SAGE Open Medical Case Reports
Volume 11: 1–6

DOI: 10.1177/2050313X231194150
journals.sagepub.com/home/sco
Sage

Primitive rectal melanoma: A rare case report

Ibtissam Elouali[1], Kawtar Imrani[1], Kenza Berrada[1], Hiba Zahi[1], Ahmed Jahid[2], Nabil Moatassim Billah[1] and Ittimade Nassar[1]

Abstract

Mucosal melanoma is a rare subtype of melanoma distinct from the cutaneous type in its clinical and biological aspects, requiring different therapeutical management. Anorectal melanomas represent less than 1% of anorectal cancers and 0.3% of malignant melanomas, and they are by far the most studied type. Proctologic examination, colonoscopy, and biopsy can establish a correct diagnosis. Imaging techniques, especially MRI can show some characteristic features, but it is essentially performed for extension assessment. We report the case of a 63-year-old man who consulted for rectal bleeding. The proctological examination found a brownish ulcerative-vegetating tumor of 3 cm in diameter located 3 cm from the anal rim. The endoscopic examination revealed a predominance of ulcerative budding lesions and the biopsy specimen confirmed a rectal melanoma. The extension assessment, based on a computed tomography scan and MRI did not show locoregional or distant metastases. Radiotherapy and abdominoperineal resection with pelvic node dissection was the treatment of choice with good evolution.

Keywords

Rectal, melanoma, imaging

Date received: 21 May 2023; accepted: 26 July 2023

Introduction

Anorectal malignant melanoma (ARMM) is a rare and highly malignant tumor representing less than 1% of all anorectal cancers and 0.3% of malignant melanomas,[1] with less than 5 years survival of 20% patients.[2]

Two-thirds of ARMM cases are misdiagnosed most often as hemorrhoids, or polyps.

Primitive mucosal melanoma is a rare disease that carries a wide spectrum of variable histological and genetic profiles leading to confusion with other malignant rectal lesions such as lymphoma, anorectal carcinomas, or sarcoma, especially in its amelanotic form.[3]

Pathogenesis and genetics of ARMM is poorly understood, which makes it difficult to make proper diagnosis and set a standardized management guideline.

Case report

We report the case of 63-year-old man, with no medical history, who consulted for tenesmus and pain preceding evacuations, in association with rectal bleeding.

Clinical examination finds nodes-free superficial ganglionic areas without any cutaneous or extra digestive suspicious lesion.

The proctological examination found a brownish ulcerative-vegetating tumor of 3 cm in diameter located 3 cm from the anal rim, involving anal sphincters.

The endoscopic examination revealed a predominance of ulcerative budding lesions.

Histopathological examination revealed pleomorphic lesions with intra- and extracellular melanin pigment suggestive of malignant melanoma. The immunohistochemical study was not performed due to the lack of laboratory reagent in our hospital.

[1]Central Radiology Department, IBN SINA University Hospital V, Rabat, Morocco
[2]Histopathology Department, IBN SINA University Hospital V, Rabat, Morocco

Corresponding Author:
Ibtissam Elouali, Central Radiology Department, IBN SINA University Hospital V, Lalla Asmaa Avenue, Salé, Rabat 11010, Morocco.
Email: Ibtissam.elouali94@gmail.com

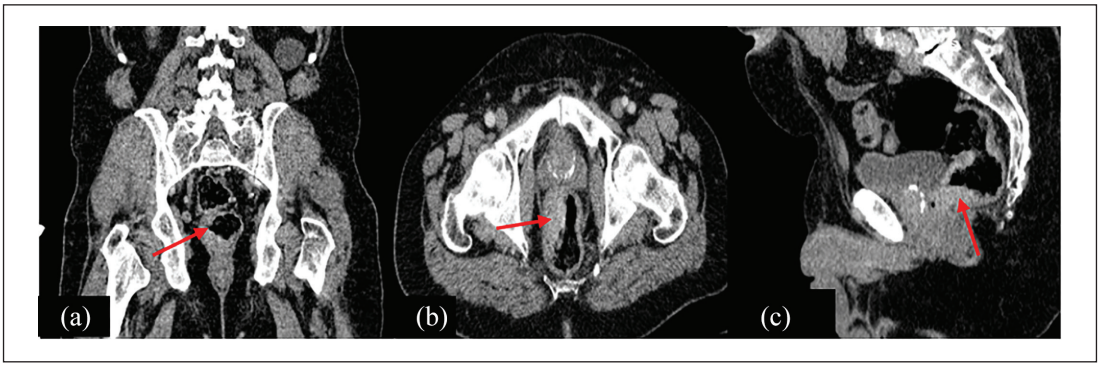

Figure 1. A contrast-enhanced computed tomography (CT) scan in a 63-year-old man with protruding anal lesion diagnosed with melanoma of the rectum shows on coronal (a) axial (b) and sagittal (c) images a heterogeneous, irregular endophytic rectal tumor without extension into peri rectal fat (red arrow).

Abdominal computed tomography (CT) scans showed an obliterating polypoidal intraluminal mass in the rectum extending to the anal sphincters, respecting mesorectal fat. There was a mesorectal lymphadenopathy but no distant metastasis (Figure 1).

MRI of pelvis was performed, for better local and regional tumor extension assessment, and showed heterogeneous intraluminal rectal mass, T1 hyperintense, intermediate signal in T2W, restrictive in diffusion sequences, strongly enhancing after GADOLINIUM administration, extending to anal sphincters, respecting the mesorectum but spreading to a mesorectal node (Figure 2). These findings are conclusive of a stage 2 anorectal melanoma and T2N1M0.

Since Breslow thickness was less than 3 mm, the surgical team opted for abdomino-perineal resection, for a better local control of the disease, pelvic lymph nodes dissection was also done and inguinal nodes were left since there was no clinical infiltration. As well, neo adjuvant pelvic radiotherapy is proposed for antalgic purposes.

Extemporaneous anatomopathological study of the mesorectal lymph node showed an infiltrated lymph node parenchyma by a melanoma (Figure 3).

This was confirmed later by histological examination of the excised anorectal tumor (Figure 4).

The evolution was unremarkable with no local recurrence or metastases after a 3 months follow-up.

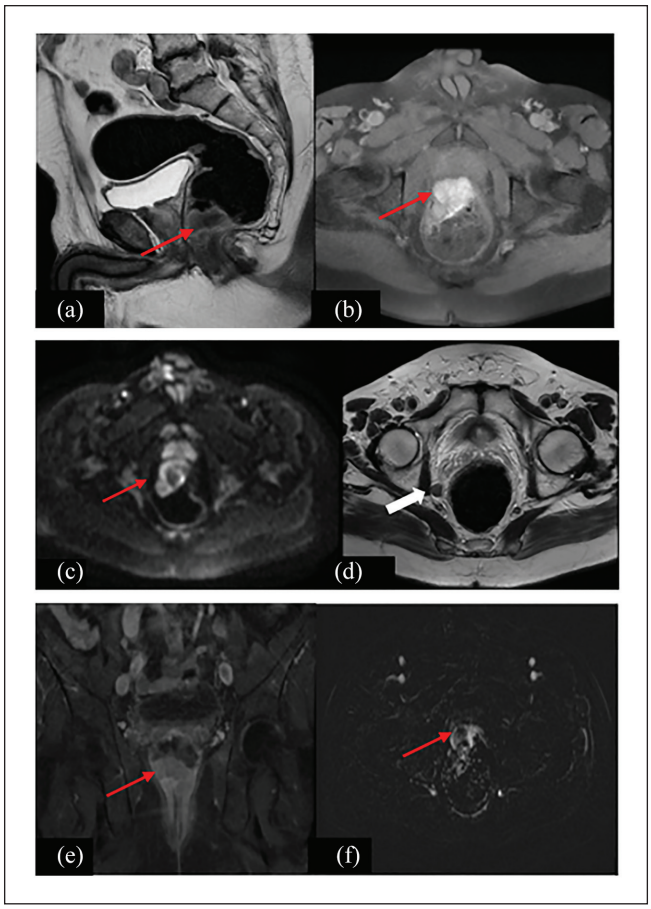

Figure 2. MRI of the pelvis of the same patient demonstrates an intermediate signal lobulated rectal mass on sagittal T2w images (a) (red arrow), T1 hyperintense (b), restrictive in diffusion sequences (c), and strongly enhancing in post-enhanced and subtracted images (e, f). This mass extends to anal sphincters but respects the peri-rectal fat. Mesorectal lymphadenopathy is also found (f) (white arrow).

Discussion

Rectal localization is rare, representing 0.3% of all melanomas and 16.5% of mucosal melanomas.[1]

The disease is more common in the elderly with a peak between the 6th and the 7th decade. There is a slightly high prevalence in female and a predominance in Caucasian race.[4,5]

As for the pathogenesis of anorectal melanomas, studies postulated some theories that are still limited and vague.[5–7]

Melanocytes are normally found in the anal squamous zone, sometimes in the anal transitional zone linked to the presence of melanocytes in the epithelial lining of the dentate line which extends proximally to the rectum.[8] These potential cells migrate, infiltrate the rectal mucosa and

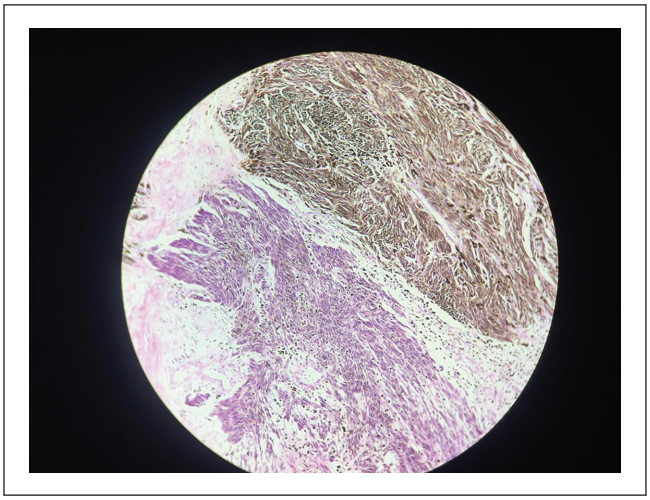

Figure 3. Extemporaneous anatomopathological study of the mesorectal lymph node showed an infiltrated lymph node parenchyma by a melanoma of cordal and massive architecture, composed of epitheloid or spindle-shaped cells of large size endowed with oval nuclei with nucleolated heterogeneous chromatin.

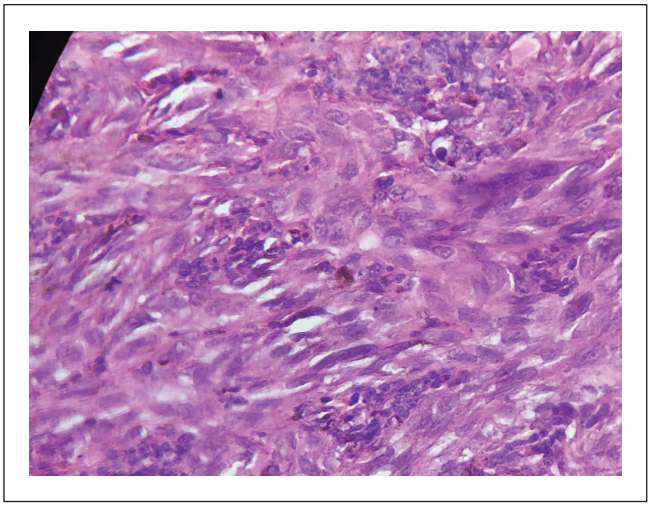

Figure 4. Squamous (anal) type mucosa. In the lamina propria, with tumoral proliferation of fusiform cells arranged in bundles and epitheloid cells arranged in thecae.

undergo neoplastic transformation probably related to oxidative stress in these regions and/or to immunosuppression. However, other valid theories have been postulated, describing that gastro-intestinal melanoma may derive from Schwannian neuroblastic cells of the autonomic intestinal innervation system or from the cells of the amine-precursor uptake and decarboxylation system.[9]

Also, a higher female incidence in anorectal melanoma is potentially due to estrogens involvement in the pathogenesis of melanoma as they increase the number of melanocytes and modify their melanin content.[10]

Melanocytes are melanin-producing somatic cells with an antioxidant activity.[11]

Melanocytes in mucosal membranes contribute to the regional immune response. Also, they are involved in immune response modulation, they increase the barrier function, constitute an antagonism of pathogens, and produce substances.[12]

The low incidence of mucosal melanoma constrains the investigations on their biological and genomic aspects.[13]

Anogenital melanomas are characterized by a highly rearranged genomes with numerous focused amplifications despite a low mitotic rate.[14] However, these mutations are responsible of a heterogeneous profile of melanomas in their pathology, describing four histologic types of anorectal melanomas: the epithelioid, the spindle-cell, the lymphoma-like, and the pleomorphic anorectal melanomas.[15–17] Studying these mutations will allow the development of specific targeted therapy for better response.[18]

The overall survival by primary site of mucosal melanoma showed a better rate for patients with an overlapping lesion of the rectum and anal canal (50%), when compared to large bowel, gall bladder, esophagus, and the pancreas.[19]

The main reasons of the delayed diagnosis are the late and nonspecific symptoms of patients with anorectal melanomas when the tumor size exceed 4 cm.

Generally, when the neoplasm involves this site, changes in bowel transit, bowel obstruction, rectal bleeding, anal pain, and/or rectal tenesmus are the most common manifestations, and they are mostly associated with a prolapsing mass through the anus.[13]

Proctological examination finds irregular polypoid lesions, mostly ulcerated, sometimes showing black or brown spots.[20] the bloody amelanotic presentation of these lesions is more frequent, often covered with mucinous/fibrinous material.[21] In this case, other differential diagnosis should be considered, especially non-Hodgkin lymphoma, adenocarcinoma, and sarcoma.

Colonoscopy with subsequent biopsy and pathological examination combined with immunohistochemical staining allows an accurate diagnosis.

Colonoscopy detects lesions and characterizes their morphology, superficial appearance and presence of melanin pigmentation, margins, colorations, origins, surface, and invasion of dentate line characteristics.[22]

Then biopsy is performed for histological and immunochemistry studies to determine cell type, degree of melanin pigmentation, and mitotic index.

These tumors are characterized by a high pleomorphism in the nucleus, epithelioid spindle-shaped, and often they enclose melanin granules.[9]

Immunohistochemical diagnosis shows a positive protein S-100, HMB-45, Melanin A, and Mart-1 antibodies.

Some reports describe a positive carcinoembryonic antigen, CD30 and CD68 and a negative AE1/AE3, CD 17 and desmin.[23]

The proliferation of atypical junctional melanocytes and atypical melanocytic cells in the basal layer in the superficial epithelium can also be considered as a histological criterion.[11]

Multiples imaging modalities are used in anorectal melanomas for primary tumor diagnosis, evaluation and staging including ultrasonography (US), endoscopic ultrasound (EUS), magnetic resonance imaging (MRI), and Positron emission tomography (PET).[24]

US's role is to characterize hepatic masses by differentiating between solid and cystic lesions.

Whereas EUS examination is good evaluation to size of the lesion, depth, and degrees of infiltration within and beyond the rectal wall. EUS shows heterogenous hypoechoic masses originating in the mucosa, with or without submucosal infiltration.[25]

On the other hand, CT scan can exhibit distant metastasis, particularly in the lungs and the liver, PET/CT is recommended in staging and response assessment of metastatic melanoma owing to the great FDG (18-Fluoro-deoxyglucose) avidity of the malignant melanocytes.[26]

MRI's key role is preoperative staging in anorectal melanoma patients.

Regardless of the site of the tumor, approximately 10%–30% of anorectal melanomas show a high intensity on T1 and a low intensity on T2-weighted sequences, because of the paramagnetic characteristic of its melanotic component which shortens the T1 relaxation time and increase the T2 relaxation time on MRI.[27]

However, in amelanotic type melanoma, these features may be absent, making it difficult to differentiate from other anorectal tumors on the basis of signal intensity alone. Also, in some cases it may show as a hyperintense mass on T1-weighted images, with a hyperintense component on T2-weighted images, due to hemorrhage or necrosis. These lesions are restrictive in diffusion, and highly enhancing after gadolinium administration.[27]

The diagnosis of anorectal melanoma as a primitive tumor can be established with complete or reasonable certainty on the basis of the following elements: No history of cutaneous or extra cutaneous melanoma, no history of removed cutaneous lesion without histological examination, the presence of atypical melanocytes along the basal epithelium in a histological sample, only at the anorectal site.[9]

Rectal melanoma spreads rapidly into inguinal lymph nodes, mesenteric lymph nodes, hypogastric lymph nodes, para-aortic lymph nodes, then into liver and lung. It shows a high incidence rate for locoregional lymph node metastases and distant metastases were found at the time of diagnosis in 26%–38% of patients.[28,29]

Melanomas can be characterized by different staging systems depending on the primary site of disease. Anorectal melanoma is staged on a clinical basis following system originally introduced by Ballantyne for the head and neck[30]:

- Stage I: clinically localized disease
- Stage II: local disease with involvement of regional lymph nodes
- Stage III: distant metastatic disease.

The TNM (tumor-node-metastasis) staging system is also used to help better communication with the surgeon, thus, a better surgical management.

There is no valid guideline for management strategies on melanoma because of the lack of randomized trials due to the low incidence of the disease.

So far, surgery is considered the cornerstone of treatment for localized diseases following abdominoperineal resection (APR) or wide local excision (WLE) techniques.[31]

APR, is a highly morbid operation used for a long period of time. Several studies describe melanoma as a systemic disease at the time of diagnosis, which means that no surgery regardless of how aggressive can make the prognosis better. Consequently, this technique has been replaced by WLE to minimize the morbidity of surgery with chances of quicker recovery and little impact on bowel function.[32,33]

Dissection of lymph nodes may be necessary in stage 2 or for occult infiltration identified with sentinel lymph node techniques.

Surgery for anorectal lesions revealed better outcomes with 33.6% 5-year survival rate, according to studies.[19]

WLE is the procedure of choices for many surgeons, followed by adjuvant radiotherapy, which leads to similar local result, with less perioperative morbidity as the radical surgery.[34]

For the locally advanced or metastatic disease, palliative surgery with partial resection with or without diverting colostomy is performed.

Radiotherapy, chemo-immunotherapy, and targeted therapy do not provide any certain results. Ongoing investigations will allow better understanding of genetic aberrations in anorectal melanoma, which will provide new effective targeted therapies (and immunotherapies).[35]

The prognosis remains poor, with a 5-year overall survival of 25%.[36]

Conclusion

Primary anorectal melanoma is a rare disease, most often presents as an ulcerative-vegetating blackish tumor, characteristic of melanoma. Imaging can show some characteristic features but its major role is the extension assessment.

Surgery is the treatment of choice in early stages, and radiotherapy can be used as adjuvant treatment to locally control the disease. Immunotherapy can also be significantly helpful due to the presence of a higher percentage of mutations in c-KIT in mucosal melanomas.

They present a poor prognosis compared to cutaneous melanomas because of the delayed diagnosis due to its

inaccessibility to physical examination and the early onset of metastases.

Acknowledgements

I would like to express my gratitude to my professors and all the colleagues who participated in the completion of this work. The authors declare that they have no competing interests.

Declaration of conflicting interests

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The authors received no financial support for the research, authorship, and/or publication of this article.

Ethical approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

Patient consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

ORCID iDs

Ibtissam Elouali https://orcid.org/0000-0001-6501-6888
Kawtar Imrani https://orcid.org/0000-0002-3710-2647
Kenza Berrada https://orcid.org/0000-0002-2322-618X

References

1. Chang AE, Karnell LH and Menck HR. The National Cancer Data Base report on cutaneous and noncutaneous melanoma: a summary of 84,836 cases from the past decade. The American College of Surgeons Commission on Cancer and the American Cancer Society. *Cancer* 1998; 83(8): 1664–1678.
2. Dasgupta T, Bowden L and Berg JW. Malignant melanoma of unknown primary origin. *Surg Gynecol Obstet* 1963; 117: 341–345.
3. Kamposioras K, Pentheroudakis G, Pectasides D, et al. Malignant melanoma of unknown primary site. To make the long story short. A systematic review of the literature. *Crit Rev Oncol Hematol* 2011; 78(2): 112–126.
4. Cress RD and Holly EA. Incidence of cutaneous melanoma among non-Hispanic whites, Hispanics, Asians, and blacks: an analysis of California cancer registry data, 1988-93. *Cancer Causes Control* 1997; 8: 246.
5. Malaguarnera M, Giordano M, Rando A, et al. Intestinal lymphoma: a case report. *Eur Rev Med Pharmacol Sci* 2011; 15: 1347–1351.
6. Takagi M, Ishikawa G and Mori W. Primary malignant melanoma of the oral cavity in Japan. With special reference to mucosal melanosis. *Cancer* 1974; 34: 358–370.
7. Holmstrom M and Lund VJ. Malignant melanomas of the nasal cavity after occupational exposure to formaldehyde. *Br J Ind Med* 1991; 48: 9–11.
8. Balachandra B, Marcus V and Jass JR. Poorly differentiated tumours of the anal canal: a diagnostic strategy for the surgical pathologist. *Histopathology* 2007; 50: 163–174.
9. Mikkelsen LH, Larsen AC, von Buchwald C, et al. Mucosal malignant melanoma—a clinical, oncological, pathological and genetic survey. *APMIS* 2016; 124: 475–486.
10. Di Rosa M, Malaguarnera G, De Gregorio C, et al. Evaluation of CHI3L-1 and CHIT-1 expression in differentiated and polarized macrophages. *Inflammation* 2013; 36: 482–492.
11. Tollenson WH. Human melanocyte biology, toxicology, and pathology. *J Environ Sci Health C Environ CarcinogEcotoxicol Rev* 2005; 23: 105–161.
12. Brady MS, Kavolius JP and Quan SH. Anorectal melanoma. A 64-year experience at Memorial Sloan-Kettering Cancer Center. *Dis Colon Rectum* 1995; 38: 146–151.
13. Flaherty KT, Hodi FS and Fisher DE. From genes to drugs: targeted strategies for melanoma. *Nat Rev Cancer* 2012; 12: 349–361.
14. Curtin JA, Fridlyand J, Kageshita T, et al. Distinct sets of genetic alterations in melanoma. *N Engl J Med* 2005; 353(20): 2135–2147.
15. Beadling C, Jacobson-Dunlop E, Hodi FS, et al. KIT gene mutations and copy number in melanoma subtypes. *Clin Cancer Res* 2008; 14: 6821–6828.
16. Cancer Genome Atlas Network. Genomic classification of cutaneous melanoma. *Cell* 2015; 161(7): 1681–1696.
17. Nakhleh RE, Wick MR, Rocamora A, et al. Morphologic diversity in malignant melanomas. *Am J Clin Pathol* 1990; 93: 731–740.
18. Hayward NK, Wilmott JS, Waddell N, et al. Whole-genome landscapes of major melanoma subtypes. *Nature* 2017; 545(7653): 175–180.
19. Shah NJ, Aloysius MM, Bhanat E, et al. Epidemiology and outcomes of gastrointestinal mucosal melanomas: a national database analysis. *BMC Gastroenterol* 2022; 22(1): 178.
20. Jensen C and Kin C. Black is the new black: prolapsing primary anorectal melanoma. *Dig Dis Sci* 2017; 62(11): 2991–2993.
21. Hillenbrand A, Barth TF, Henne-Bruns D, et al. Anorectal amelanotic melanoma. *Colorectal Dis* 2008; 10: 612–615.
22. Wang S, Sun S, Liu X, et al. Endoscopic diagnosis of primary anorectal melanoma. *Oncotarget* 2017; 8: 50133–50140.
23. Chute DJ, Cousar JB and Mills SE. Anorectal malignant melanoma: morphologic and immunohistochemical features. *Am J Clin Pathol* 2006; 126(1): 93–100.
24. Wong VK, Lubner MG, Menias CO, et al. Clinical and imaging features of noncutaneous melanoma. *AJR Am J Roentgenol* 2017; 208(5): 942–959.
25. Sashiyama H, Takayama W, Miyazaki W, et al. The diagnostic value of endoscopic ultrasonography and magnetic resonanceimaging for anorectal malignant melanoma: report of a case. *Surg Today* 2003; 33: 209–213.

26. Winkler N, Rezvani M, Heilbrun M, et al. Utility of dual phase liver CT for metastatic melanoma staging and surveillance. *Eur J Radiol* 2013; 82: 2189–2193.
27. Matsuoka H, Nakamura A, Iwamoto K, et al. Anorectal malignant melanoma: preoperative usefulness of magnetic resonance imaging. *J Gastroenterol* 2005; 40: 836–842.
28. Sachs DL, Lowe L, Chang AE, et al. Do primary small intestinal melanomas exist? Report of a case. *J Am Acad Dermatol* 1999; 41: 1042–1044.
29. Row D and Weiser MR. Anorectal melanoma. *Clin Colon Rectal Surg* 2009; 22: 120–126.
30. Ballantyne AJ. Malignant melanoma of the skin of the head and neck. An analysis of 405 cases. *Am J Surg* 1970; 120(4): 425–431.
31. Weyandt GH, Eggert AO, Houf M, et al. Anorectal melanoma: surgical managementguidelines according to tumour thickness. *Br J Cancer* 2003; 89: 2019–2022.
32. Yeh JJ, Shia J, Hwu WJ, et al. The role of abdominoperineal resection as surgical therapy for anorectal melanoma. *Ann Surg* 2006; 244: 1012–1017.
33. Iddings DM, Fleisig AJ, Chen SL, et al. Practice patterns and outcomes for anorectal melanoma in the USA, reviewing three decades of treatment: is more extensive surgical resection beneficial in all patients? *Ann Surg Oncol* 2010; 17(1): 40–44.
34. Malaguarnera G, Madeddu R, Catania VE, et al. Anorectal mucosal melanoma. *Oncotarget* 2018; 9(9): 8785–8800.
35. Knowles J, Lynch AC, Warrier SK, et al. A case series of anal melanoma including the results of treatment with imatinib in selected patients. *Colorectal Dis* 2016; 18(9): 877–882.
36. Podnos YD, Tsai NC, Smith D, et al. Factors affecting survival in patients with anal melanoma. *Am Surg* 2006; 72(10): 917–920.