Case Report

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ANO-Rectal Primary Malignant Melanoma; CT Versus MR Imaging Features

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Abstract

Anorectal malignant melanoma (AMM) is a rare and highly aggressive malignancy, shares similar appearance and clinical symptoms with low rectal cancer, presenting as an intraluminal polypoid mass accompanying a series of clinical symptoms, such as rectal bleeding, tenesmus and a change in bowel habits. In clinical practice, Computed tomography (CT) & magnetic resonance imaging (MRI) are the most commonly used modalities for evaluating and diagnosing anorectal lesions. The overlap in clinical symptoms and imaging findings make it easy to misdiagnose anorectal malignant melanoma as low rectal cancer, the most common malignancy in the anorectum. Currently, endoscopic biopsy is the routine procedure for pre-operative diagnosis of anorectal malignant melanoma. We present the clinical, CT and typical MR features seen in a 53-year-old woman diagnosed with melanoma of the anorectum.

Keywords: Anorectal Malignant Melanoma, Low Rectal Cancer; Metastatic Malignant Melanoma, Primary Malignant Melanoma.

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Introduction

Primary anorectal melanoma is rare, comprising less than 1% of all melanomas, 0.1% of all rectal malignancies, and 4% of anal malignancies.[1] It tends to spread submucosally and is often beyond complete resection at the time of diagnosis,[2] and majority of patients die of metastasis. Its prognosis is worse, with a 5-year survival rate of only 10–17%.[3,4] Patients, especially women, usually present with local symptoms in the fifth or sixth decade of life.[5] Because its clinical symptoms and signs are unexplained and include rectal bleeding, tenesmus, an anorectal lump, and a change in bowel habits,[3] which can be caused by more common benign entities, such as hemorrhoids or polyps, a timely diagnosis is sometimes difficult. In addition, although it may be suspected that tumour status, the separation of primary anorectal melanoma into more common malignancies, such as rectal and anal carcinomas, is less accurate due to the lack of reported imaging findings of primary anorectal melanoma.

Currently, pathologic confirmation of immuno-histochemical staining is a prerequisite for diagnosis of primary anorectal melanoma.[6,7]

MRI is widely used as the diagnostic tool for a variety of abnormalities; its soft-tissue resolution and tissue characterization are superior to those of CT. Determination of the reliable MRI features of primary anorectal melanoma should contribute to this challenging diagnosis and provide the potential to increase the rate of identification of primary anorectal melanoma in its earlier clinical stages.

The aim of the present study was to evaluate the CT & MRI features of malignant melanoma in a 53-year-old woman, primarily involving the anorectum and to link these features to its clinical and pathologic features.

Case Report

Female aged 53 years presented with history of bleeding and pain during defecation for 1 month. She was a non smoker with an unremarkable medical history. Her clinical examination was otherwise essentially normal. Routine blood and urine examination were unremarkable.

Per rectal examination disclosed a hard mass at 6 'o'clock position, less than 1 centimeter from anal verge. The mass was bleeding on touching. Provisionally diagnosed as Rectal carcinoma.

Biopsy was taken from the mass and sent for Histopathological examination. Hematoxylin and eosin staining of the sample revealed pleomorphic lesions displaying high mitotic rate and prominent nucleoli. It showed intra and extracellular melanin pigment on microscopic examination. Immunohistochemical staining with markers HMB 45 (Human Melanoma Black) and S 100 (S100 protein that is soluble in 100% ammonium sulfate), was suggestive of malignant melanoma.

CT abdomen [Figure 1&2] done showed an Irregular heterogeneously enhancing mass lesion seen occupying the anorectal junction extending to the right lateral wall of distal rectum. The lesion is seen extending transmurally to involve the internal and external sphincter. The lesion measures ~3.3x3.8x6.5cm (AP x Trans x Craniocaudal).

Contrast enhanced CT chest did not reveal any primary or secondary lesions in the chest.

MRI of the pelvis [Figure 3-9] done reveal an Irregular mass lesion seen occupying the anorectal junction extending to right lateral wall of distal rectum which appears hyperintense on T1, STIR and mixed signal intensity on T2W sequences. The lesion is seen extending transmurally to involve the internal and external sphincter (T3a N1 M0). Perirectal fat plane appears infiltrated. The lesion measures 3.3x3.8x6.5cm (AP x Trans x Craniocaudal). Sparing of mesorectal fascia and mesorectum noted. No involvement of vagina. Few subcentimetric perirectal lymph nodes noted.

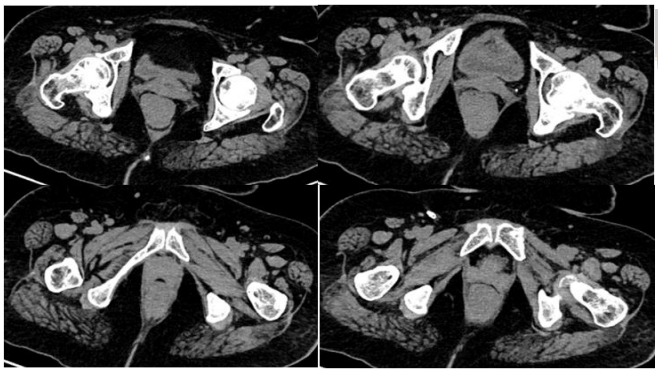

Figure 1: (A-D) Noncontrast CT Axial images reveal an irregular isodense mass in the anorectum partially occluding the lumen.

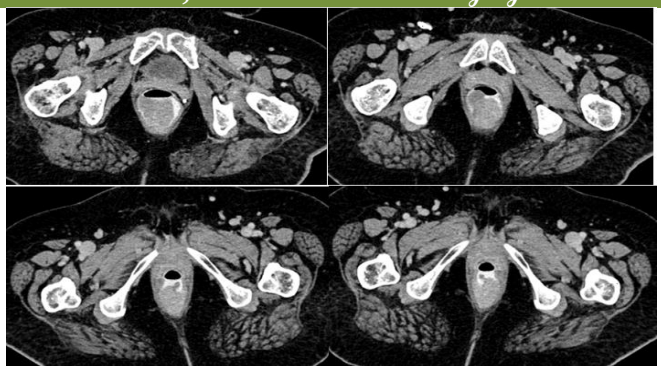

Figure 2: (A-D) Contrast enhanced CT Axial images reveal an irregular isodense heterogeneously enhancing mass lesion occupying the anorectal junction extending to right lateral wall of distal rectum.

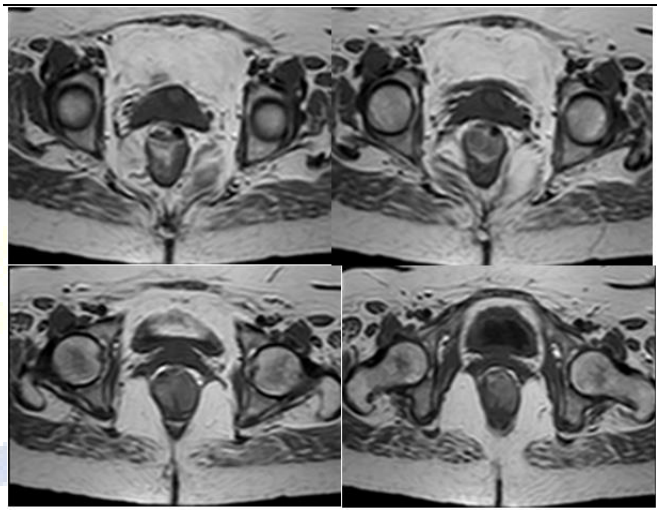

Figure 3: (A-D) Axial T1W images show an irregular mild hyperintense mass lesion occupying the anorectal junction extending to right lateral wall of distal rectum.

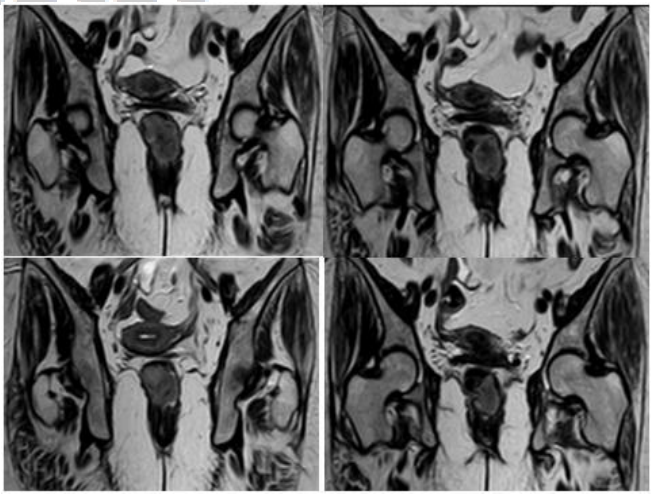

Figure 4: (A-D) Coronal T2W images show an irregular mixed signal intensity mass lesion occupying the anorectal junction extending to right lateral wall of distal rectum.

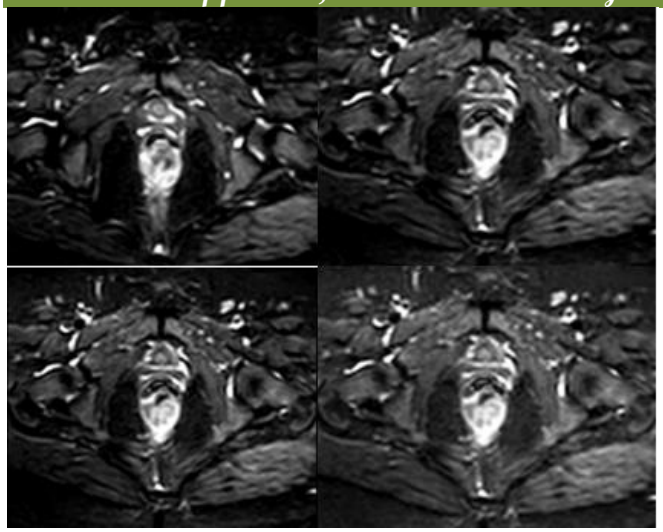

Figure 5: (A-D) Axial STIR images show an irregular hyperintense mass lesion occupying the anorectal junction extending to right lateral wall of distal rectum.

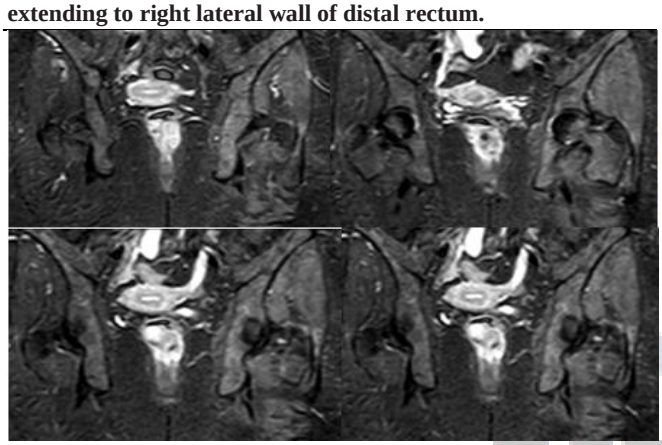

Figure 6: (A-D) Coronal STIR images show an irregular hyperintense mass lesion occupying the anorectal junction extending to right lateral wall of distal rectum.

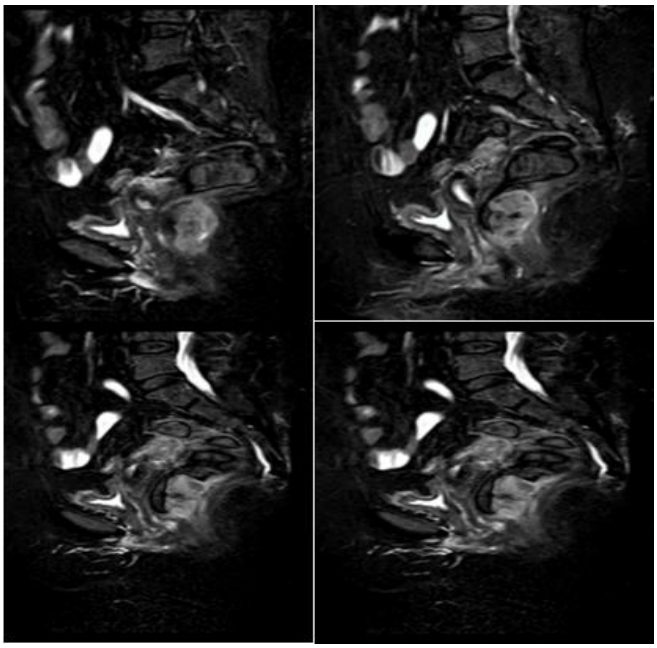

Figure 7: (A-D) Sagittal STIR images show an irregular hyperintense mass lesion occupying the anorectal junction extending to right lateral wall of distal rectum.

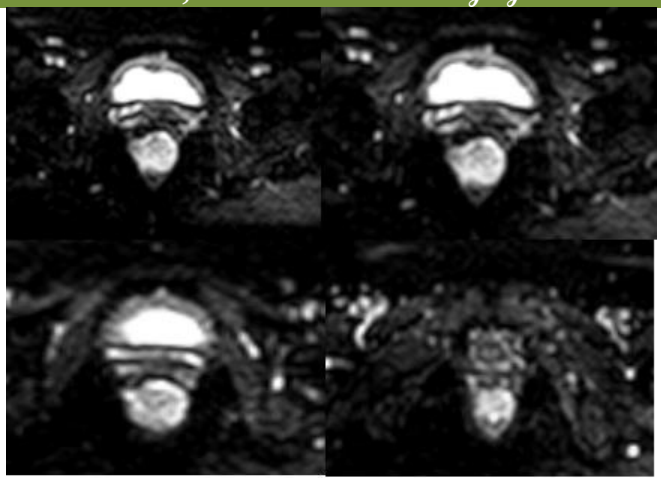

Figure 8: (A-D) Axial DWI images show an irregular mass lesion with restricted diffusion occupying the anorectal junction.

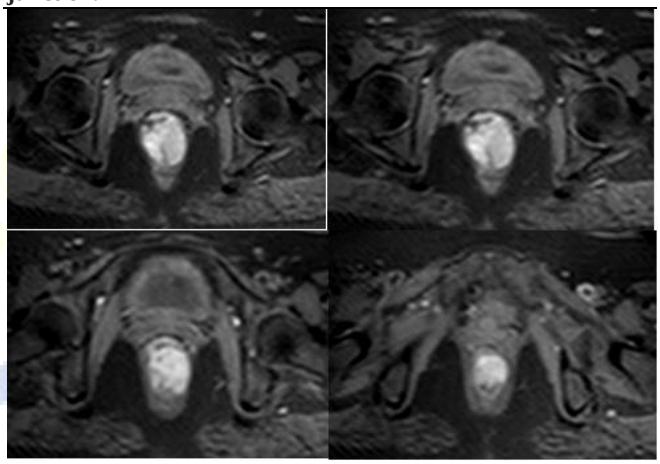

Figure 9: (A-D) Axial gadolinium contrast enhanced T1W fat satured images show an irregular mass lesion with moderate contrast enhancement occupying the anorectal junction.

Discussion

All melanoma, whether cutaneous or mucosal in origin, is derived from melanocytes, which are cells derived from embryological neural crest. During fetal development, these cells migrate to many areas throughout the body, especially to the skin. However, melanocytes also reside in the eyes (retina and uveal tract) and mucosal areas.[8,9] Therefore, cutaneous melanomas are the most common form of the disease, covering more than 90% of all melanoma. Of the remaining types of melanoma, ocular melanoma makes up 5%, melanoma of unknown origin of 2% and 1% of mucosal melanoma.[9]

In the rectum, melanocytes are found in the anal transition zone and squamous zone. Majority of anorectal melanomas arise from the dentate line and 65% are located within the anal canal or at the anal verge.[2,9] There is a potential role for immunology in the development of anorectal melanoma as the incidence is higher in patients with Human Papilloma virus (HPV) and HIV infection.[7,9]

Anorectal melanoma may appear as mucosal or submucosal mass. A rectal submucosal tumour is defined as a mass-like protrusion in the rectum covered by normal mucosa. It

comprises a variety of benign and malignant tumours of intramural or extramural origin. Those with an intramural origin include neuroendocrine tumour, gastrointestinal stromal tumour (GIST), schwannoma, melanoma, and metastatic spread to the rectum. Those with extramural origin include deep endometriosis, tailgut cyst and direct invasion by extra-colonic tumour.[10]

The incidence of AMM increases with age in both sexes and at all tumor sites. AMM lacks subjective symptoms in the early stage. The most common symptom is bleeding, with 53% to 89% of patients reporting this as major complaint. The other symptoms include altered bowel habits, constipation, decreased stool caliber, unintentional weight loss, and palpable inguinal mass. These lesions are often diagnosed as being benign hemorrhoids or polyps. In cases of diagnostic difficulty, which is frequent even on histopathology, the presence of melanin can be helpful, but it is not easily detected in anorectal disease.[2]

In most cases, AMM appears late as bulky intraluminal masses without causing obstruction and is associated with perirectal infiltration and nodal metastases. Unfortunately, few patients are diagnosed with AMM when they have already developed distant or regional metastasis.[11] The most common sites for metastases are inguinal lymph nodes, mesenteric lymph nodes, hypogastric lymph nodes, para aortic lymph nodes, liver, lung, skin, and brain.[8]

Anal melanoma is staged on a clinical basis, focusing on loco regional and distant spread [Table 1]. Stage I is local disease only, Stage II is a local disease with increased thickness and ulcerations, Stage III is local disease with involvement of regional lymph nodes, and Stage IV shows distant metastatic disease.

Table 1: American Joint Commission on cancer staging system for anorectal melanoma

Stage	Spread	Depth (mm)
IA	Localized	0.75
IB	Localized	0.76-1.5
IIA	Localized	1.5-4.0
IIB	Localized	>4.0
III	Regional nodes	X
IV	Distant metastasis	X

A sigmoido colonoscopy is essential both for evaluation of the cause of symptoms and obtaining a tissue biopsy from a suspicious lesion. Conventional colonoscopy can identify submucosal tumours but not their characteristics. Endoscopic endorectal ultrasound may be considered to evaluate tumour thickness and surrounding nodal condition.[2]

Transrectal ultrasonography can localise the specific mural layer of origin, but has limitations in the evaluation of large tumours, high rectal tumours, relationship of the tumour with the anal sphincter complex, and other pelvic organs. The diagnostic accuracy of transrectal ultrasonography–guided fine-needle aspiration / biopsy for rectal subepithelial tumours has been reported to be only 50%.[12]

Barium studies show multiple colonic submucosal nodules, intussusception, large ulcerative lesions, and extrinsic masses compressing the colon.[13]

On CT scans, primary rectal malignant melanomas appear as bulky intraluminal fungating masses in the distal rectum, focally expanding and obscuring the lumen without causing obstruction with perirectal infiltration and frequent lymph nodes enlargement.[14] Rectal carcinoma or other rectal masses present as significant obstruction. Contrast-enhanced CT scan and MRI allow characterization and assessment of the extent of the tumor.[14]

Magnetic resonance imaging (MRI) has a high soft-tissue contrast and multiplanar capability and enables discrimination of different layers of the rectal wall and evaluation of the origin and internal composition of rectal submucosal tumours. MRI is the investigative tool of choice for the complex anatomy of the pelvis and the relationship of tumours with the pelvic floor, sphincter complex, and adjacent organs to facilitate surgical planning.[14]

Melanoma cells may carry melanin pigment that shortens the T1 relaxation time due to the paramagnetic element. Melanoma also has a propensity to bleed. These account for the classic T1 hyperintensity in < 50% of melanomas. However, T1 hyperintensity is not expressed in melanomas with <10% melanin-containing cells, and up to one-third of anorectal melanomas are amelanocytic. On T2-weighted images, melanomas are usually hypointense but may contain mixed signal intensity. These are hypercellular and hypervascular tumours and hence restricted diffusion and contrast enhancement are typically present.[15,16]

Metastasis from a distant primary tumour to the rectum is extremely rare. It may occur by direct invasion of metastatic deposits in the pelvis such as by lymph nodes, serosal metastatic deposits in peritoneal carcinomatosis, or via haematogenous spread. MRI features of rectal metastasis are similar to those of primary tumors.[10]

Neuroendocrine tumours due to their origin in the superficial layers of the rectal wall, typically appear as a small superficial submucosal mass causing endoluminal bulging but without a significant exophytic component. The tumour is usually isointense on T1- weighted images and hyperintense on T2-weighted images, with moderate-to-intense contrast enhancement.[16,17,18]

GISTs are usually seen as an eccentric mural mass with well-circumscribed margins. Compared with muscle, GISTs on MRI show T1 hypointensity and mild T2 hyperintensity, with moderate heterogeneous gadolinium contrast enhancement. Mucosal ulceration may be seen.[20]

Schwannomas are usually well rounded submucosal tumour with T2 hyperintensity on MRI, representing the myxoid component. Many schwannomas may be encountered. Contrast enhancement can be heterogeneous if the tumour is large and the myxoid component does not enhance. Schwannoma is a differential diagnosis of rectal submucosal tumors when a submucosal mass is detected with enlarged lymph nodes in the mesorectum.[21]

Endometriotic tumour on MRI, manifests as an irregular, infiltrative, T2-hypointense mass with discrete fibrotic reaction due to chronic repetitive bleeding and scarring. It often penetrates the muscularis propria and invades the submucosa involving at least 40% of the circumference in the anterior rectal wall. On T2- weighted images, a ‘mushroom cap’ sign is indicative of solid invasive endometriosis of the rectosigmoid colon.[22] The low-signal

intensity of the mushroom base is attributed to hypertrophy and fibrosis of the muscularis propria, whereas the high-signal intensity of the cap represents the mucosa and submucosa that are displaced into the bowel lumen. Small internal cystic foci represent ectopic endometrial glands. Hyperintensity on T1-weighted, fat-suppressed images indicates subacute blood and is specific for endometriosis.[23]

Tailgut cyst (retrorectal cystic hamartoma) is a rare developmental tumour thought to arise from vestiges of an embryonic hindgut and is found in the retrorectal (presacral) space. It is more commonly found in middle-aged women.[24] Tailgut cyst usually presents as a multilocular cystic tumour with many small cysts intertwined adjacent to a main cyst, creating the appearance of honeycomb. The intensity of MRI signal on T1- and T2-weighted images vary depending on the protein, mucin, and blood content of the cysts.

Primary tumours arising from other pelvic organs (such as the uterus, ovary, or prostate) may invade the rectum. The primary role of MRI in this entity is to identify the primary tumour site.

Rectal adenocarcinoma commonly appears as an infiltrative ulcerative mass that frequently narrows the lumen and causes proximal bowel obstruction, because of its desmoplastic stroma.[25] On MRI, Rectal mass lesions show hypointense signal on T1 weighted imaging.

Several characteristics, including location, a large mass with polypoid contour, signal intensity reflecting paramagnetism, little perirectal or perianal infiltration, lack of colonic obstruction, and lymphadenopathy of large size, may be novel features facilitating the discrimination of primary anorectal melanoma from rectal adenocarcinoma.

In the present study, primary anorectal melanomas had a siginificantly similar morphologic appearance on MRI without intestinal obstruction and minimal infiltration of perirectal or perianal adipose tissue.

Histopathologically, AMM show considerable variability regarding the size and type of cells. Therefore immunohistochemical (IHC) analysis plays an important role in the diagnosis of AMM. The IHC stain widely used in the diagnosis of AMM is Anti-S-100 protein and it is highly sensitive for melanocytic differentiation. Also, human melanoma black (HMB-45), Vimentin, and Melan A antibody are the melanocyte-specific stains used for diagnosis of malignant melanoma.

Histopathological examination and immunohistochemical (IHC) analysis in the current study confirmed the primary anorectal malignant melanoma.

It was found that among sonography, CT, PET, and PET/CT, PET/CT had the highest sensitivity (86%) and specificity (91%) of metastases detection as deep soft tissue, lymph node, and visceral metastases.[26] In advanced stages of malignant melanoma (i.e., III–IV), FDG-PET/CT can be very valuable in detecting distant metastases, thereby influencing treatment decisions.[27]

The treatment of anorectal melanoma is controversial. While the typical therapeutic approach remains surgical resection, there is no consensus on which surgical approach, wide local excision (WLE) or abdominoperineal resection (APR) is preferred. APR is considered as the standard surgery for treatment of AMM, because it can regulate lymphatic spread and obtain a larger negative margin for local control.[28]

Role of adjuvant therapies is small. The medications used in adjuvant therapy are Cisplatin, Vinblastine, Dacarbazine, Interferon B, and Interleukins IL 2 8. Dacarbazine is one of the most widely used agents and usually initiates a partial response in 20% of patients after 4-6 months of treatment. There are no standard treatments for disseminated disease. Chemotherapy, radiation therapy, and immune therapy play a limited role.[2,8]

The patient in the present study underwent APR without adjuvant therapy. To date, she was undergoing follow-ups every 3 months and did not show signs of local recurrence or distant metastasis.

Conclusion

Malignant melanoma of the rectum is very rare, very aggressive, difficult to diagnose and has poor prognosis. The only hope for improved survival lies in early diagnosis and treatment. The chances of developing primary anorectal melanoma should be considered in patients with a large intraluminal polypoid mass that does not cause colonic obstruction in the anorectal area and shows T1 hyperintensity, T2 heterogeneous signal intensity, hyperenhancement, small perirectal or anal penetration, and large-sized lymphadenopathy. Although biopsy, histopathological and immunohistochemical tests are important for diagnosis, distinct radiological features in CT & MRI are useful in diagnosing rectal submucosal tumours in terms of tumour localisation, characterisation, preoperative staging and surgical planning. Surgery is still a pillar of treatment, yet the proper procedure is still controversial. Adjuvant chemotherapy, interferon and radiation may provide some benefit.

References

1. Klas JV, Rothenberger DA, Wong WD, Madoff RD. Malignant tumors of the anal canal: the spectrum of disease, treatment, and outcomes. Cancer. 1999; 85:1686–1693.
2. Stefanou A, Nalamati SP. Anorectal melanoma. Clin Colon Rectal Surg. 2011; 24:171–1769.
3. Chen MT, Sun HF, Zhao Y, Fu WY, Yang LP, Gao SP, et al. Comparison of patterns and prognosis among distant metastatic breast cancer patients by age groups: a SEER population-based analysis. Sci Rep. 2017;7(1):9254. doi: 10.1038/s41598-017-10166-8.
4. Liptrot S, Semeraro D, Ferguson A, Hurst N. Malignant melanoma of the rectum: a case report. J Med Case Rep. 2009;3:9318. doi: 10.1186/1752-1947-3-9318.
5. van Schaik PM, Ernst MF, Meijer HA, Bosscha K. Melanoma of the rectum: a rare entity. World J Gastroenterol. 2008;14(10):1633-5. doi: 10.3748/wjg.14.1633.
6. Meguerditchian AN, Meterissian SH, Dunn KB. Anorectal melanoma: diagnosis and treatment. Dis Colon Rectum. 2011; 54:638–644.
7. Falch C, Stojadinovic A, Hann-von-Weyhern C, et al. Anorectal malignant melanoma: extensive 45- year review and

proposal for a novel staging classification. J Am Coll Surg. 2013; 217:324.
8. Singer M, Mutch MG. Anal melanoma. Clin Colon Rectal Surg. 2006;19:78 87.
9. Row D, Weiser MR. Anorectal melanoma. Clin Colon Rectal Surg. 2009;22:120 6.
10. BTY Yuen, EHY Hung, CCM Cho, et al. Magnetic Resonance Imaging for Rectal Submucosal Tumours. Hong Kong J Radiol. 2017;20:272-81 DOI: 10.12809/hkjr1716935
11. Podnos YD, Tsai NC, Smith D, et al. Factors affecting survival in patients with anal melanoma. Am Surgeon. 2006;72:917–20.
12. Soh JS, Lee HS, Lee S, Bae J, Lee HJ, Park SH, et al. The clinical usefulness of endoscopic ultrasound-guided fine needle aspiration and biopsy for rectal and perirectal lesions. Intest Res. 2015;13:135- 44.
13. Goldstein HM, Beydoun MT, Dodd GD: Radiologic spectrum of melanoma metastatic to the gastrointestinal tract. AJR Am J Roentgenol. 1977, 129:605-12
14. Kim KW, Ha HK, Kim AY, Kim TK, Kim JS, Yu CS, et al. Primary malignant melanoma of the rectum: CT findings in eight patients. Radiology. 2004;232:181 6.
15. Matsuoka H, Nakamura A, Iwamoto K, Sugiyama M, Hachiya J, Atomi Y, et al. Anorectal malignant melanoma: Preoperative usefulness of magnetic resonance imaging. J Gastroenterol. 2005;40:836 42.
16. Kohli S, Narang S, Singhal A, Kumar V, Kaur O, Chandoke R. Malignant melanoma of the rectum. J Clin Imaging Sci. 2014;4:4
17. Kim H, Kim JH, Lim JS, Choi JY, Chung YE, Park MS, et al. MRI findings of rectal submucosal tumors. Korean J Radiol. 2011;12:487-98.
18. Bader TR, Semelka RC, Chiu VC, Armao DM, Woosley JT. MRI of carcinoid tumors: spectrum of appearances in the gastrointestinal tract and liver. J Magn Reson Imaging. 2001;14:261-9.
19. Reznek RH. CT/MRI of neuroendocrine tumours. Cancer Imaging. 2006;6:S163-77.
20. Yu MH, Lee JM, Baek JH, Han JK, Choi BI. MRI features of gastrointestinal stromal tumors. AJR Am J Roentgenol. 2014;203:980-91.
21. Kang JH, Kim SH, Kim YH, Rha SE, Hur BY, Han JK. CT features of colorectal schwannomas: differentiation from gastrointestinal stromal tumors. PLoS One. 2016;11:e0166377.
22. Yoon JH, Choi D, Jang KT, Kim CK, Kim H, Lee SJ, et al. Deep rectosigmoid endometriosis: “mushroom cap” sign on T2-weighted MR imaging. Abdom Imaging. 2010;35:726-31.
23. Coutinho A Jr, Bittencourt LK, Pires CE, Junqueira F, Lima CM, Coutinho E, et al. MR imaging in deep pelvic endometriosis: a pictorial essay. Radiographics. 2011;31:549-67.
24. Yang DM, Park CH, Jin W, Chang SK, Kim JE, Choi SJ, et al. Tailgut cyst: MRI evaluation. AJR Am J Roentgenol. 2005;184:1519-23.
25. Fleming M, Ravula S, Tatishchev SF, Wang HL. Colorectal carcinoma: pathologic aspects. J Gastrointest Oncol. 2012; 3:153–173.
26. Krug B, Crott R, Lonneux M, Baurain JF, Pirson AS, Vander Borght T. Role of PET in the initial staging of cutaneous malignant melanoma: systematic review. Radiology. 2008;249(3):836-44. doi: 10.1148/radiol.2493080240.
27. Petersen H, Holdgaard PC, Madsen PH, Knudsen LM, Gad D, Gravergaard AE, et al. FDG PET/CT in cancer: comparison of actual use with literature-based recommendations. Eur J Nucl Med Mol Imaging. 2016;43(4):695-706. doi: 10.1007/s00259-015-3217-0.
28. Cooper PH, Mills SE, Allen MS. Malignant melanoma of the anus: report of 12 patients and analysis of 255 additional cases. Dis Colon Rectum. 1982;25:693–703

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