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Shedding Light on a Rare Condition a Case Report and Literature Review on Primary Anorectal Melanoma

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Abstract: Anorectal melanoma is a rare and highly aggressive cancer, representing 0.5-1% of anorectal tumors and 1-2% of all melanomas. It predominantly affects older adults, with nonspecific symptoms like rectal discharge and pain, often leading to misdiagnosis. We present the case of a 50-year-old diabetic female diagnosed with locally advanced anorectal melanoma, who underwent abdominal-perineal amputation but succumbed to an ischemic stroke four months post-surgery. Treatment options are debated, with surgery being the primary method, though the prognosis remains poor due to early metastasis. While newer therapies, including tyrosine kinase inhibitors and immunotherapy, offer potential, their effect on survival is unclear. This case underscores the importance of considering anorectal melanoma in patients with unexplained anorectal lesions.

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I. INTRODUCTION

Anorectal melanoma is a very rare neoplasm accounting for 0.5% to 1% of anorectal tumours and only 1 to 2% of all melanomas (1) In the digestive tract, the anorectal region is the primary site of melanoma development. Its prognosis is appalling, given the non-specific nature of the symptoms and the early onset of metastases (1,2). Anorectal melanomas do not behave like other more common anal neoplasms, such as squamous cell carcinomas, which can be treated with aggressive multimodal therapy (3). Traditionally, treatment of anorectal melanoma involves complete surgical resection of the tumour in order to control the disease locally. This may be achieved by wide sphincter-sparing local resection or abdominoperineal amputation (AAR) in the case of large tumours or when wide local excision is not feasible (4).

II. CASE REPORT

We present the case of a 50-year-old female patient, type 2 diabetic on oral anti-diabetics, who presented to our clinic with a feeling of a prolapsed anal mass associated with moderate-intensity proctalgia that had been evolving for 1 year, Complicated 9 months later by the onset of low-grade rectal discharge and anal incontinence, evolving in a context of altered general condition with anorexia, weight loss and marked asthenia. Somatic examination revealed an altered patient with a PS score of 2. Examination of the anal margin revealed a tumour process approximately 7 cm in diameter, ulcerated and budding, friable and haemorrhagic on biopsy, with a suspicious appearance, blackish in places, prolapsed through the anus (Figure 1).

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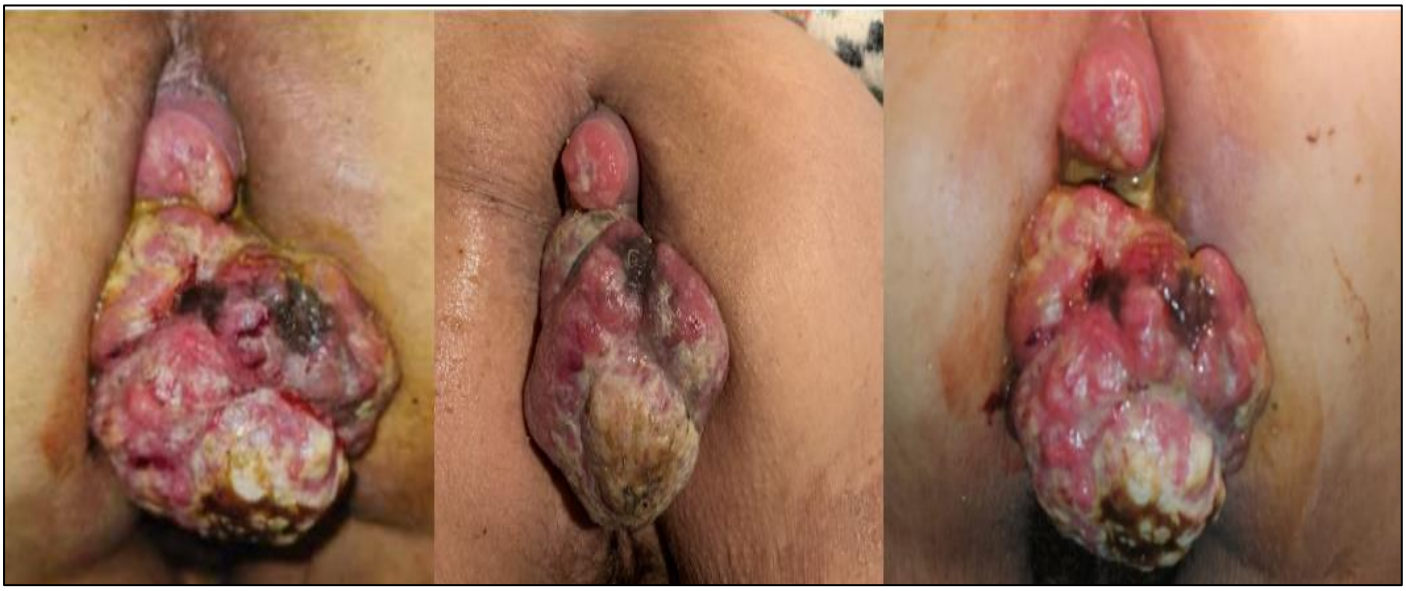

Fig 1 Tumour Process Extending into the Rectum, Friable, Bleeding On Contact and Blackish in Colour.

Biopsies were taken and macroscopic and microscopic anatomical examination revealed undifferentiated tumour proliferation, with immunohistochemical examination giving the histological appearance of a malignant melanoma, positive for Melan A 103 and HMB 45, with focal involvement and no cytokeratin expression (Figure 2).

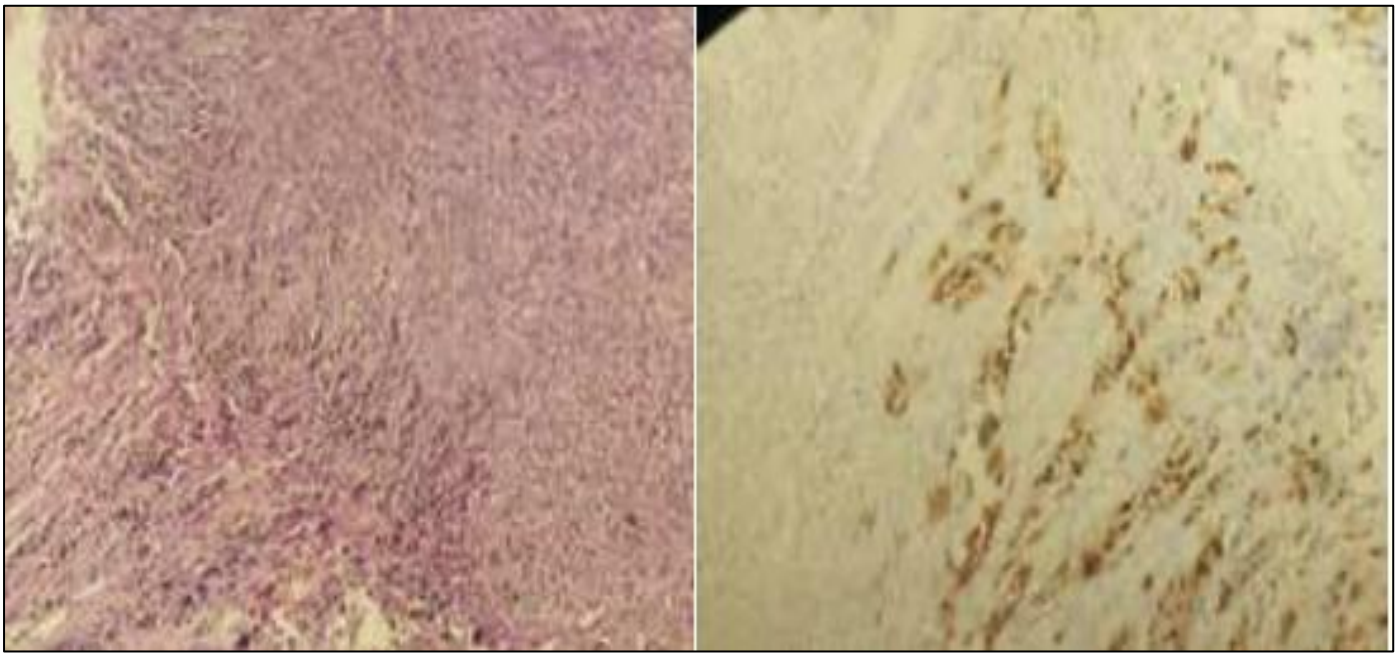

Fig 2 Histological appearance of an anal melanoma positive for Melan A103 and HMB 45

An initial laboratory work-up showed normocytic normochromic anaemia with haemoglobin at 9g/dl. A thoraco-abdomino-pelvic computed tomography scan revealed a voluminous lesion of the rectum, centred on the anal canal, measuring 7*8*15 cm, budding downwards and outside the anal margin and bulging upwards into the middle rectum, infiltrating, anteriorly, the retro-vaginal space, the meso-rectum and the pre-sacral space, with nodules measuring 11 mm in the largest case, and the sphincters. Imaging also revealed lumbo-aortic lymph nodes, the largest measuring 8 mm, associated with hypogastric lymph nodes measuring 5 mm on the left, and thoracic lingual micronodules, the largest measuring 5.6 mm, associated with a micronodule in the internal segment of the middle lobe, of secondary metastatic origin.

Additional PET scanning revealed a locally advanced hypermetabolic tumour of the anal canal, associated with right inguinal and left obturator lymph nodes and bilateral pulmonary nodules that were not hypermetabolic and did not appear pathological. The case was then discussed at a multidisciplinary consultation meeting, where the decision was taken to perform radical surgery. The patient underwent abdominal-perineal amputation, and the post-operative course was straightforward. The patient died 4 months later following an extensive ischaemic stroke.

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III. DISCUSSION

Melanomas develop in organs that contain melanocytes, such as the epidermis, eyes, nasal cavity, oropharynx, vagina, urinary tract, rectum and anus (5). Primary anorectal melanoma is a rare tumour, first described in 1857 by Moore. Since then, around 500 cases have been reported in the literature. These tumours represent 0.1-0.5% of anorectal cancers and 1.5% of all melanomas (2). This pathology of elderly subjects most often affects patients between the sixth and eighth decade, with a clear preponderance of women, since between 50% and 75% of patients in case series are women (2,3,6).

From an aetiopathogenic point of view, the theory of chronic irritation remains the most likely, given that sun exposure is ruled out in this locality. Primary anorectal melanoma implants on either side of the pectineal line, hence the term 'anorectal' (2). The rectal location of melanoma is most often secondary to invasion 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).

Clinically, the symptoms are not specific. As with any other neoplasm, the most common symptoms are rectal discharge, more or less associated with a sensation of anorectal mass in 75% of cases. To a certain extent, reports also mention proctalgia, transit disorders, anal pruritus or the chance discovery of inguinal adenopathy. (1,2). These relatively non-specific symptoms are likely to give rise to the mistaken suspicion of benign haemorrhoidal disease, leading to a delay in diagnosis. The delay between the onset of symptoms and diagnosis is in fact approximately 4 to 5 months (3).

On proctological examination, primary anorectal melanoma presents in more than 50% of cases as an ulcero-vegetative or polypoid, pedunculated tumour. The characteristic blackish colour of melanoma is present in 50% of cases (1,2), as was the case in our patient.

Histologically, malignant melanomas are known for their diversity, mimicking a number of tumours, including lymphomas, carcinomas and sarcomas (4).

The diagnosis is confirmed by the detection of melanin pigments within the tumour following Fontana Masson staining (7). However, in the majority of cases, an immunohistochemical study is essential for the diagnosis of achromic lesions. Melanoma is positive for S-100 protein, HMB-45, MelanA and vimentin. It is negative for CEA, cytokeratin and epithelial membrane antigen (1,2,4). The value of endorectal echo-endoscopy and magnetic resonance imaging in assessing depth of penetration, lymph node status and peri-anorectal soft tissue involvement is now well documented (1). The disease is polymetastatic due to lymphatic dissemination, but essentially haematogenous (8). This haematogenous dissemination is extremely frequent and often occurs early. According to some studies, 20-30% of patients have metastases at the time of diagnosis (2).

The staging of anal melanoma differs from that of cutaneous melanoma, which is based primarily on thickness in millimetres (Breslow classification), although tumour thickness is considered by some authors to be the main factor influencing prognosis (4,9). It is classified on a clinical basis, with emphasis on locoregional and distant spread. Stage I is local disease, stage II is local disease with regional lymph node metastases, and stage III is distant metastatic disease (9).

The prognosis is poor, with an estimated 5-year overall survival rate of 10-20%, with a median survival time of 12-18 months (1,5). According to the authors, although the overall misdiagnosis of anorectal melanomas did not influence the results, cases specifically misdiagnosed as haemorrhoids had a statistically significant shortening of survival (10). They also noted that these patients were more likely to have more advanced disease with systemic metastases at the time of correct diagnosis (3).

In the light of the rarity of anorectal melanomas, there has been no real randomised study of the condition, so there is no established consensus on treatment. Surgical management remains the treatment of choice, but its modalities remain controversial, ranging from abdomi- noperineal amputation (AAP) with inguinal and pelvic lymph node dissection, which appears to be a major prognostic factor influencing survival, to localised removal of the tumour(6). Initially, abdominoperineal amputation was considered to be the only technique for local control of the disease. However, recent studies have not shown this technique to be superior to local excision in terms of survival and recurrence (5). In fact, it is associated with higher morbidity and significant functional repercussions associated with definitive colostomy (11). Local resection, on the other hand, has the advantage of being less aggressive, with a shorter length of stay and reduced impact on digestive function, although with a greater number of local recurrences (4,9). Local excision is recommended as first-line treatment if the excision margins are healthy and the tumour is accessible. In this case, some teams recommend local adjuvant radiotherapy after conservative surgery and/or in invaded lymph node drainage areas, in addition to lymph node dissection (12). In the event of local recurrence, radical surgery with PAA should be performed. In metastatic forms, excision of the primary tumour using PAA or palliative local excision may be proposed if this proves necessary because of the risk of local complications, but without any benefit in terms of survival.

Other recommendations have been proposed by Weyandt et al (13), in analogy with the surgical management of cutaneous melanomas, depending on the thickness of the tumour:

- For a Breslow index of less than 1 mm, local excision with preservation of the anal sphincters and a safety margin of 1 cm is recommended.
- Between 1 and 4 mm in thickness, the safety margin is 2 cm and the anal sphincters may be preserved if they are not invaded.

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- Finally, a PAA is recommended when the Breslow index is greater than 4 mm.

This being the case, irrespective of whether patients have undergone abdominoperineal amputation or wide local excision, it is the achievement of an R0 resection with microscopically healthy margins that would positively influence the prognosis of the disease. Whichever method is chosen, in the end many patients will progress to metastatic disease, as neither resection method can control lymphatic progression (14).

Recently, progress has been made in the therapeutic arsenal used in the treatment of anorectal melanoma, notably with tyrosine kinase inhibitors, which have produced satisfactory results in the treatment of metastatic mucosal melanoma in patients with c-KIT mutation. In the case of metastatic anorectal melanoma without c-KIT mutation, certain chemotherapies alone or in combination (dacarbazine, vinblastine, cisplatin) and immunotherapies (interferon-alpha2b, interleukin 2) have induced tumour responses, although there is no certainty as to their impact on overall survival. (9) However, most patients succumb regardless of the therapeutic strategy chosen, due to the aggressive nature and rapid progression of the tumour (15).

IV. CONCLUSIONS

Due to the rarity of this pathology, it is impossible to carry out prospective studies. As a result, the management of anorectal melanomas is not well defined, as we only have data from retrospective studies of a very small number of patients, studied over a long period - sometimes decades. In conclusion, the message we wish to convey through this case is that, despite its infrequent occurrence, anorectal melanoma should always be considered in the case of an uninhabited anorectal lesion.

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