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Case Report

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Malignant spindle cell tumor of the anal canal, what are the differential diagnoses?

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Abstract

Malignant peripheral nerve sheath tumor (MPNST) is an uncommon tumor. Here, we report a case of rectal MPNST. The patient was a 75 year old man with symptoms and signs of a mass protruded from anus. There was no evidence of neurofibromatosis type 1 in him or his family. Primary diagnosis was hemorrhoid. Biopsy was taken. The microscopic examination revealed spindle cells with large and hyperchromatic nuclei. Mitotic figures were numerous. Routine and ancillary techniques confirmed the diagnosis of MPNST. MPNST should be distinguished from more common lesions encountered at this anatomic site such as hemorrhoid, malignant melanoma, sarcomatoid carcinoma, and gastrointestinal stromal tumor.

Keywords: Malignant Peripheral Nerve Sheath Tumor, Rectum, Neurofibromatosis Type 1.

INTRODUCTION

Colonoscopy is used to diagnose lower gastrointestinal tract lesions. But this method is limited in diagnosing of submucosal masses. Colonic submucosal tumors are rare compared to adenomas and carcinomas. Colorectal submucosal tumors are uncommon and are challenging to diagnose and treat. There is a wide range of benign and malignant submucosal tumors in the rectum. The more common malignant ones include GISTs, leiomyosarcomas and malignant schwannomas. Malignant peripheral nerve sheath tumor (MPNST) accounts for about 5–10% of soft tissue sarcomas. Quarters to half of MPNSTs are seen in patients with neurofibromatosis type 1 [1]. The World Health Organization has identified MPNST as a tumor that originates from the peripheral nerve or a tumor that shows nerve sheath differentiation. Although the tumor is mainly found in the soft tissues of the limbs or retroperitoneum, it has been reported rarely in the large intestine [2-5]. Here we present a case of malignant peripheral nerve sheath tumor (MPNST) of the rectum in a 75 year old man who was not shown any stigmata of neurofibromatosis or parasite infection.

CASE REPORT

A 75 year old man referred to our surgical ward with chief complaint of protruding a mass from his rectum during defecation since several months ago. He denied loss of appetite or weight. He also complained of occasional bleeding from the anus. On physical examination he was an alert conscious elderly man. There was no evidence of stigmata of neurofibromatosis.

Digital PR (prostate rectum) examination revealed a polypoid skin covered mass that was protruded from anal verge. Prostate was mildly enlarged.

Laboratory studies including CEA, CA 19-9, blood routine examination, urea and serum biochemistry analysis were unremarkable except for a low level of hemoglobin of 11 g/L. Since the primary diagnosis was hemorrhoid, he underwent surgical excision of the mentioned polypoid lesion and colonoscopy was not performed. The specimen sent to pathology ward. The histopathological examination of H&E stained slides revealed a tumoral tissue composed of cellular fascicles made of spindle cells (Figure 1). These cells had hyperchrome, pleomorphic nuclei and scant eosinophilic cytoplasm. Mitotic figures were numerous. According to the above microscopic findings and the anatomic site the possibility of sarcomatoid carcinoma, malignant melanoma, GIST, leiomyosarcoma and other spindle cell sarcomas should had to be considered. The IHC panel was designed based on these differential diagnoses. On the immunohistochemical method the tumor cells were negative for panCK, HMB45, CD117, CD34, desmin,

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alpha smooth muscle actin while weakly and focally were positive for S100 (Figure 2). Ki 67 was positive in 30% of the tumor cells. Taken together the diagnosis of MPNST was established. The patient received radiation therapy and he is well now.

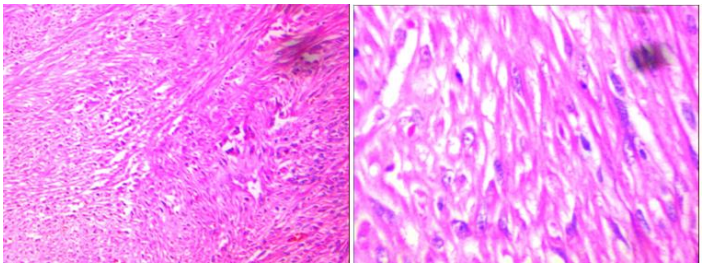

Figure 1: H&E stained slides reveales a tumoral tissue composed of cellular fassicles made of spindle cells. These cells have hyperchrome, pleomorphic nuclei and scant eosinophilic cytoplasm (X10, X40)

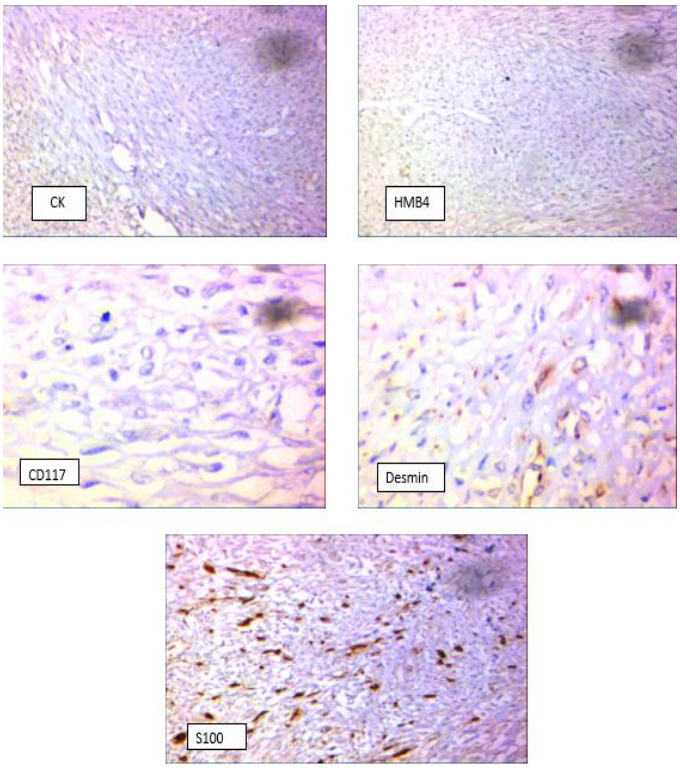

Figure 2: The tumor cells are negative for panCK, HMB45, CD117 and desmin while weakly and focally are positive for S100

RESULTS

In the present study, 118 medical students (interns) were examined. The frequency distribution of individuals based on indigenousness, age, and sex. The average score of interns' satisfaction with the process of clinical education was (26.36±6.67), which was in the average range according to the division of satisfaction (mentioned in the method section).

Most interns (41.5%), were moderately satisfied with the overall clinical education process (27.1%), had high satisfaction and 31.4% had low satisfaction. The highest level of satisfaction of medical interns was with the training on how to insert NG-tube (46.6%).

DISCUSSION

MPNST is an uncommon malignant tumor which originates from the Schwann cells [6]. MPNST of the gut is actually unusual. The disease has no gender preference and is seen in a wide range of ages [7]. However, sporadic cases are more common in patients over the age of 35 [8]. Our patient was a 75 year old man. The results of one study showed that among gastrointestinal MPNST there was just one case of MPNST in the rectum [9]. Tumors vary in size from small to quite large, causing intestinal obstruction. The most common clinical sign is due to the presence of a mass [10]. In approach to a patient with a polypoid mass protruding from the anus, in addition to benign lesions such as hemorrhoid, malignant lesions should be considered. About this patient the differential diagnoses were sarcomatoid carcinoma, malignant melanoma, GIST, leiomyosarcoma and other spindle cell sarcomas and the IHC panel was designed. Result of a study revealed that most GISTs in large intestine were positive for CD117 and CD34. None of them were positive for desmin and S100. They also found that colonic leiomyosarcoma didn't show reactivity for CD34 and CD117 but were positive for desmin and alpha smooth muscle actin. Malignant melanoma was ruled out by negative result for HMB45. Negative pancytokeratin was against sarcomatoid carcinoma. There is no specific marker for diagnosis of MPNST, however focal reactivity for S100 is helpful [11]. Preoperative diagnosis is difficult or sometimes impossible. Correct diagnosis is based on histological findings and ancillary techniques. Mention should be made that about 50% of MPNSTs occur in the background of NF1 [12]. Another predisposing factor is previous irradiation [11]. It was not true about the current case. Standard treatment is surgery with negative margin [13]. Radiotherapy is applied for high grade tumor or in the case of microscopic residual disease. Overall, radiation therapy is not useful in gastrointestinal MPNSTs. The prognosis is difficult to predict. But in general, the prognosis is not good and the tumor is resistant to chemotherapy and radiotherapy [14]. Results of one study revealed that location, size, margin status, stage, IHC result for S100, Ki67, P53 and MDM2 expression were indicator of the possibility of regional relapse and distant metastasis [2].

CONCLUSION

The rectal MPNST is rare. It should be distinguished from more common lesions encountered at this anatomic site such as hemorrhoid, malignant melanoma, sarcomatoid carcinoma, and gastrointestinal stromal tumor.

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Conflict of Interest

None declared.

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REFERENCES

1. Jo VY, Fletcher CD. Epithelioid malignant peripheral nerve sheath tumor. Am J Surg Pathol. 2015;39:673-82.
2. McCarthy AJ, Karamchandani DM, Chetty R. Neural and neurogenic tumours of the gastroenteropancreaticobiliary tract. J Clin Pathol. 2018;71:565-78.
3. Rawal G, Zaheer S, Ahluwalia C, Dhawan I. Malignant peripheral nerve sheath tumor of the transverse colon with peritoneal metastasis: a case report. J Med Case Reports. 2019;13:1-5.
4. Zhu L-B, Li P-F, Xiao W-H, Zhang P-B, Li J-Q, Sun M-F, *et al*. A distal ileum malignant peripheral nerve sheath tumor causing intussusception in a patient in China: a case report. World J Surg Oncol. 2017;15:29.
5. Kim EY, Lee SH, Yoo HM, Song KY, Park CH. Gastric malignant peripheral nerve sheath tumor: A case report. Int J Surg Pathol. 2015; 23:505-08.
6. Ducatman BS, Scheithauer BW, Piepgras DG, Reiman HM, Ilstrup DM. Malignant peripheral nerve sheath tumors. A clinicopathologic study of 120 cases. Cancer. 1986;57:2006-21.

7. Trojanowski J, Kleinman G, Proppe K. Malignant tumors of nerve sheath origin. Cancer. 1980;46:1202-12.
8. Wong WW, Hirose T, Scheithauer BW, Schild SE, Gunderson LL. Malignant peripheral nerve sheath tumor: analysis of treatment outcome. Int J Radiat Oncol Biol Phys. 1998;42:351-60.
9. Patra S, Ayyanar P, Padhi S, Purkait S, Muduly DK, Samal SC, *et al*. Epithelioid Malignant Peripheral Nerve Sheath Tumor (Epithelioid-MPNST) Presenting as Bleeding Rectal Polyp: A Case Report with Systematic Literature Review. Am J Case Rep. 2019;20:1175.
10. Maneschg C, Rogatsch H, Bartsch G, Stenzl A. Treatment of giant ancient pelvic schwannoma. Tech Urol. 2001;7:296.
11. Perrin RG, Guha A. Malignant peripheral nerve sheath tumors. Neurosurg Clin. 2004;15:203-16.
12. Neville H, Corpron C, Blakely ML, Andrassy R. Pediatric neurofibrosarcoma. J Pediatr Surg. 2003;38:343-46.
13. Choudry HA, Nikfarjam M, Liang JJ, Kimchi ET, Conter R, Gusani NJ, *et al*. Diagnosis and management of retroperitoneal ancient schwannomas. World J Surg Oncol. 2009;7:1-5.
14. Li Q, Gao C, Juzi JT, Hao X. Analysis of 82 cases of retroperitoneal schwannoma. ANZ J Surg. 2007;77:237-40.