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Table of Contents

Original Articles:

The impact of mental toughness training program on psychological well–being and resilience in cancer patients: A Randomized Controlled Trial **07**
Negin Hemmati, Kourosh Zarea. Samira Beiranvand, Saeed Ghanbari, Mehran Hosseinzadeh

Neuro–oncology Burden in Developing Countries: A Retrospective Cost–Analysis Study **19**
Said Nassier Said Alsenani, Rawan Nasser Almaskari, Yusra Abid Shah, Balqees Yousuf Hussain Al Mujaini, Alshifa Khalifa Alsaidi, Maryam Saud Mohammed AlBalushi, Fatema Dawood Mulla Hasan Almualem, Tariq Al–Saadi

Prospective observational comparative study of radiotherapy induced toxicities (xerostomia, skin reaction, and mucositis) between three–dimensional conformal radiotherapy and intensity modulated radiotherapy technique to oral cavity cancer of head and neck. **28**
Dr. Mhendr Dhakar, Dr. Hameeduzzafar Ghori, Dr. Atal Singh Parihar, Dr. Raju Prajapati, Dr. Om Prakash Singh, Dr. Vineeta Yogi, Dr. Vivek Tiwari, Dr. Rakesh Mahawar, Dr. Tushar Jassal

Epidemiological characteristics, prognostic factors, and survival of resectable colorectal cancer in a Moroccan population **36**
Rachid El Jim, Yousra.El Kirami, Laila.Chbani, Hind.Mrabti, Adil.Najdi, Karima. El Rhazi, Naoufel.Mellas, Ghizlane. Kharrasse. Said.Afkir, Khalid. Maazaz, Mohammed. El Abkari, Sidi Adil. Ibrahimi, Nada.Lahmidani

A Prospective Study Comparing Treatment Outcomes of Brentuximab Vedotin Plus AVD versus ABVD in Egyptian Patients with Advanced Classical Hodgkin Lymphoma **47**
Marwa N. Omar, Zeinab M. Elsayed, Rania M. Afify, Nervana M. Hussien, Haytham A. Abdelkader

Factors Influencing Breast Cancer Survival Outcomes in the Central Region of Saudi Arabia **56**
Amen Bawazir, Sadeem Alhalafi, Abdulrahman Jazieh, Wasif Ali Khan, Rumaisa Ali Khan

Assessing the use of anti–PD–1/PD–L1 cancer therapies in the Middle Eastern Country of Lebanon: a single–center study **65**
Reem El Hindi, Salim Moussa

Review Articles:

Artificial Intelligence Applications in Prognosis and Treatment of Neuro–oncology: A review **70**
Adham Al–Rahbi, Tariq Al–Habsi, Tariq Al–Saadi

Case Reports:

Lower Gastrointestinal Bleeding Revealing an Early–Stage Primary Anorectal Melanoma: A Case Report **87**
Hanane Aksim, Faisal El mouhafid, Mohamed Kaakoua, Meryem Belhamdiya, Ismail Essadi, Rachid El barni, Rachid Akka

Development of spontaneous subcutaneous emphysema in a case of squamous cell carcinoma of right lung during radiotherapy treatment a rare instance **92**
Dr Prarabdh Singh, Dr Arjun Agarwal, Dr Aditi Singh, Dr Siddharth Arora, Dr Kirti Arora, Dr Ankit Tebrawal

Conference Highlights/Scientific Contributions

- **News Notes** **96**
- **Advertisements** **99**
- **Scientific Activities** **101**

Case Report

Lower Gastrointestinal Bleeding Revealing an Early–Stage Primary Anorectal Melanoma: A Case Report

Hanane Aksim [1], Faisal El mouhafid [2], Mohamed Kaakoua [3], Meryem Belhamdiya [1], Ismail Essadi [3], Rachid El barni[2] , Rachid Akka[1]

[1]Hepatogastroenterology Department, Military Hospital of Avicenne, Marrakech, Morocco
[2] Visceral Surgery Department, Military Hospital of Avicenne, Marrakech, Morocco
[3] Medical Oncology Department, Military Hospital of Avicenne, Marrakesh, Morocco

Abstract

Introduction : Anorectal melanomas are rare and agressive malignant tumors of the anorectal region of the digestive tract. Due to their rarity and similarity to other more benign and common conditions, such as hemorrhoids, their diagnosis is often delayed, which worsens the prognosis of this aggressive disease. Histological evaluation with special immunohistochemical (IHC) staining is often essentiel to establish a confirmed diagnosis. In the localized stage, the primary treatment for this condition consists primarily of surgical resection combined with adjuvant immunotherapy.

Case presentation : We report the case of a 62–year–old women who presented for an etiological evaluation of mild rectal bleeding without other associated signs. Proctological examination revealed the presence of a flashy mass occupying almost the entire anal lumen, with irregular thickening of the anterior anorectal wall during rectal examination. The colonoscopy revealed a friable and ulcerated process in the lower and middle rectum, and several biopsies were performed, confirming the diagnosis of rectal melanoma. The patient was successfully treated with surgery and adjuvant immunotherapy.

Discussion/ conclusion : Our case demonstrates the importance of rapid diagnosis and a multidisciplinary therapeutic approach for a better prognosis of malignant rectal melanoma.

Keywords: anorectal melanoma, rectal bleeding, immunohistochemistry, abdominoperineal resection.

Introduction

Anorctal melanoma is a rare cancer, accounting for less than 1% of all melanomas reported worldwide. It accounts for only 0,5% to 4% of anorectal cancers [1,2]. It is more common in women and its incidence is increasing in older people [3,4]. More than 65% of cases are diagnosed in patients over 60 years of age [5].

One of the main problems in diagnosing rectal melanoma is the vague nature of its clinical presentation. Patients often report general symptoms such as rectal bleeding or anal pain, which can easily be attributed to more common conditions. The agressive nature of malignant rectal melanoma, its nonspecific clinical presentation, and its rarity make diagnosis and treatment particularity challenging. To address these issues, its is essential to have a through understanding of the epidemiology, symptoms, and therapeutic approaches to the disease. A multidisciplinary approach is also essential to improve patient outcomes.

Patient with rectal melanoma generally have a poor prognosis. The five–year survival rate is between 10% and 15%, with adjuvant immunotherapy remains the cornerstone of treatment for localized stages, other options such as chemotherapy and radiotherapy are considered to have limited efficacy [6]. This article presents the case of 62–year–old women diagnosed with primary anorectal melanoma (PARM) who was treated with abdominoperineal resection and adjuvant immunotherapy.

Case Presentation

We present the case of a 62–year–old woman admitted to our gastroenterology clinic for an etiological

Corresponding author: Hanane Aksim, Hepatogastroenterology Department, Military Hospital of Avicenne, Marrakech, Morocco.
E–mail address: maksimhanane@gmail.com

assessment of mild rectal bleeding. The patient has no significant medical history, in particular no personal or family history of digestive neoplastic disease.

The clinical history began one year ago with the onset of intermittent, mild rectal bleeding, which then became persistent without any other associated symptoms, in particular no transit disorders, anal pain, or perineal burning, all occurring in an apyretic context and with no change in her general condition.

The clinical examination found the patient to be conscious (Glasgow score 15/15), normocardic (HR 80 beats/min), normotensive, eupneic, and apyretic (36°C). The abdominal examination was unremarkable. The other examinations were unremarkable, with no skin lesions. Proctological examination revealed the presence of a fleshy mass of uniform pinkish color, without blackish spots, voluminous, occupying almost the entire anal lumen (Figure 1). The rectal examination revealed an irregular mass on the anterior rectal wall, extending from the anal margin to an upper limit that could not be reached by the finger; the posterior rectal wall was normal and the finger cot was stained with blood.

The blood test showed a hemoglobin level of 11 g/dL and a C–reactive protein (CRP) level of 40 mg/L. The rest of the blood test (blood ionogram, liver function, and kidney function) was normal. A colonoscopy revealed a friable and ulcerated process, which partially obstructed the lumen and extended approximately 12 cm into the rectum from the anal margin without other locations (Figure 2). No synchronous lesions were detected. Several biopsies were taken from this mass.

Histopathological examination of the mass was inconclusive on two occasions, while the third biopsy revealed the presence of round tumor cells with slightly enlarged hyperchromatic nuclei and indistinct cytoplasmic boundaries. Immunohistochemical (IHC) staining demonstrated the expression of melanoma markers: S–100 and SOX–10 (Figure 3). A diagnosis of anorectal melanoma was made.

The contrast–enhanced thoracoabdominal pelvic CT scan revealed a lesion centered on the anal canal extending to the lower rectum, lateralized to the left, measuring a total of 25 mm in transverse diameter, extending approximately 6 cm, with no signs of distant metastasis. Additional radiological imaging by pelvic MRI revealed a locally infiltrating anorectal tumor process involving the sphincter apparatus, budding endoluminally in the lower and initial portion of the middle rectum (Figure 4), without satellite pelvic lymphadenopathy, which could be classified as T3N0M0.

The patient was initially treated surgically, undergoing abdominoperineal resection following a multidisciplinary consultation meeting on tumor treatment. The surgical specimen was sent for histopathological examination, which confirmed the diagnosis of malignant melanoma of the rectum (Figure 5). The postoperative period was uneventful and unremarkable. The patient was then referred to the medical oncology department for possible adjuvant immunotherapy with pembrolizumab 200 mg every 3 weeks for one year (18 cycles). After a median follow–up of 2 months.

Discussion and Conclusion

Melanomas affect organs that contain melanocytes, such as the nasal cavity, eyes, epidermis, vagina, oropharynx, rectum, anus, and urinary tract [7]. While rectal localization is rare, it accounts for 0.3% of all melanomas [8]. Anorectal melanoma is a very aggressive

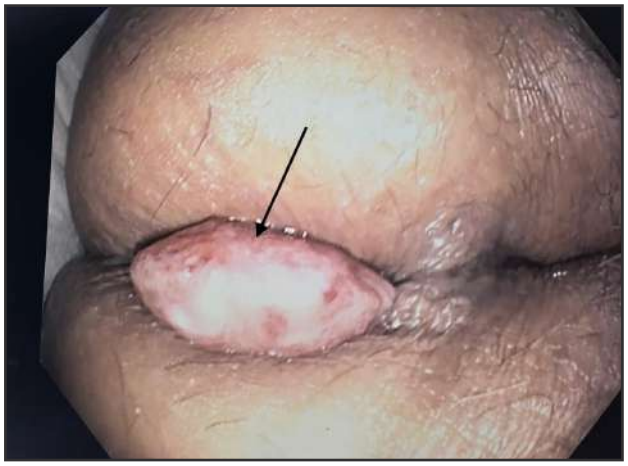


Figure 1: Photo showing the protrusion of the anorectal mass; the arrow indicates the mass protruding into the anal canal, which does not show any black pigmentation.

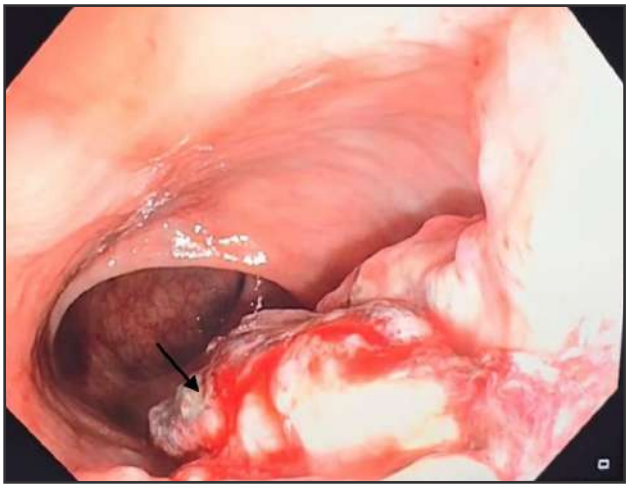


Figure 2: Photo taken during colonoscopy showing a congested and friable mass (arrow) partially obstructing the lumen.

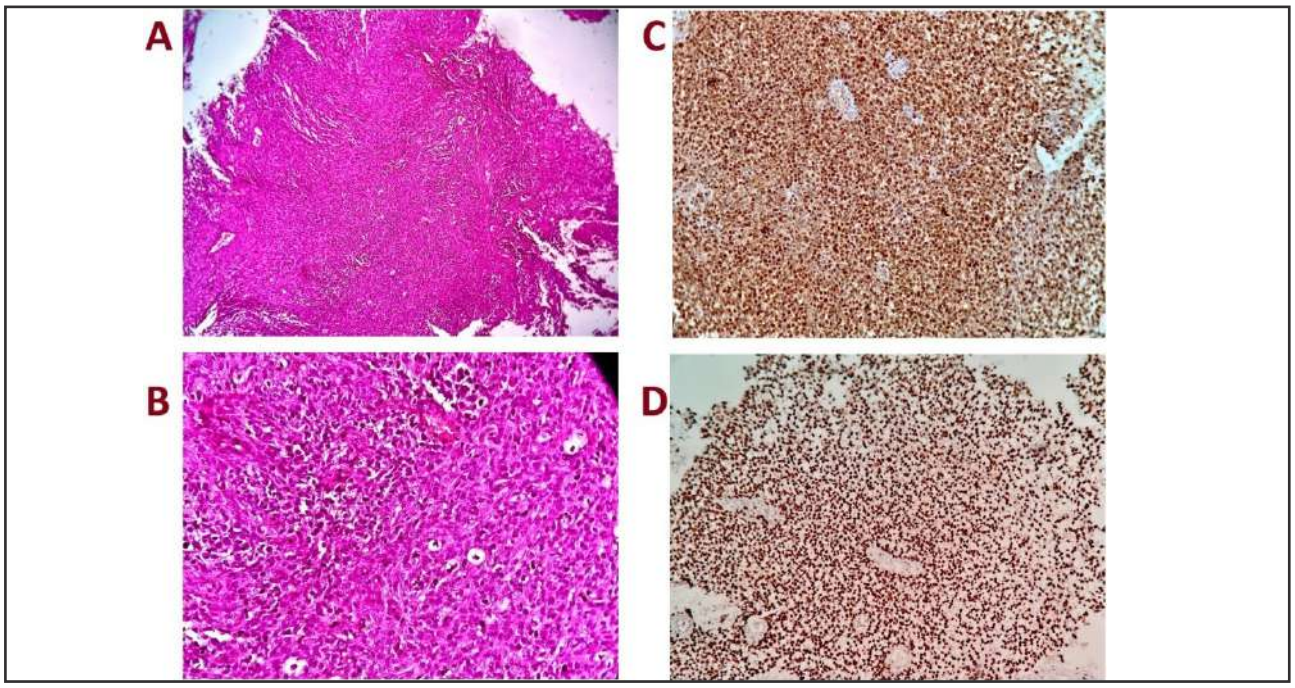


Figure 3: pathological study, A: microscopic features showing dense and undifferentiated tumor proliferation (HE,x40), B :tumor cells are round with slightly enlarged, hyperchromatic nuclei and unclear cytoplasmic boundaries,(HE,x200), C: positive labeling of tumor cells by anti–PS100 antibody, D: positive labeling of tumor cells by anti–SOX10 antibody.

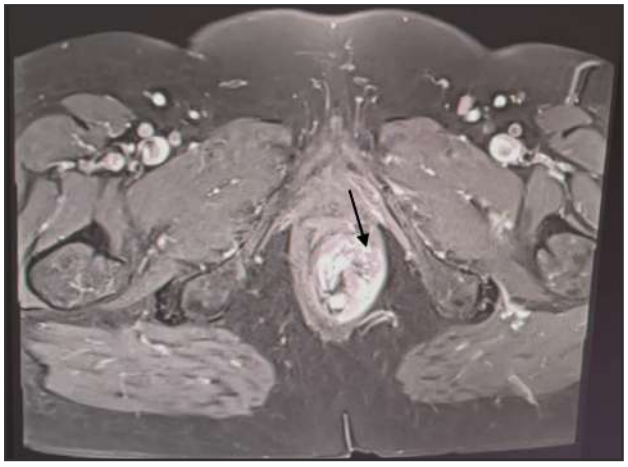


Figure 4: Axial pelvic MRI showing an irregular and heterogeneous anorectal mass (arrow).

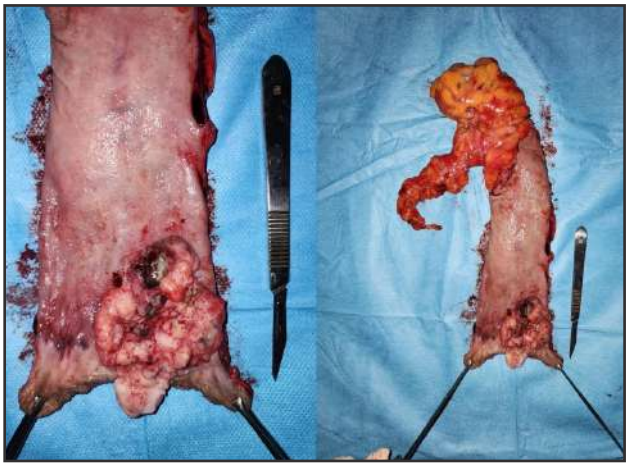


Figure 5: Abdominoperineal amputation specimen, showing the anorectal tumor.

tumor that tends to spread via the lymphatic system to the inferior mesenteric or inguinal pelvis. The most common secondary sites are the mesenteric, inguinal, hypogastric, and para–aortic lymph nodes, the lungs, the brain, the liver, and the skin. Almost 60% of cases present with locoregional lymph node metastases at initial diagnosis, while distant secondary sites are observed in 26 to 38% of cases [1–9].

It occurs at all ages, but mainly affects people over the age of 60. It generally affects women more than men.

The most common clinical manifestations of these tumors are bleeding, anal pain during defecation, changes in bowel habits, and palpable masses in the anorectal region. Other manifestations may include tenesmus, hemorrhoidal prolapse, changes in bowel habits, pruritus, and diarrhea. In advanced stages, anemia, weight loss, and fatigue may also occur [9,6]. Anorectal conditions may be misdiagnosed as hemorrhoids or benign polyps, leading to a delay in diagnosis [10,6].

Rectosigmoidoscopy is indicated for taking biopsy samples and investigating the cause of symptoms, while a full colonoscopy is recommended to investigate other identical locations. Endorectal ultrasound, computed tomography, and magnetic resonance imaging are

indicated to determine the size of the mass and the presence of secondary locations.

Approximately 61% of patients with anorectal melanoma have regional lymph node involvement [11], and 26% to 38% of patients already have metastases at the time of diagnosis [12]. Our patient showed no signs of lymph node or distant metastasis at the time of presentation on CT and pelvic MRI.

Despite the difficulties associated with detecting melanin in anorectal diseases, its presence can be invaluable in cases where diagnosis is difficult. Immunohistochemical markers such as S–100, HMB–45, and vimentin are essential for confirming the diagnosis of melanoma and differentiating it from squamous cell carcinomas. In addition, mutations in the KIT gene, associated with gastrointestinal stromal tumors and leukemia, have been linked to the development of malignant melanoma [9,13].

Despite their rarity, anorectal melanomas are a cause for serious concern due to their poorer prognosis than other melanomas and their aggressive nature. As with malignant tumors, early diagnosis is essential to optimize survival rates [14,15]. Several authors consider that the prognosis of anorectal melanoma is generally determined by the stage of the tumor at the time of diagnosis. The presence of deep infiltration of the rectal wall, tumor necrosis, perineural invasion, lymph node metastases, and distant metastases are associated with poor overall survival [16,6]. However, survival rates have been shown to improve when the tumor thickness is less than 4 mm[17]. Patients with anorectal melanoma have a five–year survival rate of between 16% and 34%, with a decrease in recurrence–free survival to 16% for those who have metastases at the time of diagnosis [9,6].

Unfortunately, the results of treatment for anorectal melanoma remain suboptimal. The recommended first–line treatment for localized stages is surgery, either abdominoperineal resection or wide local excision (WLE). Recent studies suggest that wide local excision may offer comparable disease control with less surgical morbidity [1,18,9,6]. Lymph node dissection, however, is crucial when the disease is clinically evident or detected by sentinel lymph node techniques [12–9].Until a few years ago, adjuvant systemic treatment in localized stages was not recommended due to the lack of benefit from chemotherapy in this type of cancer. However, since 2018, adjuvant immunotherapy has become standard in the treatment of localized melanomas with a high risk of recurrence (stage IIb/III). Two molecules have been approved for their efficacy in this situation: nivolumab (anti–PD–1), which has been shown to reduce the risk of recurrence by 58% (CheckMate 76 K study) [19] and pembrolizumab (anti–PD 1), which also improved recurrence–free survival by 35% (Keynote 716 study) [19].

With regard to post–treatment follow–up, there is no clear consensus, but a clinical examination including a detailed proctological examination, palpation of the lymph nodes, periodic colonoscopy, as well as a thoraco–abdominal–pelvic CT scan and a brain CT scan seem to be strongly indicated in the monitoring of patients with melanoma [20].

Anorectal melanoma is difficult to diagnose and treat due to its vague clinical presentation and the fact that it is often diagnosed at a late stage. Early diagnosis and prompt, appropriate treatment are the only means of achieving a cure. Surgical treatment remains the cornerstone of treatment for localized anorectal melanoma, and adjuvant immunotherapy has significantly improved the prognosis for this condition.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Contact :
The Gulf Federation for Cancer Control
P. O. Box 26733 Safat 13128 Kuwait
Tel. (00965) 22530186 / 22530184 Fax: (00965) 22510137
website : http://www.gffcc.org
email : gffccku@yahoo.com

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