Primary malignant melanoma of small intestine – A rare case report

Palak Patel[1], Prashant Parikh[2], Bhavna Mehta[3], Rutvik Raval[4], Rahul Yadav[5], Sandip Shah[6], Mandava Rao[7]

From [1]Histopathologist, [2]Senior Histopathologist, [3]Head, Department of Histo-Pathology, [4]Junior Scientist, [5]Bioinformatician, Department of Genomic Medicine, [6]Lab Director, [7]Research Director, Neuberg-supratech Referral Laboratory, Ahmedabad, Gujarat, India

Correspondence to: Mandava Rao, Neuberg-supratech Referral Laboratory, Paldi, Ahmedabad - 380 006, Gujarat, India. E-mail: manvrao@gmail.com

Received - 06 November 2020 Initial Review - 16 November 2020 Accepted - 25 November 2020

ABSTRACT

Primary malignant melanoma of the intestine is an extremely rare lesion that may arise from large bowel. Hereby, we report a case of a 35-year-old female who presented with abdominal pain of 1 month duration. On positron emission tomography-computed tomography, a hypermetabolic mass observed in the pelvic ileal loop with no lesions elsewhere in the body. The surgically excised specimen was diagnosed as primary small intestinal melanoma by histopathological examination, which was further confirmed by the presence of melanoma markers, that is, Melan-A, HMB45, and S100 protein.

Key words: *Immunochemistry, Positron emission tomography-computed tomography, Primary melanoma, Small intestine*

Majority of the malignant melanomas in the small intestine are usually metastases from primary cutaneous lesions; however, it can also develop as a primary mucosal tumor in the gastrointestinal (GI) tract [1]. Distinguishing metastatic melanoma of the GI tract from primary melanomas and other primary lesions can be very challenging. Hereby, we present a rare case of primary malignant melanoma of the small intestine along with review of the relevant literature for diagnosis and management of the same.

CASE REPORT

A 35-year-old female presented with abdominal pain of 1 month duration. A CT enterography was performed because of repeated colicky abdominal pain, it revealed the presence of a solid tumor possibly originating in the distal ileum. In the whole body positron emission tomography-computed tomography (PET-CT), a hypermetabolic mass in the pelvic ileal loop was observed; however, there was no indication of cutaneous, retinal, or anal primary lesions. Exploratory laparotomy revealed an intraluminal mass in the distal ileum. Careful examination of the abdominal cavity revealed no macroscopic evidence of metastases. Small bowel resection was performed with side-to-side anastomosis. The post-operative course was uneventful, and the patient was discharged on post-operative day 6. Surgical specimen was 18 cm in length and included a 6 cm × 5 cm mass, the resection margins retrieved seven lymph nodes. Histopathology showed melanoma condition and was free from tumor invasion (Fig. 1). Immunohistochemistry revealed the presence of tumor cells that were positive for the melanoma markers such as Melan-A and HMB45 as well as S100 protein (Fig. 2a and b).

After the diagnosis of melanoma was established, the patient underwent a complete clinical and laboratory evaluation. Since the examination of skin, eyes, esophagus, and anus was negative for primary melanoma and the investigation with chest CT, brain magnetic resonance imaging, and PET-CT scan did not report metastatic disease, a final diagnosis of primary melanoma of the small intestine was considered. The patient was scheduled for follow-up at regular intervals every 3 months for the 1st year and every 6 months for up to 5 years. At each follow-up, physical examination and an abdominopelvic CT scan were obtained. During regular follow-up, there was no recurrence or metastases. No adjuvant or alternative therapy was used.

DISCUSSION

Primary mucosal melanoma can arise at any site within the GI mucosa, but it is most common in anorectal (anal canal – 31.4%; rectum – 22.2%) and oropharyngeal (32.8%) regions, whereas esophagus (5.9%), stomach (2.7%), small intestine (2.3%), gallbladder (1.4%), and large intestine (0.9%) are extremely rare sites of origin [2]. The presence of melanocytes has not yet been demonstrated in the small intestine, and the origin of primary melanoma of the small intestine remains unknown. One potential origin of the primary melanoma of small intestine is melanoblastic cells of the neural crest that migrates to the distal ileum through the omphalomesenteric canal. Accordingly, the ileum, which represents the distal end of the omphalomesenteric canal, should be the most common site of primary malignant melanoma within the small intestine [3]. Another hypothesis suggests that these tumors originate from the enteric neuroendocrine non-cutaneous tissue, in the form of amine precursor uptake decarboxylase

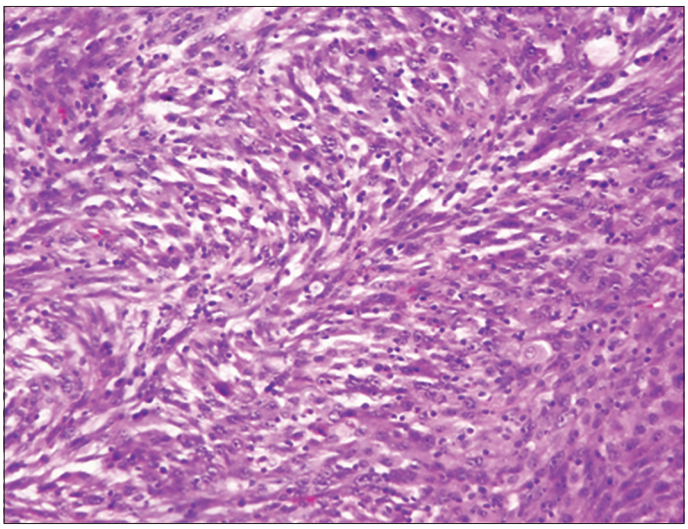

Figure 1: Small intestinal melanoma with characteristic cell types ×400

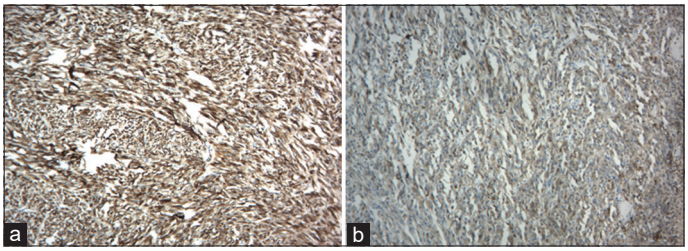

Figure 2: (a) Melanoma marker – HMB45 in the small intestine ×400. (b) Melanoma marker – S100 in the small intestine ×400

cells that have undergone neoplastic transformation. This would also account for the remaining non-ileal intestinal malignant melanomas [3].

A primary GI mucosal melanoma is considered in patients with no obvious primary cutaneous melanoma or those with an isolated GI lesion in the absence of other extraintestinal metastases. Blecker *et al*. [1] suggested the following criteria for the diagnosis of primary intestinal melanoma: No evidence of concurrent melanoma or atypical melanocytic lesion of the skin, absence of extraintestinal metastatic spread of melanoma, and presence of intramucosal lesions in the overlying or adjacent intestinal epithelium. In our study, histopathology of post-operative showed melanoma condition of the intestine. It is further supported by immunochemistry performed using tumor markers, such as Melan-A, HMB45, and S100 protein. Since the no evidence of tumor was observed elsewhere in the body, primary intestinal melanoma is considered.

Prognosis is worse for primary intestinal melanomas which tend to grow faster and more aggressively. Surgery is the main treatment option for primary melanoma of the small intestine and should include excision of the intestine with tumor-free margins and of mesentery to remove regional lymph nodes. In these patients, systemic adjuvant therapy has a limited role, and chemotherapy regimens have very low response rates [3]. Recently, some articles reported that immune checkpoint inhibitor (PD-1 antibody and/or CTLA-4 antibody provide unprecedented efficacy gains in metastatic or advanced melanoma [4]. However, the effect of immunotherapy as an adjuvant treatment on mucosal melanoma has not been proved yet.

CONCLUSION

A primary small bowel melanoma is an extremely rare neoplasm. A definite diagnosis can only be made after a thorough investigation, to exclude the coexistence of a primary lesion. Curative resection of the tumor remains the treatment of choice.

ACKNOWLEDGMENT

We are thankful to staff of the Neuberg-Supratech Referral Laboratory, Paldi, Ahmedabad-380006, India.

REFERENCES

1. Blecker D, Abraham S, Furth EE, *et al*. Melanoma in the gastrointestinal tract. Am J Gastroenterol 1999;94:3427-33.
2. Cheung MC, Perez EA, Molina MA, *et al*. Defining the role of surgery for primary gastrointestinal tract melanoma. J Gastrointest Surg 2008;12:731-8.
3. Lens M, Bataille V, Krivokapic Z. Melanoma of the small intestine. Lancet Oncol 2009;10:516-21.
4. Byrne EH, Fisher DE. Immune and molecular correlates in melanoma treated with immune checkpoint blockade. Cancer 2017;123:2143-53.

Funding: None; Conflict of Interest: None Stated.

How to cite this article: Patel P, Parikh P, Mehta B, Raval R, Yadav R, Shah S, Rao M. Primary malignant melanoma of small intestine – A rare case report. East J Med Sci. 2020;5(4):91-92.

Doi: 10.32677/EJMS.2020.v05.i04.006