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Case Report

Primary Small Bowel Amelanotic Melanoma and Intestinal Intussusception: a case report

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Abstract: Primary gastrointestinal melanomas (PGM) are usually asymptomatic in the early stages, leading to misdiagnosis. We reported the case of a 75-year-old male patient admitted to our service due to hematochezia, and fatigue. Primary investigation showed small bowel intussusception and surgical treatment was performed. Histopathology and immunohistochemistry analysis identified primary small bowel amelanotic melanoma at an advanced tumor stage. Whereas the patient had a sudden impairment of performance status during hospitalization, no curative oncological approach could be performed. This report highlights the clinical and imaging findings that led to the primary gastrointestinal amelanotic melanoma diagnosis in an elderly patient with intestinal intussusception. The early recognition of prior intussusception signs in adults and elder patients may lead to an earlier oncological diagnosis and a better prognosis.

Keywords: Amelanotic Melanoma; Small Intestine Cancer; Intestine Intussusception.

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1. Introduction

Melanomas are melanocytes malignant tumors. The skin is the primary neoplastic site with the highest incidence, accounting for approximately 324,635 new cases worldwide [1, 2]. Since gastrointestinal tract melanomas occurrence is commonly related to neoplastic metastasis, primary gastrointestinal melanomas (PGM) are rarely diagnosed and described [3, 4]. The origin of PGM is uncertain. The most accepted theories include the migration of melanoblasts cells from the neural crest to the small bowel, malignant mutations in non-cutaneous neuroendocrine enteric tissue cells, and also metastatic lesions of underdiagnosed cutaneous melanomas [5-7].

The clinical presentation of small bowel melanomas is often asymptomatic and non-specific at an early stage of the disease [8]. Although intestinal intussusception is a rare condition in healthy adults, PGM are responsible for 60% of colonic intussusceptions and 30% of small bowel intussusceptions in adult patients [4, 8, 9]. Hence the low PGM incidence and the uncommon association with small bowel intussusception, the present study reports a case of a 75-year-old male with primary small bowel amelanotic melanoma who presented intestinal intussusception on admission.

2. Case Report

A 75-year-old ex-smoker (low smoking load), non-elitist, and hypertensive male patient presented a thirty-day evolution of hematochezia and fatigue on minimal efforts at the emergency service of a Brazilian university hospital. Although the patient was lucid

and orientated, he exhibited a regular general state with tachycardia and a slowed capillary refill time. There were no peritoneal irritation signs, nor other alterations in the abdominal examination.

Upon admission, the laboratory evaluation revealed severe hypochromic microcytic anemia (4.8 g/dL hemoglobin) with no other associated abnormalities. No infectious signs were noticed in the laboratory analysis. The patient was then hospitalized and stabilized by the administration of crystalloids and two concentrated blood bags. For further investigation of the clinical etiology, digestive endoscopy and colonoscopy were performed. The endoscopy exam evidenced areas of intestinal metaplasia in the distal esophagus, a positive urease test for H.Pylori, and moderate enanthematous gastritis of the antrum. The colonoscopy findings did not reveal any significant clinical or anatomopathological changes.

A Fourteen-day H. Pylori eradication treatment (Amoxicillin 1g, Clarithromycin 500mg, and Omeprazole 40 mg) was prescribed. The patient was discharged after stabilization and improvement of his condition. An outpatient clinic appointment was then scheduled. Four days after dischargement, the patient returned to the emergency service of the hospital presenting new complaints of abdominal pain, hyporexia, hematochezia, dyspnea, nausea, and vomiting. The symptoms were not related to food intake. No fever, dehydration or peritoneal irritation signs were noticed.

Abdominal ultrasound was then performed. The images suggested intestinal intussusception in the left mesogastrium with adjacent parietal thickening of small bowel loops. No signs of distension of the intestinal loops upstream were identified. Computed tomography of the upper abdomen and pelvis corroborated the previous ultrasonographic findings (Figure 1). Based on the image findings, a neoplasm was suspected as the primary cause of intestinal intussusception. An exploratory laparotomy with enterectomy and end-to-end enteroenteric anastomosis was performed. The collected material was sent for histopathological analysis.

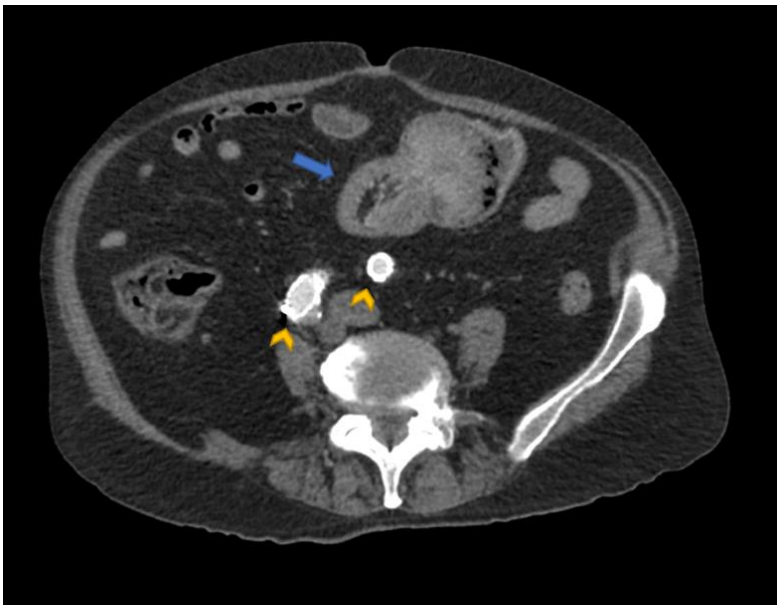

Figure 1. Contrast-enhanced arterial-phase and reformatted axial oblique computed tomography demonstrates small bowel intussusception, as well as fat and mesenteric vessels (blue arrow), at the level of the jejunoileal transition, in the left mesogastrium. As an additional finding, an aortobiiliac vascular prosthesis is observed at the level of the common iliac arteries (yellow arrowheads).

The anatomopathological findings showed a poorly differentiated, pleomorphic, and ulcerated epithelioid malignant neoplasm measuring 4.2 cm in its longest axis with the unifocal serosa mucosal invasion. The mesentery and local lymph nodes were infiltrated by the neoplasm. To complement the findings, tumor immunohistochemical analysis revealed positivity for HMB-45 and S-100 markers. Also, Fontana–Masson staining did not reveal any melanin pigment in the analyzed specimen. A final diagnosis of small bowel amelanotic melanoma was established.

An abdominal and pelvis MRI was performed for cancer staging, revealing hepatic metastasis (Figure 2). As the case reports a small bowel amelanotic melanoma, a mildly low signal on T1 was noticed. The patient also underwent a careful dermatological examination and no skin lesions were identified. The patient also had no previous history of skin lesions, nor oncological familial history. As no other site had neoplastic manifestations, the authors attributed the small bowel as a primary site.

The patient had a sudden impairment in performance status during hospitalization, evolving with neoplastic cachexia, deep vein thrombosis, symptomatic anemia, urinary tract infection, acute renal failure, and rapid disease progression. Thus, it was decided not to start any curative oncological therapy, and the patient was referred to palliative care. A few days later, the patient progressed to cardiorespiratory arrest and subsequent death.

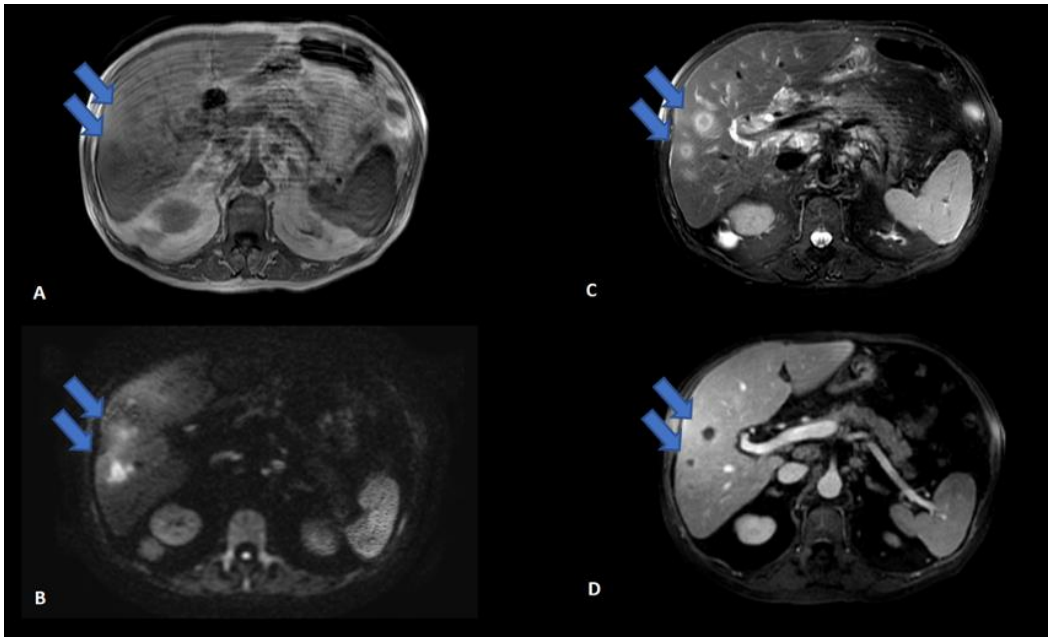

Figure 2. Magnetic resonance imaging sequences in T1 (A), T2 (B), DWI (C), and T1 post-gadolinium MRI in portal phase (D) revealed solid hepatic nodules (blue arrows), with mildly low signal on T1, predominantly high signal on peripheral T2, water diffusion restriction demonstrated on the diffusion-weighted sequence and confirmed on apparent diffusion coefficient map (not shown), and faint peripheral enhancement by the paramagnetic contrast agent.

3. Discussions

The PGM is an uncommon neoplasm affecting with major prevalence the anorectal area in 50% of cases (31.4% occur in the anal canal and 22.2% in the rectum) and the oropharyngeal in 32.8%. Other affected sites may include the esophagus (5.9%), stomach (2.7%), small bowel (2.3%), gallbladder (1.4%) and large bowel (0.5%) [10]. Most gastrointestinal tract melanomas correspond to metastatic lesions from cutaneous sites. As

reported in the present case, primary small bowel amelanotic melanoma accounts for less than 1% of all gastrointestinal tumors [3, 9, 11].

In the initial stages, PGM are usually asymptomatic. The diagnosis majorly occurs when the disease progresses and nonspecific symptoms (chronic abdominal pain, weight loss, and bleeding of the anorectal mucosa) appear [8]. Primary small bowel amelanotic melanoma remains a diagnostic challenge, due to the absence of specific symptoms, lack of precise clinical diagnosis criteria, and the need for a biopsy to define its diagnosis [12, 13].

Intestinal intussusception in the small bowel is a rare diagnostic finding. It can be secondary to intra- or extraluminal causes, dealing with numerous etiological diagnoses which could be inflammatory, oncological, structural, or iatrogenic. Lu (2015) and Honjo (2015) described malignant neoplastic lesions as the most common causes for the intussusception finding [14,15]. As occurred in the reported case, the intussusception sign evidenced by ultrasonography and confirmed by the tomography was fundamental for further investigation of a subjacent neoplasm. Similar reported cases of intestinal intussusception secondary to PGMs were also diagnosed only after thorough investigation with imaging methods, as both admission and prior symptoms were non-specific in both cases [16, 17].

Primary small bowel amelanotic melanomas are associated with a more aggressive behavior and a worse prognosis when compared to cutaneous melanomas. This finding is due to the rapid disease growth supported by an abundant vascular-lymphatic vessel network of the gastrointestinal mucosa [9, 13, 18]. In such a manner, the clinical evolution of the reported case was extremely fast, cursing with hepatic metastasis. Hence the advanced cancer stage and the significant decrease in patient's performance status, a curative oncological approach could not be performed in time.

There are only a few cases of PGM described in the medical literature, resulting in non-existing established standardized oncological reference therapy protocols. The current treatment of choice includes the extensive intestinal resection with free neoplastic margins associated with mesenteric resection and local lymphadenectomy [19,20]. Although its radical approach, surgical resection is the only potential curative therapy that may prolong patients' survival. The chemotherapy, immunotherapy and targeted therapy regimens are considered exclusively palliative therapeutic options [21].

The case reported in this study highlights the nonspecific presentation of primary small bowel amelanotic melanoma and reinforces the need to consider PGM as a differential diagnosis of intestinal intussusception. In this report, the definitive diagnosis was achieved through the association of intussusception images with the anatomopathological and immunohistochemical analysis of the collected material in the resolutive obstructive abdomen surgery.

4. Conclusions

Primary small bowel amelanotic melanoma is a rare condition with a challenging diagnosis and poor prognosis. There are limited studies and reports on this disease regarding a standardized reference therapy protocol. Extensive intestinal resection is the current treatment of choice. The present study emphasizes small bowel melanomas as differential diagnoses in adult patients presenting intestinal intussusception. An early neoplasm diagnosis may provide a better prognosis.

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