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Rectal Melanoma: Rare Disease Managed in District Hospital

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Authors' contributions

This work was carried out in collaboration among all authors. Author RSK designed the study, performed the statistical analysis, wrote the protocol, and wrote the first draft of the manuscript. Authors SS and SE managed the analyses of the study. Author HA managed the literature searches. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Rectal melanoma is an extremely rare disease which can be aggressive invading locally and metastasize during the cause of disease. Hereby, we are reporting a case of a lady presented to us with anal mass sensation since 2-month duration. Per rectal examination revealed mass about 3cm from anal verge with a melanotic skin lesion at perianal region. Colonoscopy done revealed anorectal mass 2cm from anal verge and biopsied which revealed malignant melanoma and subsequently we proceeded with abdominal perineal resection (APR). Histopathology examination report as malignant melanoma.

Keywords: Rectal; malignant melanoma; abdominal perineal resection (APR).

1. INTRODUCTION

Rectal melanoma was first reported by Moore in 1857 [1]. Is an extremely rare and aggressive malignancy [2]. Rectal melanoma constitutes around 0.5-4% of overall anorectal malignancy and less than 1% of all melanoma [3,4]. Predominantly affects female gender in fifth or

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sixth decade of life. Presenting complain in this group of malignancy is vague which includes rectal bleeding, anorectal pain, altered bowel habit or rectal mass, hence leading to diagnosis at early stages difficult [5]. Other causes of delay in early diagnosis at early stages are up to 80% of this lesion lack pigmentation and the rest 20% are amelanotic [6, 7].

2. CASE REPORT

58 years old lady presented with anal mass associated with per rectal bleed since 2 months duration to department of General Surgery, Sultan Haji Ahmad Shah Hospital, Temerloh, Pahang, Malaysia. Per abdomen revealed no significant finding. Per rectal examination evidence of mass 3cm from anal verge, mobile, able to get beyond and presence of melanotic skin lesion over perianal region and proctoscopy examination revealed a fungating anorectal mass at 7 o'clock. (Fig.1). Colonoscopy showed an anorectal mass 3cm from anal verge, able to pass scope beyond and presence of pedunculated polyp at ascending colon. Both lesion biopsied and reported as malignant melanoma for anorectal mass and tubovillous adenoma with low grade dysplasia for ascending colon polyp. Carcinoembryonic antigen (CEA) was 1.6. Computed tomography of thorax, abdomen and pelvis showed anorectal lesion with possible lymph node metastasis. She underwent abdominal perineal resection (APR) and intra-operative finding was fungating rectal mass at anterior wall of rectum measuring 4 x 4cm, 3cm from anal verge, multiple matted pre-sacral lymph nodes, the rest organ were normal (Fig. 2). Histopathology examination revealed malignant melanoma with lymph node involvement, following was the staining performed by our Pathologist which were Melan A and HMB 45 (Figs. 3,4 and 5). Patient was discharge home well. Patient was referred to Oncology team and planned for adjuvant immunotherapy, pembrolizumab. Patient is under the follow up at Oncology centre and once has completed her treatment will be follow up at our centre for surveillance. In my centre, this is first reported case of such pathology.

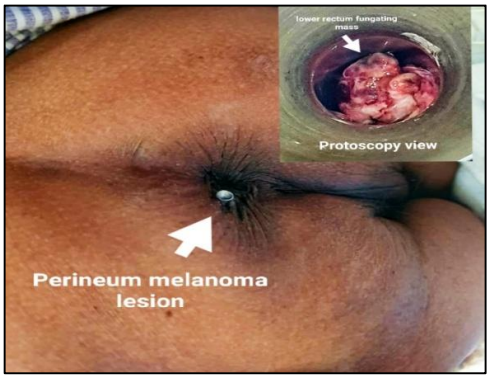

Fig. 1. Showing an anorectal mass (3cm from anal verge), with melanotic skin lesion over perianal region. Proctoscopy examination revealed a fungating anorectal mass at 7 o'clock

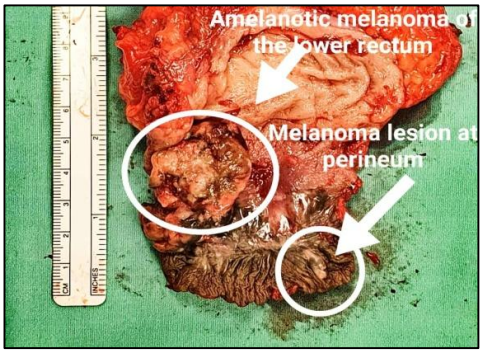

Fig. 2. Showed the resected specimen showed a fungating rectal mass at anterior wall of rectum measuring 4 x 4 cm, 3 cm from anal verge, with melanoma lesion at perineum

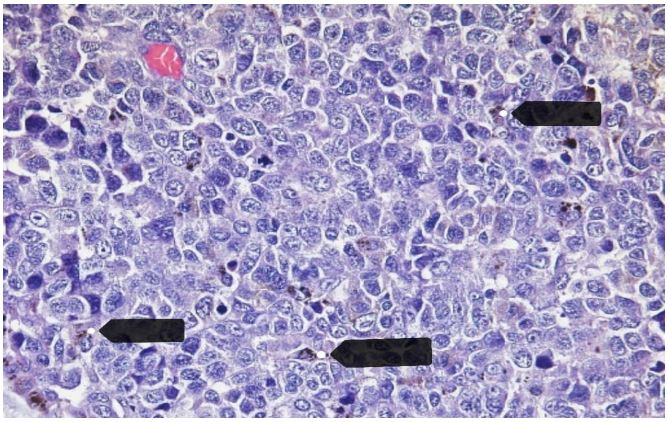

Fig. 3. The tumour cells exhibit enlarged vesicular nuclei with moderate nuclear pleomorphism, prominent nucleoli and moderate cytoplasm. Intracytoplasmic pigments (arrow) are also seen (H&E, x400)

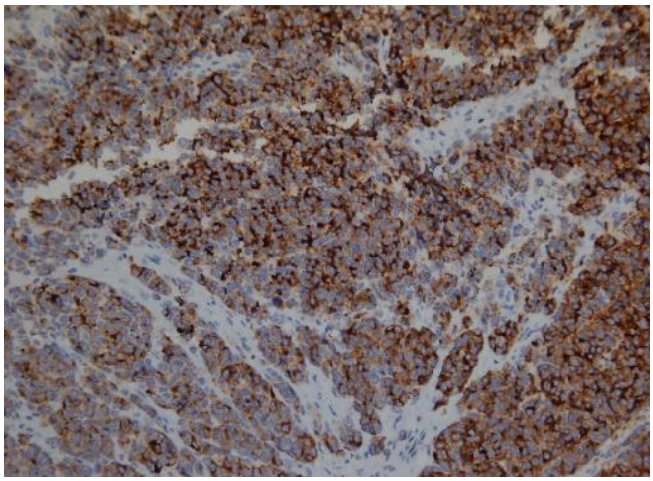

Fig. 4. The tumour cells are immunoreactive with Melan A (Melan A, x100)

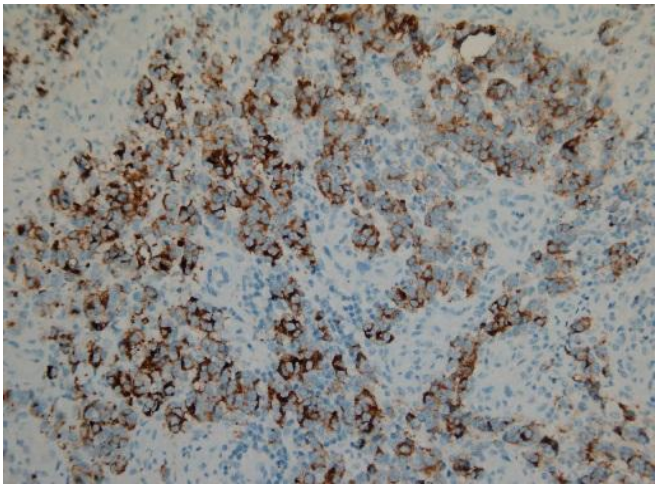

Fig. 5. The tumour cells are immunoreactive with HMB45 (HMB45, x100)

3. DISCUSSION

Rectal melanoma is most common primary melanoma of gastrointestinal tract [9]. It accounts around 0.5% of all colorectal and anal cancers [10]. Is unlikely to be caused by exposure to ultraviolet light like in other cases of melanoma [11], its incidence is higher in immunocompromised patients [12]. Melanoma originates from melanocytes which are derived from embryological neural crest. Melanoma can be found in various location of body, primarily cutaneous followed by ocular and finally gastrointestinal. In rectum, melanocytes are located at the anal transition zone and squamous zone. Majority occurring within 6 cm of the anal canal [13]. Classification of rectal melanoma is according to American Joint Commission on cancer staging system (AJCC) and according to Breslow thickness of lesion (Table 1 and Table 2). Histological variant of rectal melanoma according to Chute et al. are epitheloid, spindle cell, lymphoma like and pleomorphic [14].

Common presenting complaints can be presence of anal mass as in this patient, other complaints are bleeding, anorectal pain, rectal tenesmus or discomfort or change in bowel habit. In cases of metastasis it can be presented as constitutional symptoms such as fatigue, weight loss and anaemia. Rectal melanoma almost 80% are misdiagnosed as haemorrhoids, polyps, adenocarcinoma or rectal ulcer. Modalities available to aid in obtaining diagnosis of rectal melanoma as below (Table 3)

Histopathology finding can be helpful, the presence of melanin can be helpful although it is not easily detected [11]. Other immunohistochemistry markers are melanoma antigens S-100, HMB 45 and vimentin. Polyclonal anti-serum and monoclonal antibodies to carcinoembryonic antigens are helpful in distinguishing rectal melanoma from poorly differentiated epidermoid carcinoma. Activating KIT gene mutation has also been associated in this disease. Surgery remain the mainstay of treatment. As this disease is very rare there are lack trials regarding its management, options of management recommended are based on retrospective studies with limitation in number of cases and collection of data were over period of time leading to dilemma in optimal treatment for this disease. Surgery consist of abdominal perineal resection (APR) or wide local excision (WLE). Previously APR been the modality choice of surgery however recent studies have showed WLE are adequate for local control and minimise the morbidity of surgery. There is no difference in outcome between these 2 modalities [2,3,4,11]. Other benefits are the need of stoma not required and minimal effects on bowel function [11]. Sentinel lymph node dissection is useful in apparent disease [3,4]. Patient diagnosed with rectal melanoma, 5 year survival rate range from 16-34% and in advanced disease 5 year survival rate drop to 16% [4,11]. Prognostic factor includes stage of disease at time of diagnosis [15] and tumour thickness [16]. In terms management with chemotherapy, radiotherapy

Table 1. Classification of rectal melanoma

AJCC stage	TNM stage	Definition
Stage I	T1N0M0	Tumour invades submucosa
	T2N0M0	Tumour invades muscularis propria
Stage IIa	T3N0M0	Tumour invades through muscularis propria into subserosa or non-peritonealised pericolic tissues
Stage IIb	T4N0M0	Tumour directly invades other organs or structure and/or perforates visceral peritoneum
Stage IIIa	T1-2, N1M0	Metastasize to 1-3 regional lymph nodes
Stage IIIb	T3-4, N1M0	Metastasize to 1-3 regional lymph nodes
Stage IIIc	Any T, N2M0	Metastasize > 4 regional lymph nodes
Stage IV	Any T, any N, M1	Distant metastasis

Table 2. Rectal melanoma stages with depth

Stage	Depth (mm)
Stage I	0.75-1.5
Stage IIa	1.5-4.0
Stage IIb	>4.0
Stage III	X
Stage IV	X

Table 3. Modalities available to aid in obtaining diagnosis of rectal melanoma [11]

Colonoscopy	For evaluation of cause of symptoms and obtaining tissue biopsy from lesion if present
Endoscopic endorectal ultrasound	Evaluate tumour thickness and nodal status
Contrast enhanced CT	Assess regional disease, determine presence of lymphadenopathy or metastasis. On CT it appears as intraluminal fungating mass expanding and obscuring with no obstruction and perirectal infiltration.
MRI	High signal intensity on T1-weighted images and mixed signal intensity on T2-weighted imaging

and immune therapy there show no added benefit in term of overall survival. Dacarbazine has shown a partial response in 20% of patients within 4-6 months after treatment [3,11]. A combination chemotherapy was reported to be effective by Kim et al. [17] a study done on 18 patients with cisplatin combining with interferon alpha-2b or interleukin-2.

4. CONCLUSION

Rectal melanoma is extremely rare, aggressive and difficult to diagnose. Early diagnosis and treatment may improve survival rate. To be able to advocate an early diagnosis and treatment remain crucial. APR may offer a higher rate of local control and can be carried out safely in district hospital setting. However, wide local excision offers a much less morbid operation but need to be carried out tertiary centre with colorectal subspecialty.

CONSENT

As per international standard or university standard, patient's consent has been collected and preserved by the authors.

ETHICAL APPROVAL

It is not applicable

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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