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RESEARCH ARTICLE

ANAL MALIGNANT MELANOMA PRESENTING AS A PRIMARY TUMOR: A RARE CASE REPORT

Dr. Balachandran Premkumar, *Dr. Sayed Mohammed Afsal, Dr. Sheena Ali, Dr. Shubankar, Dr. Ranjini Ramachandran

Department of General and Laparoscopic Surgery, Apollo Speciality Hospital, Chennai, Tamil Nadu, India

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**Corresponding author*

ABSTRACT

A fifty year old hypertensive male presented in the outpatient department with the chief complaints of bleeding per rectum associated with tenesmus while passing stools along with a complaint of a mass protruding from the rectum. A colonoscopic evaluation and biopsy revealed a lesion at the anal region which was suggestive of a malignant melanoma, while the PET CT revealed a hypermetabolic circumferential wall thickening involving the anal canal and distal rectum with metastatic pelvic nodes, and hepatic metastasis. He was then taken up for a Laproscopic Abdomino perineal resection with an end colostomy. His histopathology with immunohis to chemistry studies were suggestive of high grade malignant melanoma

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INTRODUCTION

Anal melanoma is an uncommon and aggressive disease. The anorectum is the third most common location of malignant melanoma after the skin and retina. The most common symptom is rectal bleeding, which is often mistaken for bleeding associated with hemorrhoids. Diagnosis is very difficult, and initial diagnosis may be incorrect in 80% of all cases. For patients with anorectal malignant melanoma, treatment strategy includes surgery, chemotherapy, and radiotherapy. However, the tumor tends to be considerably resistant to radiotherapy and shows a poor response to chemotherapy. The choice of wide local excision (WLE) or abdominoperineal resection (APR) is also controversial. The prognosis is very poor, with less than 20% survival five years after diagnosis. We in this case have outlined current treatment options.

Case Presentation: This fifty year old hypertensive male patient was evaluated first at the outpatient department, where he presented with the chief complaints of bleeding per rectum associated with tenesmus while passing stools along with a complaint of a mass protruding from the rectum. On a digital per rectal examination, the patient had a black pigmented mass protruding at the anal verge, which was tender and bleed to touch. Following this, a colonoscopic evaluation revealed a lesion at the anal region with circumferentially thickened distal rectal area. A biopsy was retrieved from this mass which revealed Malignant melanoma. A PET CT was also done, which revealed a hypermetabolic circumferential wall thickening involving the anal canal and distal rectum with metastatic pelvic nodes, and hepatic metastasis. The evaluation of other primary sites like the eyes and skin were ruled out. After basic surgical and general evaluation of the patient, he was then taken up for a Laproscopic Abdomino perineal resection and total mesorectal excision with an end colostomy. A bilateral inguinal lymphadenectomy was performed, which helped create a larger negative margin. The patient was shifted to the Critical care unit, for further management with IV antibiotics, analgesics and other supportive measures. The surgery was uneventful and patient recovered well. He was mobilized and started on sips on post operative day 1, which he tolerated well. He was soon started on oral feeds and had a well functioning colostomy, and was discharged from the hospital to follow up with the oncologist for further management. The final histopathology report with immuno-histo chemistry revealed Malignant melanoma Anorectum located below the peritoneal reflection, the tumour infiltrates through the muscularis propria, proximal and distal margins are free of tumour, circumferential resection margin is involved by the tumour, lymphovascular invasion present, 4/5 lymphnode are involved with perinodal spread, perineural invasion present. Pathological staging –pT3N2AM. A macroscopic image of the specimen is shown in Figure 1.

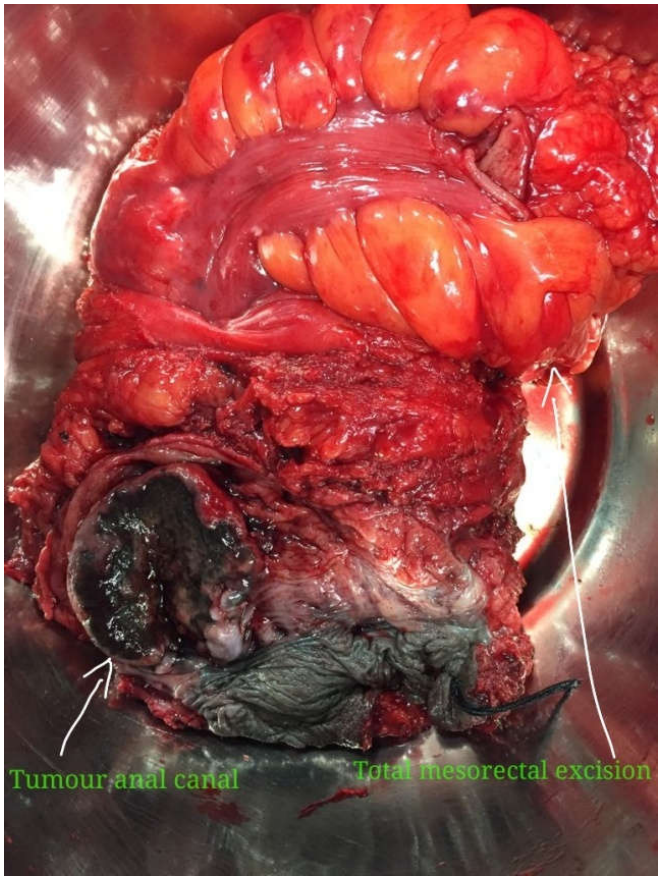

Figure 1.Macroscopic image of anorectal melanoma

DISCUSSION

Primary malignant melanoma of the anus and rectum is a rare and highly lethal malignancy of the elderly, which often manifests at an advanced stage (Chute *et al*., 2006). Mucosal melanomas represent approximately 1.2% of all melanomas and anorectal melanomas account for less than 25% of all mucosal melanomas (Chang *et al*., 1998). It is the third most common location after cutaneous and ocular melanomas (Singer, 2006). In addition, it is the most common primary melanoma of the gastrointestinal tract (Cheung *et al*., 2008) and accounts for approximately 0.5% of all colorectal and anal cancers (Roumen *et al*., 1996). The tumor commonly affects females in their fifth or sixth decade (McLaughlin *et al*., 2005; Coté, 2009) with a 1.7-fold higher prevalence in Caucasians than in African Americans (Coté, 2009). The incidence rate is reported as 0.4% per million (McLaughlin *et al*., 2005) with a 1.8-fold increase in incidence in the last 2 decades, suggesting either a true increase in incidence or an improvement in diagnosis (Coté, 2009). A melanoma of the anus and rectum was first reported by Moore (Moore, 1857) in 1857. Lesions can affect the anal canal, the rectum or both, with the majority occurring within 6 cm of the anal rim (Zhang *et al*., 2010). Common presenting symptoms include rectal bleeding, anorectal pain or discomfort, change in bowel habits, prolapsed tumor mass and hemorrhoids. This represents a significant clinical challenge since early diagnosis and treatment are crucial. Our patient had a similar presentation. Primary anorectal malignant melanomas are in almost 80% of the times mostly misdiagnosed as hemorrhoids, polyp, adenocarcinoma or rectal ulcer (Zhang *et al*., 2010). Grossly, the majority of lesions appears polypoid, with or without pigmentation, and can be ulcerated as well (Chute *et al*., 2006). The tumor is amelanotic in about 30% of the cases (Hillenbrand *et al*., 2008) and with considerable morphologic variability, misdiagnosis as lymphoma, carcinoma or sarcoma is common (Banerjee, 2006; Gatter *et al*., 1985). The use of immunohistochemistry panels, including S-100 proteins, MelanA, HMB-45 and tyrosinase, can help in the diagnosis (Chute *et al*., 2006). In our case, immunohistochemistry analysis was positive for the S-100 protein. Chute *et al*. (5)reported 4 histologic cell types: epithelioid, spindle cell, lymphoma-like and pleomorphic. The mitotic rate averaged 2.8 mitotic figures per high-power field in 17 cases of a primary anorectal malignant melanoma. It is presumed that primary anorectal malignant melanoma arises from normal melanocytes in the intestinal epithelium distal to the dentate line and extending proximally into the rectum (Morson, 1963). KIT expression can be present in anorectal malignant melanomas and, when present in spindle cell subtypes, can lead to confusion with gastrointestinal stromal tumors (Chute *et al*., 2006). As in cutaneous melanomas, loss of c-kit expression is associated with aggressive clinical behavior, it was postulated that a loss of KIT might play a role in the pathogenesis (Chute *et al*., 2006; Ni *et al*., 2012), therefore suggesting a role of kinase inhibitors such as imatinib (Ni *et al*., 2012). The 5-year survival rate has been reported to be less than 20% for anorectal melanomas, with a median survival of 24 months (Chang *et al*., 1998). Prognostic factors include the stage of the disease at the time of diagnosis (Slingluff *et al*., 1992) and the tumor thickness (Ballo *et al*., 2002). Common sites of distant metastasis are the liver and lung (DeMatos *et al*., 1998). As this is a relatively rare entity, we lack randomized control trials regarding appropriate management, and current evidence is mostly based on retrospective studies, reported as a limited number of cases or data collected over prolonged time periods, including patients with an age of up to 64 years (Brady *et al*., 1995; Weyandt *et al*., 2003). Optimal treatment is still controversial. Surgical approaches include WLE and APR.

Our patient underwent APR. A meta-analysis of 426 patients did not demonstrate any survival advantage with either approach (Thibault *et al*., 1997). Preoperative tumor thickness may be a valuable tool to plan the surgical approach (Ross *et al*., 1990). Studies reported that local disease seems to be more effectively controlled with APR (Brady *et al*., 1995; Zhou *et al*., 2010; Iddings *et al*., 2010). Recent studies suggested the initial treatment of choice to be WLE because radical surgery failed to show any survival advantage and also to avoid the need for colostomy (Zhou *et al*., 2010; Iddings *et al*., 2010). Local recurrences are common with WLE and with no documented effect on survival (Zhou *et al*., 2010). Radiation therapy has reported to provide a better local control after WLE (Chute *et al*., 2006) and also seems beneficial for sphincter preservation (Brady *et al*., 1995). Most patients die regardless of the chosen therapeutic strategy due to the aggressive nature and the rapid progression of the tumor. Kim *et al*. (2004) conducted a retrospective review on 18 patients with metastatic anorectal melanoma treated with cisplatin-based chemotherapy in combination with interferon alpha-2b or interleukin-2. They reported that combination chemotherapy was effective against metastatic anorectal melanoma. The response was similar to that of cutaneous melanoma.

Conclusion

Anal Malignant melanoma is a rare malignancy that has a tendency to be misevaluated and misdiagnosed for other

benign diseases. The diagnosis usually portends a poor prognosis, with high morbidity and mortality rates, and distant metastases being a very common and grave development. While neither surgery nor radiation has had a clear impact on survival, this surviving case had experienced good results postoperatively.

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