



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

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 malignancy 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 tumor.

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 mm × 25 mm, originating from the muscularis propria, compressing the bladder and coming in contact with the distal ileum (**Figures 1 and 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 (**Figure 3**).

Case 2

A 49 year old male, presented with abdominal pain and cramp-

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ing, 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 25 cm, repeated biopsies were negative.

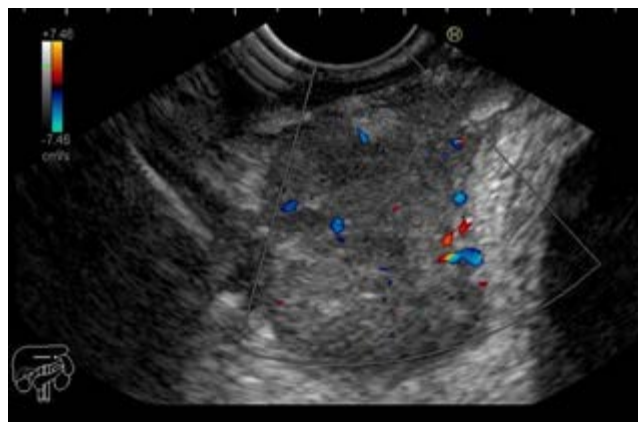


Figure 1: EUS showed a well-defined heterogeneous slightly echogen-

ic 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

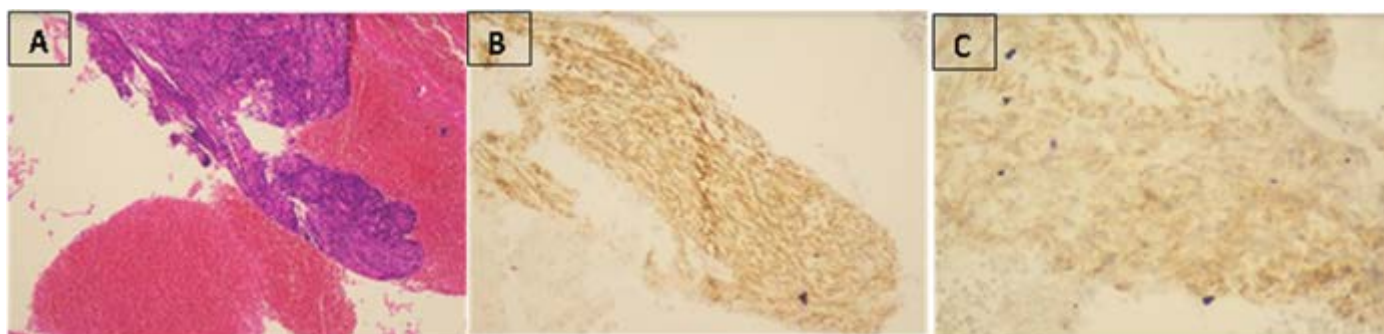


Figure 3: Cytological and immunohistochemical findings. Cell blocks showing spindle cells containing ovoid nucleus with fine chromatin x (A, hematoxylin and eosin [H and E], x 100), weak nuclear staining for dog1 (B x 400) and positive staining for CD117 in tumor cells (C x 400),

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 are 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 micro vascularity 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 FNB 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. In our case report, EUS helped the diagnosis of two case of rectal Gists by providing tissue sampling for cytological and immunohistochemical analysis.

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