Radiology Case Reports 19 (2024) 1215–1221

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Case Report

Primary malignant melanoma of the small bowel: A case report ☆

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ARTICLE INFO

Article history:
Received 12 March 2023
Revised 27 November 2023
Accepted 6 December 2023
Available online 6 January 2024

Keywords:
Malignant melanoma
Small intestine malignancy
Aggressive tumor
Gastrointestinal Radiology
Oncoimaging

ABSTRACT

Malignant melanoma comprises 1%-3% of all malignant tumors of the gastrointestinal tract. The small bowel melanoma is an extremely rare malignancy. Very few cases have been reported in the literature. The small intestine is mostly affected by the metastatic tumors of the primary lesions especially cutaneous. This malignancy is diagnosed at the late stages as the patient remains asymptomatic. In this report we present a case of malignant melanoma arising from the small bowel in a 58-year-old male. There was no primary lesion in the eye, skin, anus, rectum or elsewhere in the body. The patient was treated with surgery. Afterward the patient presented to the emergency room with respiratory distress for which he was on ventilator support, sadly the patient died after 10 days. Malignant melanoma is an aggressive tumor and does not respond well to chemotherapy or radiotherapy.

Introduction

Melanoma develops from the melanocytes which are commonly located in the skin, eye's choroid, meninges and anal margin. Malignant melanoma comprises 1%-3% of all malignant tumors of the gastrointestinal tract [1,2]. Most of the tumors are metastatic from the other primary lesion [3]. A thorough search in Medline/ PubMed, nearly 40 cases of primary malignant melanoma of the small bowel have been reported so far [4]. Primary melanoma of the small bowel remains a diagnostic challenge due to its rarity and the difficulty in excluding another primary carcinoma [5]. It is an aggressive tumor with a poor prognosis in comparison to other non-GIT melanoma. The median survival is 4-6 months with a 5 years survival rate of less than 10% [6,7]. There is no standard treatment protocol of primary melanoma of the GI. However, surgery remains an important management since chemoradiotherapy does not affect the overall survival of the patient [8].

Case report

A 58 year old male presented to our institute with pain abdomen for the last 4 months which was unrelieved by analgesics. It was progressive in nature. The patient also com-

☆ Competing Interests: There is no interest to declare.

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https://doi.org/10.1016/j.radcr.2023.12.005

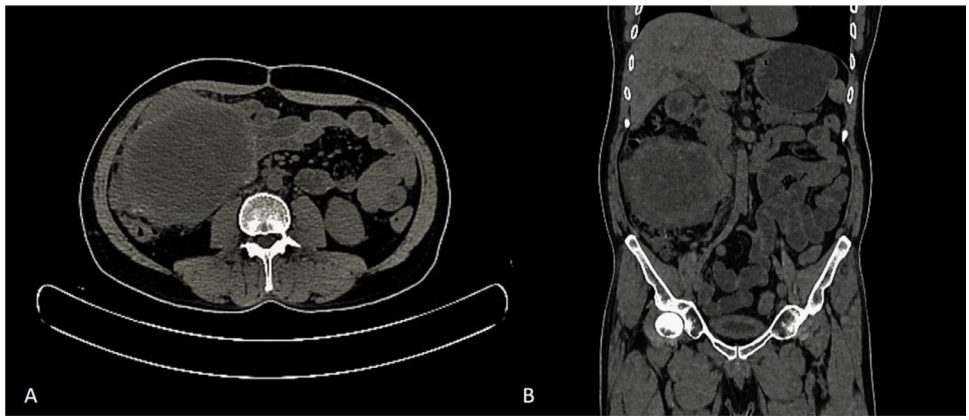

Fig. 1 – Primary malignant melanoma of the small intestine: noncontrast CT showing hypodense lesion with no evidence of intratumoral calcification or hemorrhage (A) axial (B) coronal.

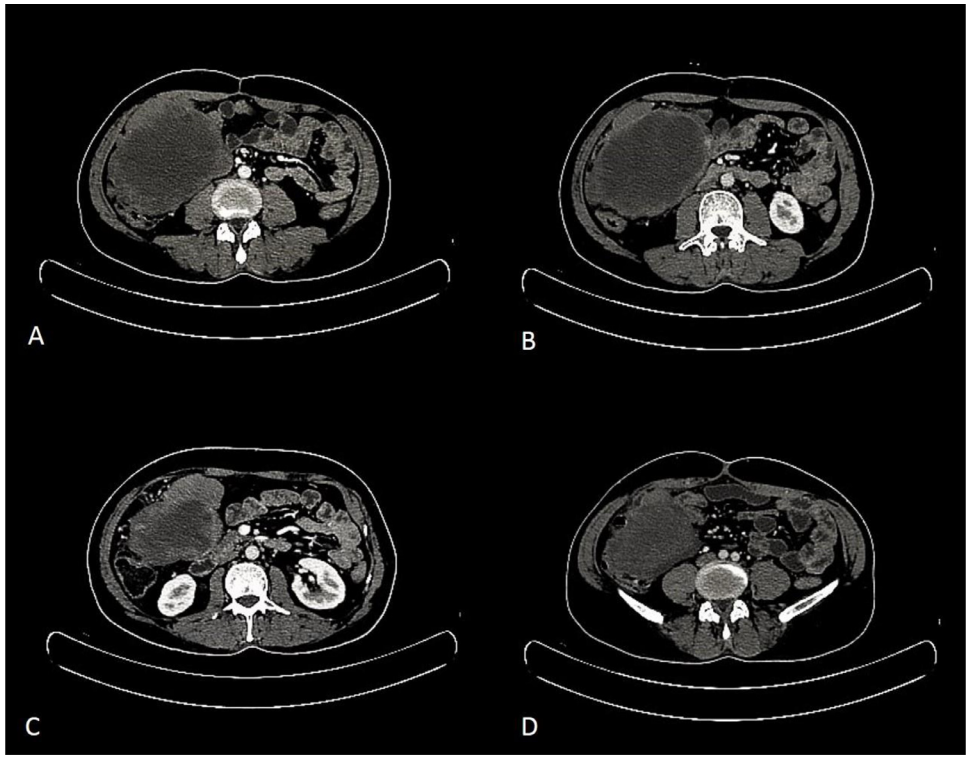

Fig. 2 – Primary malignant melanoma of the small intestine: contrast enhanced CT axial showing heterogeneously enhancing lesion with areas of hypodensity suggesting necrosis.

plained of swelling of abdomen since 2 months. The symptoms were aggravated by melena and asthenia. Clinical examination revealed a palpable and mobile abdominal mass on the right side.

Laboratory tests showed hypochromatic and microcytic anemia with low hemoglobin and hematocrit levels. Tumors markers CEA, CA19.9, and AFP were normal. Other blood parameters were within normal limits. Abdominal ultrasound demonstrated a heterogeneously hypoechoic mass lesion in the right lumbar region arising from the bowel loop. Plain and contrast enhanced CT was performed with a 64 slice Philips Brilliance scanner, 80 mL nonionic contrast (ultravist, 370mg I/mL) was injected with a rate of 4 mL per second through 18G intravenous cannula. Non contrast CT demonstrated a large hypodense mass (Hounsfield unit [HU] value 28.45) showing no intratumoral calcifications or hemorrhage (Fig. 1). Contrast enhanced CT demonstrated a large heterogeneously enhancing mass lesion measuring $\sim$ 10.5 $\times$ 12 $\times$ 10 cm with areas of necrosis, arising from the ileal loop. Anteriorly the lesion was abutting the anterior abdominal wall with no evidence

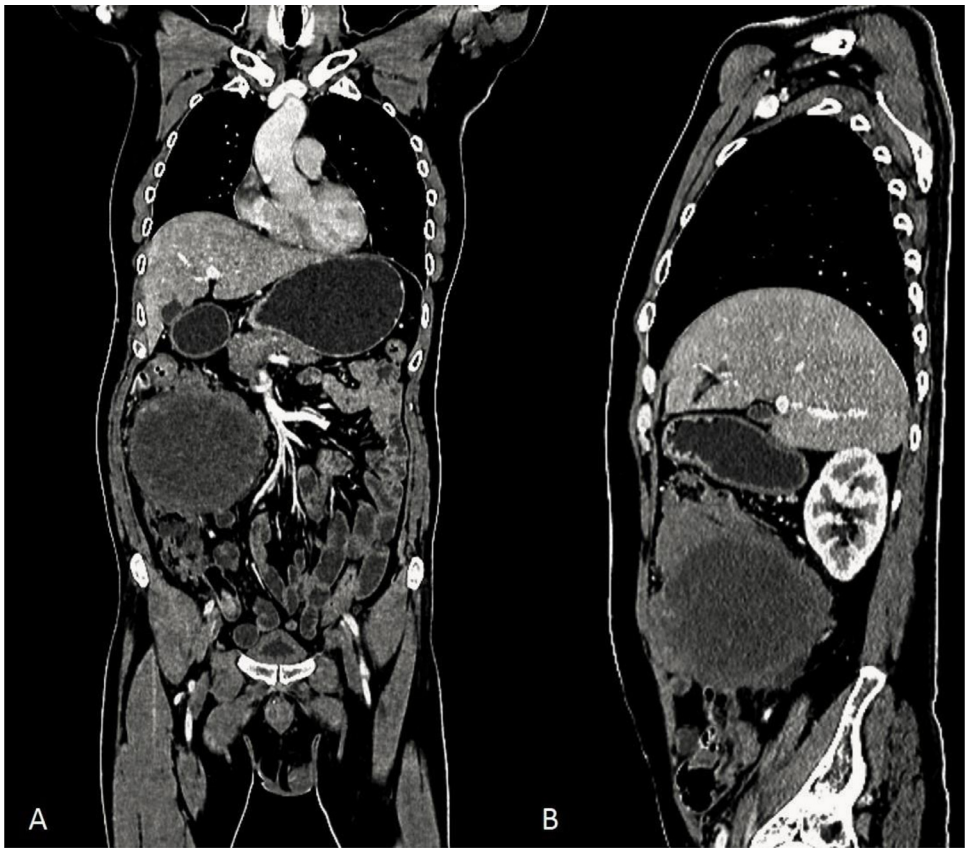

Fig. 3 – Primary malignant melanoma of the small intestine: contrast enhanced CT, showing heterogeneously enhancing lesion with areas of necrosis in mid abdomen on right side (A) coronal, superior mesenteric vein is normally visualized, (B) sagittal, fat plane with the right kidney is maintained.

of infiltration. Superior mesenteric artery and vein were normally visualized (Figs. 2 and 3). After careful examination of the mass lesion on imaging, differential diagnoses included was GIST and primary malignancy of the small bowel like adenocarcinoma. Imaging guided biopsy was done to confirm the diagnosis.

A histological examination revealed a malignant proliferation of large cells with prominent round nuclei with spots of eosinophilic cytoplasm and melanin pigments. Tumor necrosis was evident. On immunohistochemistry, Melan-A, HMB-45 antigens and PS-100 were diffusely positive, suggesting malignant melanoma. The etiological investigations revealed no primary melanoma elsewhere in the body. (Fig. 4)

PET-CT showed heterogeneously increased pathologic FDG uptake (SUV max 12.9) in the periphery of the lesion and the solid component of the lesion. No evidence of uptake in the rest of body (Figs. 5 and 6).

To our knowledge, this case is scarcely report in the literature as to its imaging appearance and management. The patient was treated with surgery. Afterward the patient presented to the emergency room with respiratory distress for which he was on ventilator support, sadly the patient died after 10 days

Discussion

Primary small bowel melanoma rarely occurs; however, it commonly occurs as a metastatic tumor of the underdetermined cutaneous melanoma. In this case, the patient had no previous cutaneous lesion. In such cases, the intense diagnostic work up is necessary to define if the tumor is primary or secondary. Melanoma can originate at any site within the GI mucosa, it is the most common in the anorectal region (anal canal - 31.4%, rectum -22.2%), followed by the oropharyngeal region (32.8%), esophagus (5.9%), stomach (2.7%), small intestine (2.3%), gall bladder (1.4%) and large intestine (0.9%) [9,10].

There are many hypothesis for the genesis of the small bowel melanomas [2]. Small bowel does not have the melanocytes, however occasionally these cells can be present in the alimentary tract, thus supporting the hypothesis of primary lesion in these sites [2]. Second, malignant melanoma originates from the intestinal Schwann cells, confirmation of this theory is debatable [2]. Finally, malignant melanoma originate from the neural crest, and these potential cells relocate via the umbilical-mesenteric canal and later transform into the specialized cells (amine precursors uptake and de-

Fig. 4 – Primary malignant melanoma of the small intestine: 18FDG PET-CT shows peripheral pathologic FDG uptake in the lesion with FDG nonavid central necrosis.

carboxylation cells) which is subjected to the neoplastic transformation [2]. According to this hypothesis (APUD theory), the commonest location of the primary malignant melanoma of the small bowel should be ileum which represent the distal end of the umbilical mesenteric canal [2,11,12]. To determine whether the small intestine melanoma is a primary lesion, few diagnostic criteria are postulated. Sacks et al., established the three diagnostic criteria: 1) single lesion, 2) no primary lesions in the other organs with the absence of enlarged lymph nodes, 3) more than 1 year of survival after the diagnosis [13]. In another study, Blecker et al., proposed the diagnostic criteria which is important to strengthen the diagnosis of the true primary small intestine melanomas [14,15]. This includes 1). The presence of a single solitary tumor in the mucosa of the intestine, 2). The existence of the other intramucosal melanocytic lesions in the adjacent intestinal epithelium; 3). The absence of any cutaneous or mucosal melanoma or other melanocytic dermal lesions. Nonspecific clinical manifestations include rectal bleeding, melena, chronic persistent abdominal pain, vomiting, diarrhea, weight loss and microcytic anemia. Few cases of the small intestine malignant melanoma also presented with acute small bowel obstruction, intussusception and perforation for which the patients underwent emergency laparotomy [4,14], however these symptoms were absent in our case. Reemst et al., reported less than 5% of the patients presented late with nonspecific symptoms [16].

Upper GI endoscopy and colonoscopy failed to detect the small intestinal pathology in all the reported cases. Abdominal ultrasound, CT scan, technetium studies, PET-CT and capsule endoscopy must be considered as an alternating diagnostic tool [2]. The diagnostic sensitivity of CT scan is only 86% [17]. PET-CT is necessary to monitor the disease progression, its advanced stage and to direct the treatment.

Ultrasound guided percutaneous biopsy of small bowel lesions is an important safe alternative when an endoscopic procedure fails to obtain the tissue for histopathological diagnosis. Few studies have shown the efficacy of USG guided biopsy with the diagnostic accuracy ranging from 97% to 99%, 97% to 99.1% sensitivity and 100% specificity [18-20]. Endoscopic ultrasound guided biopsy is the procedure of choice in cases of the submucosal or subserosal lesions.

The histological criteria for the primary small bowel melanoma is the increase of the atypical junctional melanocytes and atypical melanocytic cells in the basal layer in the superficial epithelium. The other important feature of the primary lesions are the presence of lymphocytes infiltrating surrounding the tumor mass [21,22].

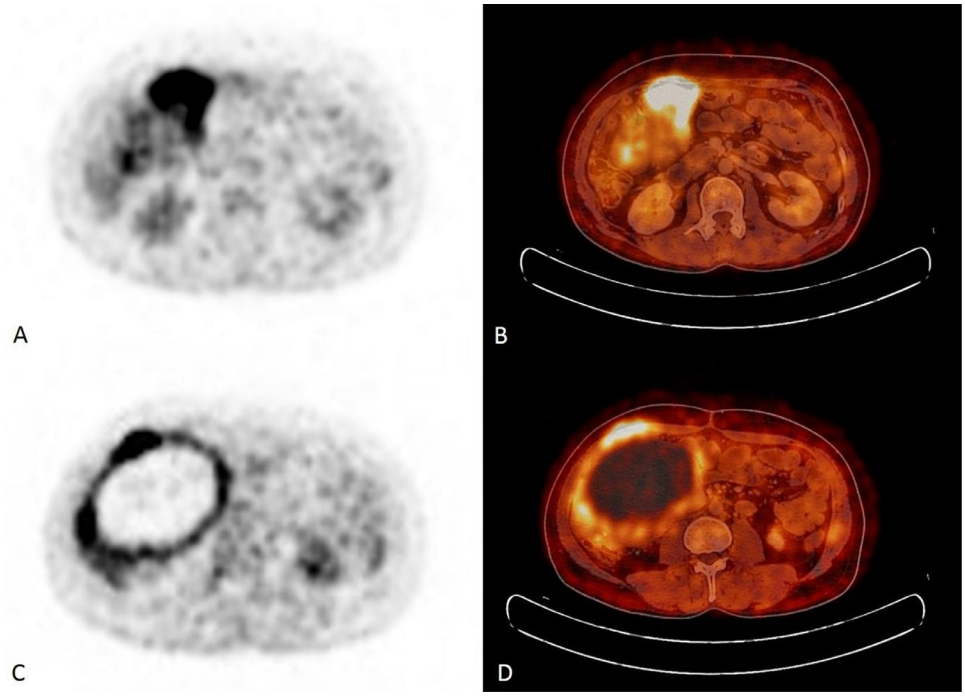

Fig. 5 – Primary malignant melanoma of the small intestine: 18FDG PET-CT shows pathologic FDG uptake in the solid component of the lesion (A, B), as well as in the margin of the lesion (C, D).

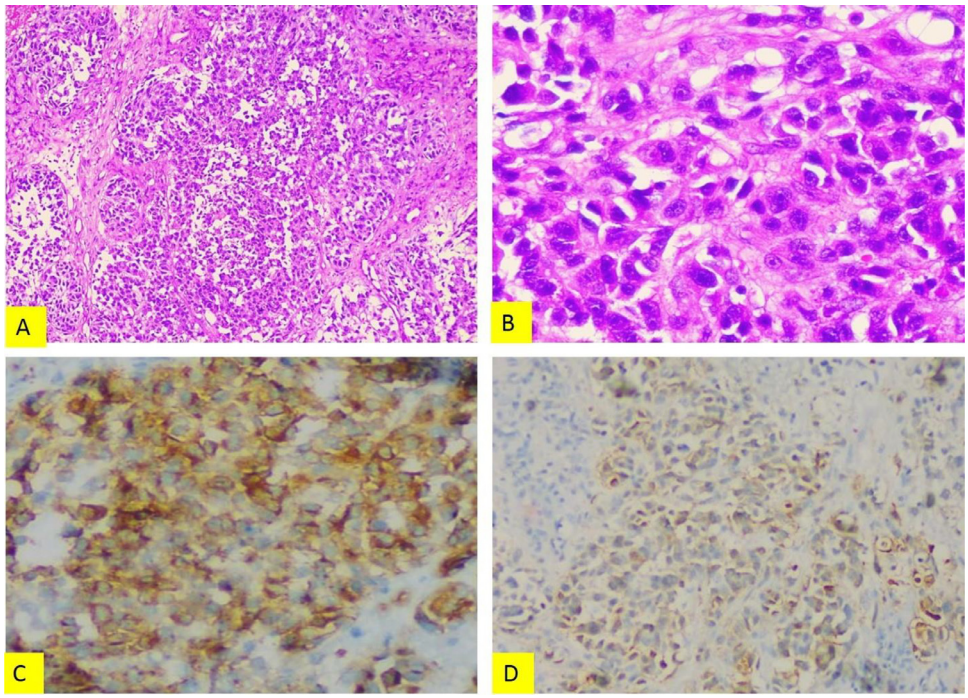

Fig. 6 – Primary malignant melanoma of the small intestine: (A): Hematoxylin & Eosin (H&E) stained section showing tumor arranged in form lobules, nests and sheets, 100x; (B): Hematoxylin & Eosin (H&E) stained section showing round to polygonal tumor cells with coarse chromatin, prominent nucleoli and moderate cytoplasm, 400x; (C): HMB45 immunostaining showing granular cytoplasmic positivity in tumor cells, 400X; (D) S100 positivity in tumor cells, 200x.

Surgical resection of the tumor is the mainstay of treatment [23,24]. GI metastatic tumor resection is advised when curative surgery is not possible in order to relieve the acute symptoms or any future complications [25,26]. Adjuvant therapies like chemotherapy, immunotherapy and biochemotherpy have been attempted, however their efficacy has not been established [27]. Primary intestinal melanomas has worse prognosis since they tend to grow faster and more aggressively [14,23]. The fatal outcome has been observed in a high percentage of cases [28].

Conclusion

Primary malignant melanoma of the small bowel is extremely rare with an aggressive clinical course and poor prognosis. Fatal outcome has been observed in a high percentage of cases. The possibility of other primary sites should be considered, but the possibility of a single primary lesion cannot be neglected. Its early diagnosis and appropriate treatment can improve the overall survival.

Patient consent

Written informed consent for the publication of this case report was obtained from the son of the patient.

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