterior wall of the anal canal. For this reason he was referred for a colonoscopy.

The colonoscopy performed five days later showed a blackish, highly proliferative lesion of about 2 cm in diameter which was compatible with a malignant neoplasm of the anal canal associated with prolapsed hemorrhoids next to the lesion. There were no other alterations found in the colonic mucosa. Histopathological diagnosis of a biopsy confirmed epithelial malignant anal amelanotic melanoma (Figure 1).

Physical examination, colonoscopy and biopsy were confirmed by another higher level facility in the city of Lima eight months after the first consultation. Further immunohistochemistry was positive for S-100 positive and negative for HMB-45 n.

Subsequently, a multicut helical thoracic abdominopelvic CT scan ruled out the presence of tumors and metastatic lymph nodes. With the diagnosis confirmed, the patient underwent a partial transanal local excision of the pigmented tumor. The lesion was located between the anal base and the dentate line and occupied 60% of the circumference with small three millimeter in diameter satellite nodes in the surrounding mucosa. The patient’s outcome was favorable, and he was discharged three days later.

The postsurgical histopathological results were positive for S-100, weakly positive for Melan-A and positive Ki-67 and confirmed a diagnosis of malignant anal amelanotic melanoma (Figures 2, 3 and 4).

DISCUSSION

Malignant anal amelanotic melanoma is a rare entity which accounts for between 0.5% to 2% of all anal malignancies and less than 2% of melanomas in general. It has been reported that this entity is more common in women and occurs most often from the fifth decade of life onwards. It is mainly spread via the lymphatic system and evolves rapidly with extensive local invasion. The prognosis for this very aggressive cancer is bad. Despite surgery, patients have median survival time of only 20 months, and most patients die within 5 years of diagnosis. Death usually occurs due to patients’ advanced ages and early metastases. According to reported cases, the most frequent dissemination location is in the lungs (40% of cases) followed by the central nervous system (CNS) (38% of cases) and less often to places such as the liver, bones and intestine.

Clinically, patients present rectal bleeding or diarrhea with tenesmus. The most common lesion is a nodular mass protruding through the anus. It can be pigmented or amelanotic (lacking pigment or with minimal residual pigmentation). It is worth adding that, because the main symptom is rectal bleeding, many of these lesions are confused with hemorrhoid disease or external hemorrhoid thrombosis. This confusion delays diagnosis. Furthermore, the differential diagnosis must consider multiple anal conditions such as anal squamous cell carcinoma, basal cell carcinoma, Bowen's disease and Paget’s disease. In this case, the patient initially presented pain and the sensation of a foreign body followed later by stools with traces of
blood. He was later diagnosed with malignant anal amelanotic melanoma associated with hemorrhoids.

A biopsy is necessary for diagnosis of malignant anal amelanotic melanoma. Immunohistochemistry markers highly suggestive of melanoma such as S-100 protein, HMB-45, and the expression of Melan-a are very useful for diagnosis. The S-100 protein has a high sensitivity (97% to 100%) but a low specificity (75% to 85%) while HMB-45 melanoma antigen is very specific and less sensitive (69% to 93%). (13) In this patient tests for these markers were requested in order to confirm the diagnosis. The resulting positive S-100 and negative HMB-45 indicated that the lesion was amelanotic melanoma.

We also found that only 48% of patients had definitive pathological diagnoses before surgery. This indicates that the diagnosis of this disease is complex and requires the use of various tests for confirmation. (2)

The treatment of choice for this type of cancer is surgery. (5) In a study comparing the median survival time and disease-free time of local excision versus abdominal-perianal resection, similar results were obtained for the two techniques. (15) In addition, adjuvant melanoma treatment including radiation therapy and interferon can be considered (16) although it should be noted that this was not included in this patient’s treatment.

One limitation of this study is that we have not been able to follow the evolution of the patient over time, so we will not know if there is a recurrence of the malignancy. Also, no brain image study was done to rule out central nervous system metastases, one of the most common locations. On the other hand, since this was a patient referred from an isolated rural area, there is no clinical history of the first health facility he went to nor any information about this patient’s evolution during the months he waited for treatment in Lima.

According to the literature, in most reported cases of malignant anal amelanotic melanoma patients are in advanced stages due to metastases which implies a poor prognosis. For this reason it is important to encourage medical practitioners to perform digital rectal exams as part of clinical examinations and to perform colonoscopies to screen for colon cancer in the entire adult population over 50 years for early detection and treatment of this cancer.

Finally, from the case report and literature review we conclude that malignant anal amelanotic melanoma is a rare disease that is usually detected in later stages. However, despite the development of various forms of adjuvant therapy and improvements in surgical techniques, it continues to present poor prognoses. Therefore, it is highly important to consider this disease during differential diagnosis for early diagnosis and management.

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REFERENCES