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Case Report

Pathology Section

Primary Rectal Malignant Melanoma: A Case Report

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ABSTRACT

Primary Rectal Malignant Melanoma (RMM) is an uncommon and highly aggressive tumour that affects older individuals. Individuals with RMM frequently exhibit regional and distant tumour spread at the time of diagnosis and hence poor long-term survival. Complaints like bleeding per rectum often lead to incorrect diagnosis such as haemorrhoids. Hereby, the authors present a case of a 56-year-old male with primary RMM which was clinically suspected to be rectal carcinoma. On abdomino pelvic Contrast-enhanced Computed Tomography (CECT) a well-defined hypodense enhancing mass involving proximal and distal rectum was visualised along with multiple peri-rectal lymph nodes. Fluorodeoxyglucose (FDG) Positron Emission Tomography (PET) scan showed FDG uptake in regional lymph nodes suggesting metastasis. Colonoscopic biopsy of the mass exhibited diverse histomorphology of the cells comprising of round, oval and spindly cells along with tumour giant cells. The patient succumbed to the disease one year after diagnosis, despite receiving post-operative adjuvant chemotherapy. In present case, the patient presented with a rare case of rectal melanoma, exhibiting varied histomorphology and significant metastases at the time of diagnosis, leading to a poor long-term survival. Diagnostic work-up in elderly patients with rectal bleeding must include CECT to rule out neoplastic lesions, colonoscopic biopsy in case of mass lesion. Histopathology remains gold standard in diagnosis of the lesion, and Immunohistochemistry (IHC) can be advised when required. PET scan is crucial in identifying metastatic extent of the tumour. Thus, prognosis depends solely on early diagnosis by colonoscopic biopsy with IHC study to confirm the diagnosis of melanomas of anorectal region.

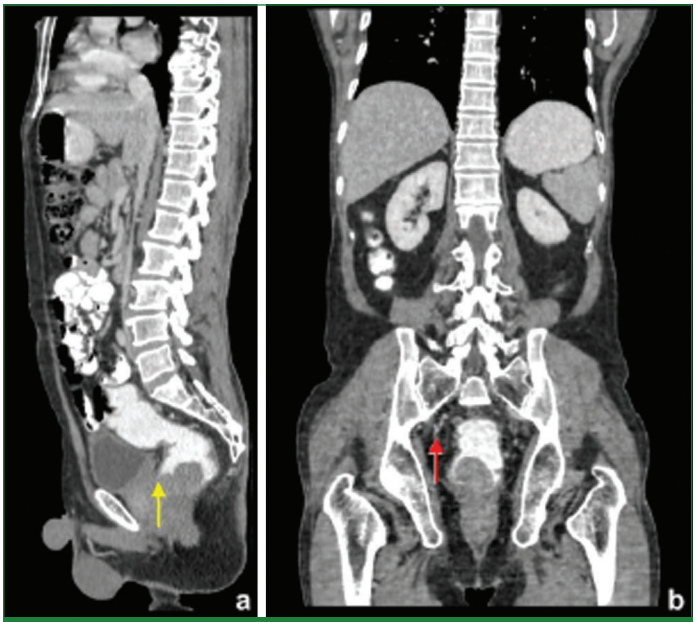
Keywords: Melanoma antigen recognised by T-cells 1 (MART-1), Metastasis, Positron emission tomography scan (PET scan), Rectal tumour, S100 protein

CASE REPORT

A 56-year-old male visited Surgery Outpatient Department (OPD) complaining of per rectal bleeding (PR) for three months which was intermittently associated with hard stools. Recently he suffered from constipation since three days for which he sought medical help. There was no history of vomiting, or abdominal pain or anal pain. On digital rectal examination, a firm to hard growth was palpated 3 cm from the anal verge measuring approximately 1.5-2 cm, with irregular surface and that did not bleed on touch. General and other system examinations were normal. He had no significant past medical history. Possible clinical diagnosis considered was rectal carcinoma.

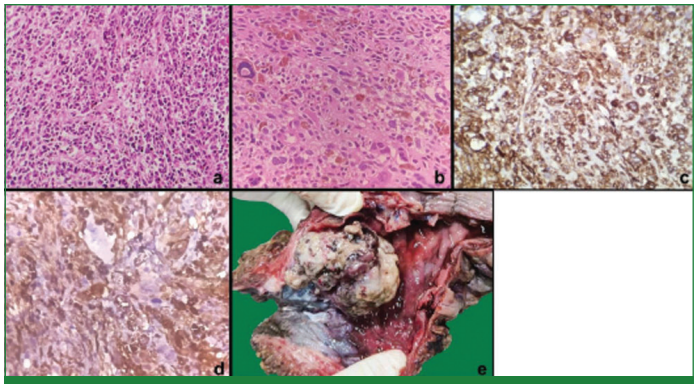
Abdomino-pelvic CECT was performed which revealed a well-defined lobulated hypodense enhancing intraluminal soft-tissue density lesion involving proximal and distal rectum, measuring 38×31 mm [Table/Fig-1a]. Multiple enlarged, sub-centimeter sized necrotic lymph nodes were seen in the peri-rectal region, largest measuring 10×11 mm suggestive of neoplastic aetiology [Table/Fig-1b]. FDG PET scan revealed an FDG avid mass in the lower rectum, measuring 52×42 mm. It also showed a contiguous uptake in the upper anal canal, right inguinal nodes, right external iliac, left obturator, mesorectal and right iliac lymph nodes. No uptake was seen in any distant organ. Suggestive of malignant rectal tumour with regional lymphnode metastases.

Colonoscopic biopsy of the lesion showed normal stratified squamous epithelium with underlying tumour arranged in sheets [Table/Fig-2a]. The tumour cells were round, oval, and spindle in morphology with pleomorphic, vesicular to hyperchromatic nuclei, moderate amount of eosinophilic cytoplasm, few abnormal mitotic figures along with bizarre and tumour giant cells. Some cells contained a small amount of intra cytoplasmic brown pigment [Table/Fig-2b], and few had prominent nucleoli. Possible differential diagnoses which were considered were poorly differentiated carcinoma, sarcoma and melanoma. The tumour cells were strong and diffusely positive for S100 [Table/Fig-2c] and Melan-A [Table/Fig-2d] immunohistochemical markers therefore, a diagnosis of malignant melanoma was offered.



[Table/Fig-1]: a) Sagittal CECT showing rectal mass (yellow arrow); b) Coronal CECT showing peri-rectal lymph nodes (red arrow).

The patient underwent laparoscopic Abdomino-perineal Resection (APR) with end colostomy. Resected specimen sent for Histopathological Examination (HPE) measured 17 cm in length with an unremarkable external surface. On cut-surface a firm polypoidal mass in the rectum measuring 5×4×2.5 cm, with a solid blackish-brown appearance, was seen abutting to the dentate line and reaching the anorectal junction [Table/Fig-2e]. A total of 15 peri-rectal lymph nodes were isolated, with the largest one measuring



[Table/Fig-2]: a) Tumour cells in sheets (H&E, 400X); b) Cells with prominent nucleoli, intracytoplasmic melanin pigment and bizarre cells (H&E, 400X); c) IHC image of S100 (400X); (d) IHC image of Melan A (400X); e) Gross image showing polypoidal growth with black pigment.

1.5 cm in diameter. Microscopic examination on Haematoxylin and Eosin stained sections (H&E) revealed histomorphological features consistent with those observed in the biopsy specimen. The tumour cells were round, oval, and spindle in morphology with pleomorphic, vesicular to hyperchromatic nuclei, prominent nucleoli, few abnormal mitotic figures, within intracytoplasmic brownish pigment. The tumour demonstrated invasion into the muscularis propria, while the serosa was free of tumour. Among the15 peri-rectal lymph nodes examined, two exhibited metastatic tumor involvement. All resection margins including radial margin were free of tumour. Final diagnosis of malignant melanoma of rectum was offered. Follow up abdomino pelvic CECT scan after two months did not reveal any new lesions. The patient was given postoperative adjuvant chemotherapy at another hospital, details of which are unavailable. Despite the treatment efforts, patient passed away after one year of diagnosis.

DISCUSSION

Mucosal melanoma constitutes about 1.3% of all reported melanomas, of which 23.8% are seen in anorectal region [1]. The annual incidence of anorectal melanoma was estimated to be 0.343 per 1 million in a population based study conducted by Chen H et al., [2]. RMM is seen predominantly in elderly women in their fifth to sixth decades of life and show increased incidence with advancing age however, the present case was seen in an elderly male [1-4]. RMM is highly aggressive tumour with poor long-term survival. Among RMM cases approximately 41% presented with regional lymph node metastasis at the time of diagnosis and 22% cases presented with distant metastases commonly involving the brain, liver, lungs, and bone [4]. Five year survival rate of the patients was found to be as low as 12% according to Wanebo HJ et al., study and 12-26% in study by Weinstock MA, similar to present case where patient passed away after one year of diagnosis [3,4]. In present case PET scan was instrumental in identifying regional lymphnode metastases and staging the disease.

Although melanomas are typically pigmented but, they are often diagnosed late. A pigmented anorectal lesion presenting with PR bleed is often mistaken for haemorrhoids [2,5]. Studies have found that RMM frequently presents as a polypoid mass, which can lead to misdiagnosis as benign polyps or carcinomas similar to present case [3,5]. Unlike rectal carcinomas, which are often infiltrative and cause bowel obstruction, rectal melanomas, despite their polypoid morphology, does not result in obstruction [6].

Pathogenesis of rectal melanoma is not well established unlike the more common Cutaneous Melanoma (CM). Molecular analysis in anorectal melanoma cases identified KIT mutations in 33% of cases [7]. These cases also harboured distinct mutation in Neurofibromatosis Type 1 (NF 1) and Mitogen-activated Protein Kinase (MAPK) pathway effectors in contrast to CM. These differences in mutational profiles and clinical presentation underscore the distinct nature of RMM relative to the more common CM.

The RMM can exhibit diverse histomorphological features, such as pleomorphic, spindle-shaped, small lymphocyte-like, and epithelioid cells [3,8,9]. Therefore, immunohistopathology is considered as the gold standard test for diagnosing melanomas. Spindle or epithelioid and small melanoma cells mimic sarcoma, or lymphoma. Trabecular or focal organoid pattern may resemble carcinoid. Melanoma cells may be arranged in sheets resembling poorly differentiated adeno carcinoma or squamous cell carcinoma [8]. Presence of melanin pigment helps in narrowing down the differentials and conserve efforts on performing additional IHC markers. Therefore, anorectal lesion presenting as a polypoidal mass with varied histomorphology; poorly differentiated carcinoma, carcinoid, sarcoma, lymphoma and melanoma a must be considered among the differentials. IHC like Human Melanoma Black (HMB)-45, S100 and Melan-A/MART-1 exhibit strong positivity in melanomas and helps in ruling out the differentials, similar to the present case [8,9].

The optimal management of RMM is not clearly defined in previous studies. Management comprises a multimodal approach that incorporates immunotherapy, chemotherapy, with targeted treatment, and/or surgical removal [10]. Surgery is usually the primary mode of treatment adopted by most surgeons which comprises of APR or wide local excision, but neither of them provide superior survival benefits [2,10]. Chemotherapeutic drugs like cisplatin, vincristine, dacarbazine and immunotherapeutic agents like interferon alpha have been used as adjuvant agents in treatment of anorectal melanomas [10]. A study conducted by Vitagliano NL et al., have demonstrated effectiveness of neoadjuvant immunotherapy followed by surgery with maintenance immunotherapy leading to complete remission [11]. Immunotherapeutic agents like anti Cytotoxic T Lymphocyte-associated protein 4 (CTLA-4) agent ipilimumab, and Programmed cell death-1(PD-1) inhibitor nivolumab were utilised in Vitagliano NL et al., study [11]. Increased Overall Survival (OS) with immunotherapy in comparison to chemotherapy is still debatable. Ongoing trials focus on targeted therapies aimed at Cellular homolog of the feline sarcoma viral oncogene v-kit (C-KIT) mutation.

A three-stage clinical staging for RMM: Stage I (local disease), stage II (local disease with regional lymphnode involvement), and stage III (with distant metastasis) has been used by few studies in literature [1,3,10]. Studies have shown that the patients with local disease had better prognosis than the patients with regional or distant metastases [2,4]. However, there remains no formal staging system for this tumour till date, unlike for CMs [1]. Wanebo HJ et al., study showed a correlation between OS and tumour size. Patients with tumour measuring ≤2 mm exhibited higher OS [3]. In present case, tumour size was larger than 2 cm and patient did not survive more than one year. RMM typically remains undetected and presents at sizes exceeding 2 cm similar to the present case. Since a significant number of individuals with RMM already exhibit metastases at diagnosis like the present case, early detection is the only way to enhance patient survival rates. Authors reported the present unusual case of RMM for creating awareness thereby early diagnosis and better prognosis.

CONCLUSION(S)

The rarity, aggressive nature and differences in behaviour from CM, makes diagnosis of RMM more challenging. When an anorectal lesion appears as a polypoid mass with varying histomorphology, melanoma should always be considered among the potential differential diagnoses. Histopathology and IHC study are crucial for diagnosing RMM. While surgery remains the mainstay of treatment, the optimal therapeutic approach is still debatable. Newer adjuvant immunotherapies have shown effectiveness in certain cases, and targeted therapies are currently undergoing trials. Since a significant number of individuals with RMM already exhibit extensive metastases at diagnosis, early detection by biopsy is the only way to enhance patient survival rates.

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