Case Report

Gastrointestinal stromal tumor of rectum diagnosed by three-dimensional anorectal ultrasound

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ABSTRACT

Gastrointestinal stromal tumors (GIST) are relatively rare lesions of mesenchymal origin, being more frequent in the stomach and small intestine. These are clinically asymptomatic lesions, and in advanced stages may present with nausea, vomiting, bleeding, abdominal pain, a palpable mass, and even intestinal obstruction. The only effective treatment consists of a complete tumor resection. We report two cases of GIST located in the distal rectum and evaluated with three-dimensional anorectal ultrasonography, a procedure of great value in identifying the size of the lesion, its involvement toward nearby structures and lymph node invasion.

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Tumor estromal gastrointestinal de reto diagnosticado por ultra-sonografia anorretal tridimensional

RESUMO

Os tumores estromais do trato gastrointestinal (GIST) são lesões relativamente raras de origem mesenquimal, sendo mais frequentes no estômago e intestino delgado. Clinicamente, são lesões assintomáticas e em estados avançados podem cursar com náusea, vômito, sangramento, dor abdominal, massa palpável e até obstrução intestinal. O único tratamento efetivo é a ressecção completa do tumor. Relatamos dois casos de GIST...
Gastrointestinal stromal tumors (GIST) are relatively rare lesions. They are characterized by having a mesenchymal origin, which may be present from the distal half of the esophagus to the anorectal region. Its most frequent location is the stomach, with rates of 50–70%, followed by the small intestine (25–35%), colon and rectum (5–10%), and esophagus (<5%).1–3

With an incidence estimated at 10–20 cases/10 million inhabitants,4 GIST represents about 0.6% of all malignant rectal tumors,5 comprising only 1–3% of all cancers of the gastrointestinal tract. Its relevance stems from the fact that they represent 80% of all primary tumors of the gastrointestinal tract.6,7 The largest number of cases occurs in older adults, especially between the fourth to sixth decade of life.8 There is no prevalence in gender.9

Clinical manifestations of GIST are related to its location, which can be multiple, and its dimensions.5 Clinically, these are asymptomatic lesions, and in an advanced state, there may be nausea, vomiting, bleeding, abdominal pain, a palpable mass, and even intestinal obstruction.9–11 Tests such as abdominal ultrasound, upper gastrointestinal (UGI) endoscopy, colonoscopy, echoendoscopy, computed tomography (CT) and nuclear magnetic resonance (NMR) are imaging methods important for the investigation of injury.9 The only effective treatment is surgical resection.10 Anorectal ultrasonography (USG-A) is indicated in benign and malignant anorectal diseases, including anal incontinence, infectious and inflammatory diseases (abscesses, anorectal fistulae), chronic anorectal pain (endometriosis), and in the staging and follow-up of anorectal neoplasms.12–14 Thus, USG-A allows for the evaluation of anatomical structures that form the anal canal, rectum and perianorectal tissues with low cost and in a safe manner, because this is a very little invasive test, being well tolerated and which does not expose patients to radiation. Thus, USG-A has become a critical imaging modality for clinical diagnosis of GIST and its differentiation from other submucosal neoplasms.

The objective of this study is to report two cases of GIST diagnosed by three-dimensional anorectal ultrasonography (USG-A 3D), confirmed by histopathological and immunohistochemical studies.

Case report 1

Male patient, 57, industrial worker, previously healthy, attended the Coloproctology Clinic of Gastroclinica Cascavel to perform colonoscopy as part of a medical check-up. During symptomatology questioning, the patient did not report complaints or comorbidities. The digital rectal examination revealed an elevated, hardened, well-defined, painless lesion, fixed on the anterior rectal wall, without rectal mucosa changes. The lesion was located about 5 cm from the anal margin and measured 4 cm in diameter. The patient underwent colonoscopy and USG-A (Fig. 1). Local transanal excision was indicated. The patient was hospitalized on the procedure day, and a rectal cleaning with fleet enema was carried out 2 h before the procedure. The surgical procedure was uneventfully carried out, evolving on the first postoperative day without complaints; the patient was discharged in good condition, and has been followed-up at our coloproctology outpatient clinic, being asymptomatic for 3 years.

Case report 2

Female patient, 58, complained of a rectal lump which she noticed when passing a vaginal cream. Denied pain or bleeding. On physical examination, a hardened lump was observed at the rectovaginal septum. The patient underwent colonoscopy (Fig. 2A), and the examination showed a rectovaginal septum wall bulging without compromising the mucosa. The USG-3D resulted in a heterogeneous picture with regular borders, situated between the vagina and the

Fig. 1 – USG-A 3D showing a hypoechoic lesion in the upper region. (A) Axial cut; (B) diagonal cut.
anorectal junction (rectovaginal septum) measuring about 2.0 cm × 2.0 cm × 2.0 cm, without penetration of rectal muscle and with no lymph node evidence (Fig. 2B–D). The nodule was excised transvaginally, and the histopathology revealed mesenchymal neoplasm, with immunohistochemistry positive for CD117 (polyclonal) (Fig. 3).

**Discussion**

Histologically, GIST is characterized by the proliferation of spindle cells, forming small bundles arranged in multiple directions. The immunohistochemical study is characterized by CD34 marker positivity, ranging from 46% to 100% and with CD117 (c-kit) positivity in all cases.

It is difficult to predict the degree of malignancy for GIST. Hsu et al. identified histological features indicating malignancy, such as tumor size and mitotic activity. Tumors greater than 5.0 cm or with more than two mitoses by ten high-power fields (HPF) indicate increased risk of metastasis or recurrence.

The only effective treatment is by surgical resection of the tumor with tumor-free margins. The laparoscopic approach is a satisfactory option in the resection of small tumors; in several series of cases of gastric GIST resection, this approach proved to be safe and with low recurrence rates.

In patients with large distal rectal GIST, an abdominoperineal amputation will be needed.

Neoadjuvant or adjuvant radiotherapy and chemotherapy does not provide good results. However, imatinib mesylate (STI571 or Gleevec), which acts as a tyrosine kinase inhibitor, has been described with good results as monotherapy for the treatment of metastatic GIST. As a neoadjuvant agent, imatinib can produce tumor downsizing and allow for sphincter preservation.

To this end, a good diagnostic evaluation for choosing the appropriate surgical treatment should be obtained. USG-A has been widely applied to clarify the diagnosis of anorectal

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**Fig. 2** – (A) View of a septal bulging; (B) USG-3D, sagittal cut; (C) USG-3D, cross cut; (D) USG-3D axial cut.

**Fig. 3** – Surgical specimen. (A) Rectal nodule; (B) longitudinal section.
diseases, because this method evaluates the anatomical structures that form the anal canal, rectum and perianorectal tissues.21–39 Thus, USG-A was suggested as the best method to study the integrity of the layers of the intestinal wall and adjacent areas.40–41 USG-A also makes it possible to establish the distance from the endometrial focus to the sphincteric apparatus, suggesting which anastomosis will be the most appropriate in each case. Thus, we can plan the appropriate surgical approach for each case.42

In this sense, one can make use of several types of tools and probes.36,42–51 More recently, a probe was introduced with three-dimensional reconstruction capability of a transaxial parallel imaging sequence (0.25 mm), characterized by the formation of a cube.25,26,30,32,52–54

The mobilization mechanism may be manual or automatic, depending on the type of transducer used. The image formed into a cube can be recorded and widely deployed, enabling the operator to acquire all types of cuts, even in multidivision, which constitutes in simultaneous viewing four and six specialized images, besides the possibility of subsequent revision of these images as often as desired. This significantly improves the accuracy of the test, in addition to increasing the amount of information obtained.52

In cases of rectal GIST, the scarce literature suggests performing nuclear magnetic resonance54 so that one can attain accurate assessments. The ultrasound may be an alternative in this area, but the authors used a low-frequency, two-dimensional endoscopic probe,55 which results in a low-resolution image quality.

In a comparative study of resonance and two-dimensional anorectal ultrasonography in the evaluation of patients with endometriosis, these two techniques were equivalent in terms of image quality. But a study related to the evaluation of perianorectal layers and tissues with three-dimensional mode versus MRI has not yet been published.56

Despite the growing number of publications on GIST, there is little information on its rectal presentation and on the evaluation with USG-A. This technology has the ability to analyze the size of the tumor and its location, extent and depth, as well as its relationship to adjacent structures, besides analyzing lymph node involvement.

This study aims to take into account the importance of USG-3D in the evaluation of anorectal region and perianorectal tissues, in order to predict the best treatment to be chosen in the case of suspicion of an anorectal injury, and more specifically, a rectal GIST, because this method allows a good assessment of the affected rectal layers, besides diagnosing if there is an invasion of adjacent organs and blood/lymphatic vessels.

Conclusion

The use of anorectal ultrasonography in cases of rectal GIST has great value in identifying affected rectal layers, lesion size, and involvement of adjacent organs and of blood/lymphatic vessels, helping in the guidance for the best treatment to be proposed.

Conflicts of interest

The authors declare no conflicts of interest.

REFERENCES


