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Primary Anorectal Melanoma: A Case Report

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ABSTRACT

Malignant anorectal melanoma is a malignant tumour that develops from the pigmentary system. It is a rare tumour, accounting for less than 1% of anorectal cancers and 0.3% of malignant melanomas. Its prognosis is poor due to the early onset of metastases. We report a case.

A 78-year-old female patient with hypertension was admitted for rectal bleeding that had been ongoing for two months, with no other associated symptoms but a deterioration in her general condition. A proctological examination revealed a polypoid tumour with a blackish appearance located 3 cm from the anal margin. Abdominal examination revealed hepatomegaly with a hepatic splenic flexure at 14 cm. Colonoscopy revealed a polyploid mass 2 cm in diameter extending 3 cm in a hemicircular pattern, with a blackish appearance, and scattered nevi on the rectosigmoid mucosa.

Pathological examination of the biopsies concluded that the diagnosis was anorectal melanoma, with the presence of anti-PS100 and anti-HMB45 antibodies on immunohistochemistry. Staging investigations revealed liver metastases.

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KEYWORDS:

Rectal bleeding; Anorectal melanoma; Liver metastases

INTRODUCTION

Anorectal melanomas account for less than 1% of all anorectal cancers and 0.3% of malignant melanomas, ranking third after the skin and the eye. This site is characterised, in addition to its rarity, by the absence of standardised management protocols and by its poor prognosis, with a five-year survival rate of less than 20%. Early diagnosis can improve the prognosis for these patients. Based on our observations and a review of the literature, we discuss the clinical, therapeutic and progressive characteristics of this rare condition.

CASE REPORT

A 78-year-old female patient with hypertension was admitted for rectal bleeding that had been present for two months, with no other associated symptoms but a deterioration in general health.

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Clinical examination revealed a performance status of 4 and hepatomegaly. Proctological examination revealed a blackish mass 2 cm from the anal margin; total colonoscopy showed a blackish polypoid tumour located 2 cm from the anal margin, extending 3 cm in a hemicircular pattern, and nevi of the rectosigmoid mucosa. Histological examination of the biopsies confirmed anorectal melanoma, with immunohistochemical positivity for anti-PS100 and anti-HMB45 antibodies. The thoraco-abdominal-pelvic CT scan revealed a budding parietal thickening of the lower rectum with hepatomegaly, the site of seven lesions suspected to be of secondary origin.

DISCUSSION

Malignant melanoma is a malignant tumour that develops from the pigmentary system, affecting organs containing melanocytes such as the epidermis, eyes, nasal cavity, oropharynx, vagina, urinary tract, rectum and anus. Anorectal localisation is rare, accounting for 0.3% of all melanomas.

The disease is more common in older individuals, with a peak incidence between the ages of 60 and 70, as reported by LEILA et al [1]. These findings are consistent with those of our patient.

In terms of aetiopathogenesis, the hypothesis of chronic

irritation remains the most likely, given the exclusion of sun exposure at this site. The rectal location of the melanoma is most often secondary to infiltration of the rectal mucosa by a process originating in the anus, arising from the melanocytes normally present in the squamous epithelium of the pectinate zone and in the transitional epithelium above the pectinate line [1, 2].

As for the diagnosis, it can only be confirmed in the absence of any synchronous sites (skin, eye) and in the absence of a history of melanoma excision, regardless of its location [1,2]. The clinical symptoms are varied and non-specific, dominated by rectal bleeding, an anal mass, rectal syndrome and a deterioration in general health in the event of metastases, as reported by Haddad and colleagues [2,3]. Our patient reported rectal bleeding and a deterioration in general health.

Clinical examination may immediately reveal a mass protruding from the anus or a blackish discolouration. On proctological examination, primary anorectal melanoma most commonly presents as an ulcerative, vegetative tumour or a pedunculated polypoid lesion. The characteristic blackish colour of melanoma may be observed; this feature was also noted by Haddad et al,[2] which was the case in our patient. Histological confirmation of the diagnosis of anorectal melanoma is identical to that for melanomas in other locations. It relies on the detection of melanin pigment within the tumour using the classic Fontana stain. The use of immunohistochemistry may be necessary in atypical forms, utilising anti-Melan A, anti-HMB45 and PS100 antibodies [1,2]. In our patient, immunohistochemistry with PS100 and HMB45 staining was positive. This is a polymetastatic tumour due to lymphatic and, above all, haematogenous spread. Therefore, a comprehensive staging assessment must always be carried out, including a colonoscopy to look for synchronous lesions, a rectal echoendoscopy or a pelvic MRI to assess wall thickness, lymph node involvement, and a thoraco-abdomino-pelvic CT scan or PET scan to investigate lymph node involvement and distant metastases. A colonoscopy was performed on our patient and the thoraco-abdomino-pelvic CT scan revealed liver metastases.

With regard to treatment options, surgery, endoscopy and radiotherapy may be used. The surgical procedure is most commonly an abdominoperineal resection (APR) with inguinal and pelvic lymph node dissection, or localised tumour excision as described in the study by Othmane et al. [4].

However, endoscopic treatment via mucosal resection or submucosal dissection has recently been proposed as a therapeutic alternative to surgery in localised forms [1,5]. The role of immunotherapy as a neoadjuvant treatment is now recognised as effective, as demonstrated in the study by Radabe.

N. Amaria et al. observed a high rate of complete tumour response in operable patients who received neoadjuvant immunotherapy combining Relatlimab and nivolumab [6]. Adjuvant chemotherapy or radiotherapy following surgery does not appear to offer any survival benefit compared with surgery alone. Radiotherapy offers a major benefit in palliative cases. Its combination with local excision allows for local tumour control similar to that achieved with abdominoperineal resection [1]. This observation is of diagnostic interest as rectal bleeding may indicate malignant anorectal melanoma, a tumour most often diagnosed at the metastatic stage.

CONCLUSION

Anorectal melanoma is a rare condition; its prognosis is poor due to the frequency of metastatic forms at the time of diagnosis. Surgery via abdominoperineal resection or local excision is the treatment of choice for these tumours, although it does not significantly improve survival. New targeted therapies (BRAF and MEK inhibitors) and immunotherapy (ipilimumab, pembrolizumab, nivolumab) offer real hope and have a significant impact.

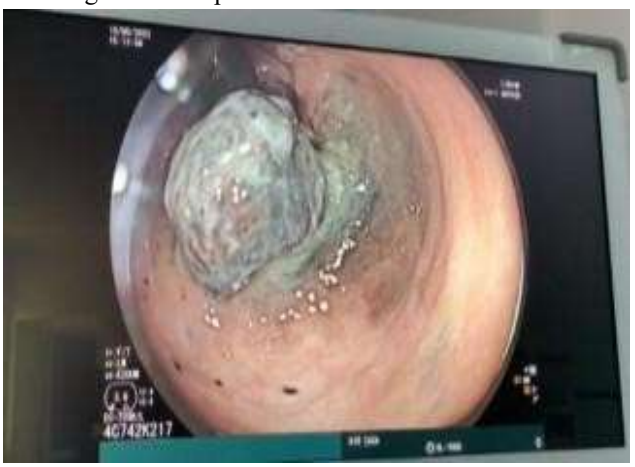

Figure: Colonoscopy: a blackish polypoid mass 2 cm from the anal margin, associated with naevi: a pigmented form of anorectal malignant melanoma

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