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Case Report

Rectal Gastrointestinal Stromal Tumors: Usefulness of Endoscopic Ultrasound

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Abstract

Rectal gastrointestinal stromal tumors (GIST) account for 0.1% of all rectal tumors. They are generally diagnosed in adults over 50 years of age with non-specific symptoms. Immunohistochemistry performed on biopsy tissue sample confirms the diagnosis. In this study, we report two cases of histologically and immunohistochemically confirmed rectal GISTs diagnosed using Endoscopic ultrasound (EUS) with fine needle aspiration (FNA), a procedure of crucial value in the diagnosis of these tumors. Both patients were male, presenting with non-specific symptoms. Standard endoscopy showed a rectal mass in both cases, biopsies were negative. EUS showed a well-defined hypoechoic mass and evaluated adjacent organs and of blood/lymphatic nodes involvement. EUS-FNA was performed in both cases. Cytology and immunocytochemistry studies showed spindle cell pattern with positive immunohistochemical staining for CD117, typical of GISTs.

Keywords: Rectal gastrointestinal stromal tumor (GIST), Rectal tumor, Endoscopic ultrasound, Fine needle aspiration, Case report.

INTRODUCTION

GISTs are neoplasms of mesenchymal origin rarely located in the rectum, making up about 0.1% of all tumors arising in the rectum and accounting for about 2% of all GISTs, with an estimated incidence rate of 0.45 per million individuals [1, 2].

EUS is an important tool for rectal GIST diagnosis, by determining the wall layer of origin and assessing malignity features [3]. The addition of EUS-guided biopsy allows tissue acquisition for histopathological confirmation.

We report two new cases of rectal GISTs, detailing the clinicopathological and endoscopic features of this tumors.

CASE PRESENTATION

Case 1

A 60-year-old male, with no relevant past medical or surgical history, complained of lower abdominal pain located in the hypogastric region and constipation. Clinical examination was unremarkable and digital rectal exam (DRE) was normal. Colonoscopy identified an ulcerated mass measuring 20 mm in diameter, located about 12 cm from the anal verge. EUS showed a well-defined hypoechoic heterogenous mass with central calcifications located in the middle rectum, measuring 36,6* 25 mm, originating from the muscularis propria, compressing the bladder and coming in contact with the distal ileum (Fig 1 & 2). EUS-FNA has been successfully performed using a 19-gauge (G) needle, with 2 passes. The cytopathological analysis showed spindle cells containing ovoid nucleus with fine chromatin. Immunohistochemical staining showed positive expression for CD117 and Dog1, consistent with a gastrointestinal stromal tumor (Fig 3).

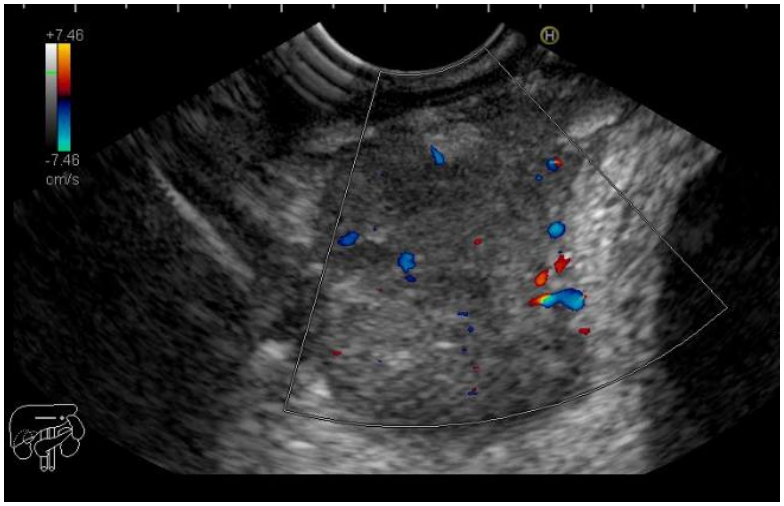

Figure 1: EUS showed a well-defined heterogeneous slightly echogenic solid mass with central calcifications and no significant flow on color Doppler

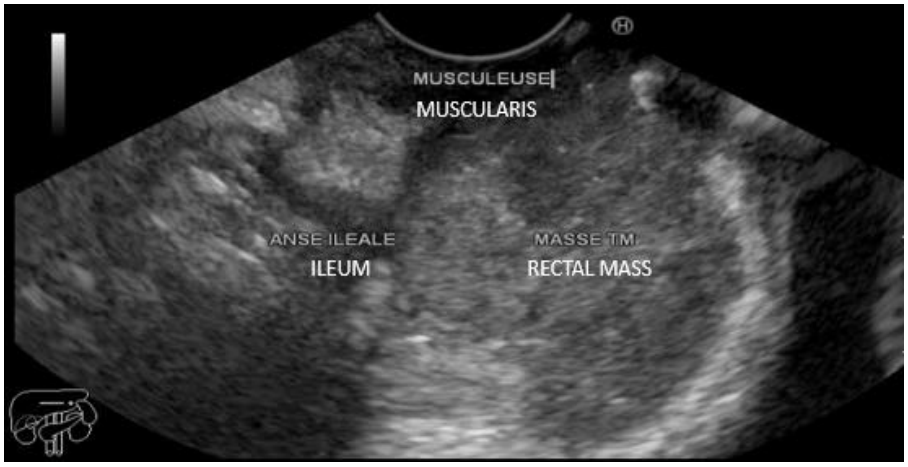

Figure 2: EUS revealed contact between the rectal mass and the distal ileum

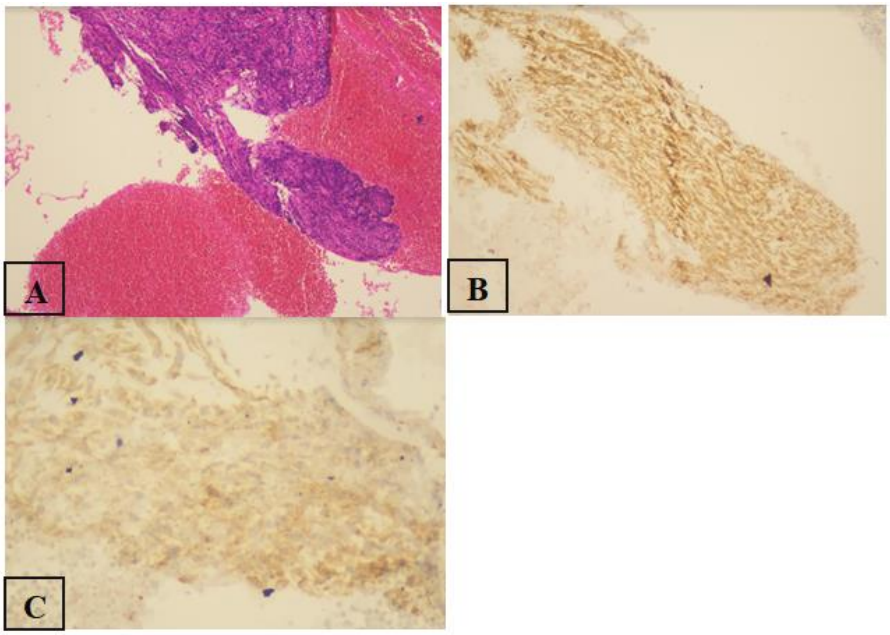

Figure 3: Cytological and immunohistochemical findings. Cell blocks showing spindle cells containing ovoid nucleus with fine chromatin x (A, hematoxylin and eosin [H&E], x 100), week nuclear staining for dog1(B x400) and positive staining for CD117 in tumor cells (C x400)

Case 2

A 49-year-old male, presented with abdominal pain and cramping, tenesmus, and incomplete stool evacuation stool. DRE found a palpable indurated mass rectal mass 5 cm from the anal verge. Flexible sigmoidoscopy showed an ulcerative budding mass extending from 5 cm from the anal verge to 25cm, repeated biopsies were negative.

EUS revealed a noncircumferential submucosal solid hypoechoic mass invading the mucosa, located in the lower rectum extending to 25 cm from the anal margin, where it became stenosing. The rectal sphincters were not invaded, multiples perirectal lymph nodes were detected.

EUS-FNA performed using a 19-G needle with 3 passes. Cytology and immunocytochemistry studies showed spindle cell pattern with positive Immunohistochemical staining for CD117, typical of GISTs.

DISCUSSION

Rectal GISTs generally occur in middle-aged individuals between the fifth and sixth decades, predominantly in males [4]. In our study both patients were male with a mean age of 54 years, in line with the pathological literature.

GISTs probably originate from the interstitial cells of Cajal, are spindly-shaped (70% of cases) and typically defined as CD 117/KIT and/or DOG1 positive, making immunohistochemical staining for these markers an important diagnostic tool [5].

Rectal GISTs have a worse prognosis than other gastro-intestinal locations and high local recurrence rate [2]. After rectal GIST resection, the most common cause of death GIST is distant metastasis, with a frequent involvement of the liver [1].

Clinically, the reported symptoms of rectal GISTs are rectal bleeding, tenesmus, abdominal pain with painful contractions, abnormal evacuation (mucus, pus, etc.), transit disorders, urinary complaints or altered general condition [6-8]. In this study, the most frequent symptoms were abdominal pain and constipation.

Rectal GISTs are frequently found incidentally, either on cross-sectional imaging, screening colonoscopy, or clinical examination. Low-lying GISTs may be felt as a smooth, firm mass on physical examination [2]. In our study, both patients were symptomatic and physical examination identified a rectal mass in one case.

Computed tomography and magnetic resonance imaging are useful imaging methods for detection and staging of these neoplasm [2, 9]. Rectal GISTs appear as large, bulky, exophytic rectal masses with heterogenous enhancement. The presence of features such as well-demarcated margins, prominent extraluminal location and no surrounding adenopathy, and lack of bowel lumen constriction despite the large size of the tumor, allow GISTs to be differentiated from malignant epithelial neoplasms [8].

Endoscopic examinations are key in rectal GIST's diagnosis. GISTs can have an exophytic and/or intramural pattern of growth. The main endoscopic finding is a round smooth mass covered with normal mucosa in endoluminal tumors, and a stiffening of the rectal wall in exophytic forms [10]. Other findings on endoscopy such as Irregular borders, ulceration, and growth during endoscopic follow-up are considered clinically malignant features [11].

Conventional biopsy by endoscopy is frequently negative since biopsy sample provides only mucosal tissue [10, 12] the Reported diagnostic yield of this approach is poor, ranging from 17 to 42% [12].

In this context, EUS is an invaluable tool for the diagnosis, by determining the wall layer of the origin and assessing its echogenicity [3]. Since rectal GISTs typically arise within the muscularis propria [8].

The typical EUS features of a GIST is a hypoechoic, round to oval, homogeneous mass, with well-defined contours. GISTs are seen as fourth layer tumors in EUS. Large or clinically malignant GISTs can display irregular margins [12]. EUS finding such as irregular shape, heterogeneous echotexture, central necrosis, echogenic foci or calcification are associated with high-risk GISTs [13].

Contrast-enhanced harmonic (CH) imaging of EUS is a useful tool in differentiating GISTs from benign SELs and evaluating the malignancy potential of GISTs. Hyper-enhancement pattern on CH-EUS has an accuracy ranging from 82.2 to 100% in the diagnosis of GISTs [3].

CH-EUS allows the visualization of the intratumoral microvascularity to estimate the malignancy potential of GISTs [14]. CH-EUS findings predicting high-grade malignancy are: irregular and abundant intra-tumor blood vessels , heterogeneous enhancement patterns and the existence of non-enhancing spots [3].

The principal differential diagnoses of GISTs by imaging methods and EUS are schwannoma, leiomyoma, leiomyosarcoma [14]. Thus, tissue sampling is required for a conclusive diagnosis.

EUS-FNA is an effective tissue sampling method for the diagnosis of submucosal lesions [12],

with a diagnostic accuracy rate ranging from 70 to 90% [11].

For the diagnosis of GISTs, the sensitivity of EUS-FNA cytology was 78.4%, influenced by size, location, shape, and layer of origin in a study by Sepe PS and al [15]. However, despite EUS-FNA good accuracy in identifying spindle cells and mitotic activity, it has a limited ability to procure tissue for immunohistochemistry, which is a critical element in establishing the diagnosis and prognosis of GISTs [16].

EUS-FNA has an inconsistent diagnostic yield in different studies, ranging from 46% to 93% [17]. The non-diagnostic FNA-specimens in GISTs, can leads to a lack of prognostic information based on the tumor mutation profile and proliferation rate, making it an obstacle for the early personalized management of GISTs [18].

Core needles for EUS-guided fine-needle biopsy (EUS-FNB) emerged to overcome the limitations of EUS-FNA and optimize the diagnostic yield of lesions such as GISTs [17].

In a retrospective large multicenter study, FNB for suspected GISTs was superior to EUS-FNA in establishing the diagnosis of GISTs, with a diagnostic yield for FNB more than double that of FNA, making FBN an effective method in preventing repeated EUS procedures or unnecessary surgery on benign lesions discovered [17]. In our study, EUS-FNA was the mainstay of obtaining tissue and allowed an accurate pathological diagnosis of the tissue specimen in both cases of rectal GISTs.

CONCLUSION

EUS is a crucial technique in the diagnosis of rectal GISTs. It identifies the affected rectal layer, assesses the tumor size, and evaluates the involvement of adjacent organs and blood/lymphatic vessels for tumor staging. The addition of FNA or FNB to EUS enables tissue analysis for diagnosis confirmation and high-risk features identification, thus helping in the guidance for the best therapeutic management.

REFERENCES

1. Hamada, M., Ozaki, K., Horimi, T., Tsuji, A., Nasu, Y., Iwata, J., & Nagata, Y. (2008). Recurrent rectal GIST resected successfully after preoperative chemotherapy with imatinib mesylate. *International Journal of Clinical Oncology*, *13*(4), 355-360.
2. Kane, W. J., & Friel, C. M. (2019). Diagnosis and treatment of rectal gastrointestinal stromal tumors. *Diseases of the Colon & Rectum*, *62*(5), 537-540.
3. Tamura, T., & Kitano, M. (2019). Contrast enhanced endoscopic ultrasound imaging for gastrointestinal subepithelial tumors. *Clinical Endoscopy*, *52*(4), 306-313.
4. Miettinen, M., Furlong, M., Sarlomo-Rikala, M., Burke, A., Sobin, L. H., & Lasota, J. (2001). Gastrointestinal stromal tumors, intramural leiomyomas, and leiomyosarcomas in the rectum and anus: a clinicopathologic, immunohistochemical, and molecular genetic study of 144 cases. *The American journal of surgical pathology*, *25*(9), 1121-1133.
5. Lee, S. J., Hwang, C. S., Kim, A., Kim, K., & Choi, K. U. (2016). Gastrointestinal tract spindle cell tumors with interstitial cells of Cajal: prevalence excluding gastrointestinal stromal tumors. *Oncology Letters*, *12*(2), 1287-1292.
6. Rejab, H., Kridis, W. B., Ameur, H. B., Feki, J., Frikha, M., & Beyrouti, M. I. (2014). Tumeur stromale rectale: à propos d'une observation. *Pan Afr Med J*, 17, 119.
7. Jallouli, A., Jarti, M., & Haida, M. (2021). Rectal Gastrointestinal Stromal Tumor (GIST): About a Rare and Unusual Case. *Int J Innov Research Med Science*, 6(11), 814-817.
8. Jiang, Z. X., Zhang, S. J., Peng, W. J., & Yu, B. H. (2013). Rectal gastrointestinal stromal tumors: imaging features with clinical and pathological correlation. *World Journal of Gastroenterology: WJG*, *19*(20), 3108-3116.
9. Puckett, Y., & Aryaie, A. (2017). Case report of diffusely metastatic rectal GIST. *International journal of surgery case reports*, *37*, 4-9.
10. Samlani-Sebbane, Z., Diffaa, A., Charaf, K., Rabbani, K., Narjis, Y., El Mansouri, F., ... & El Idrissi Dafali, A. (2011). Le GIST rectal: à propos de deux observations et une revue de la littérature. *Journal Africain d'Hépato-Gastroentérologie*, *5*(1), 60-62.
11. Nishida, T., Kawai, N., Yamaguchi, S., & Nishida, Y. (2013). Submucosal tumors: comprehensive guide for the diagnosis and therapy of gastrointestinal submucosal tumors. *Digestive Endoscopy*, *25*(5), 479-489.
12. Sepe, P. S., & Brugge, W. R. (2009). A guide for the diagnosis and management of gastrointestinal stromal cell tumors. *Nature reviews Gastroenterology & hepatology*, *6*(6), 363-371.
13. Kang, J. H., Lim, J. S., Kim, J. H., Hyung, W. J., Chung, Y. E., Choi, J. Y., ... & Kim, K. W. (2009). Role of EUS and MDCT in the diagnosis of gastric submucosal tumors according to the revised pathologic concept of gastrointestinal stromal tumors. *European radiology*, *19*(4), 924-934.
14. Sakamoto, H., Kitano, M., & Kudo, M. (2010). Diagnosis of subepithelial tumors in the upper gastrointestinal tract by endoscopic ultrasonography. *World Journal of Radiology*, *2*(8), 289-297.
15. Sepe, P. S., Moparty, B., Pitman, M. B., Saltzman, J. R., & Brugge, W. R. (2009). EUS-guided FNA for the diagnosis of GI stromal cell tumors:

sensitivity and cytologic yield. *Gastrointestinal endoscopy*, *70*(2), 254-261.

16. Obuch, J., & Wani, S. (2017). EUS–guided tissue acquisition in GI stromal tumors. *Gastrointestinal Endoscopy*, *86*(3), 516-518.
17. Trindade, A. J., Benias, P. C., Alshelleh, M., Bazarbashi, A. N., Tharian, B., Inamdar, S., ... & Ryou, M. (2019). Fine-needle biopsy is superior to fine-needle aspiration of suspected gastrointestinal stromal tumors: a large multicenter study. *Endoscopy International Open*, *7*(07), E931-E936.
18. Hedenström, P., Nilsson, B., Demir, A., Andersson, C., Enlund, F., Nilsson, O., & Sadik, R. (2017). Characterizing gastrointestinal stromal tumors and evaluating neoadjuvant imatinib by sequencing of endoscopic ultrasound-biopsies. *World Journal of Gastroenterology*, *23*(32), 5925-5935.