



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

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localizados no reto distal e avaliados com ultra-sonografia anorretal tridimensional, que se apresenta de grande valia na identificação do tamanho da lesão, acometimento de estruturas vizinhas e invasão linfonodal.

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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.⁸

Clinical manifestations of GIST are related to its location, which can be multiple, and its dimensions.⁹ 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 peri-anorectal 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 Gastroclínica 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

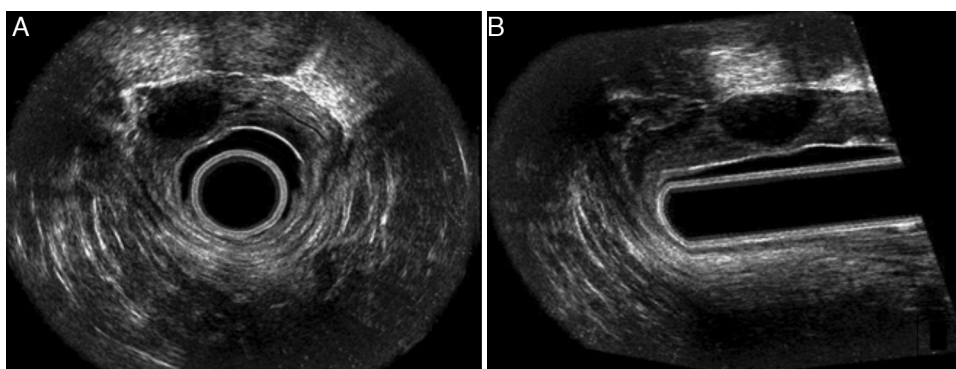


Fig. 1 – USG-A 3D showing a hypoechoic lesion in the upper region. (A) Axial cut; (B) diagonal cut.

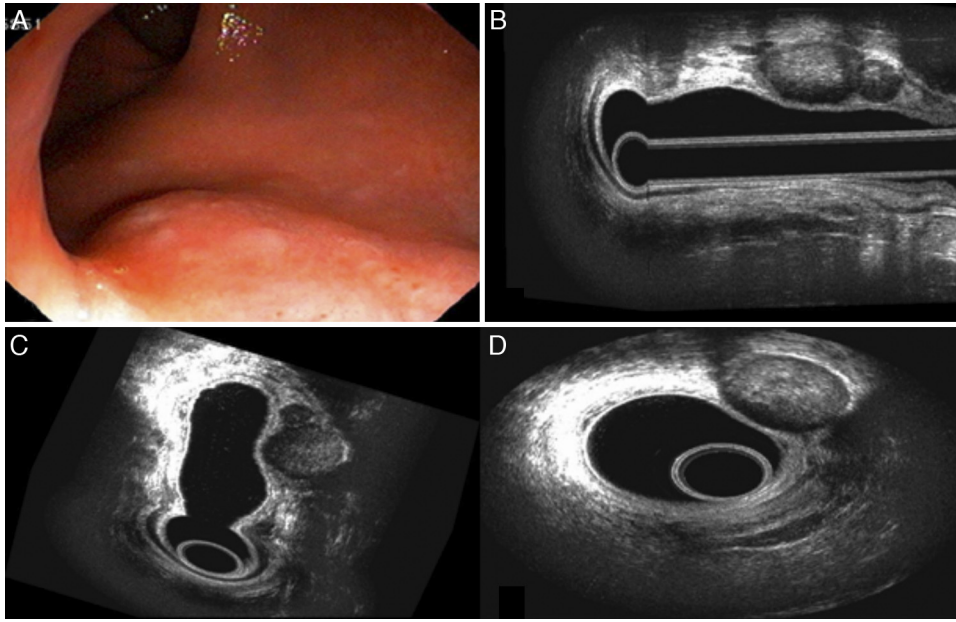


Fig. 2 – (A) View of a septal bulging; (B) USG-3D, sagittal cut; (C) USG-3D, cross cut; (D) USG-3D axial cut.

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 CD 117 (polyclonal) (Fig. 3).

Discussion

Histologically, GIST is characterized by the proliferation of spindle cells, forming small bundles arranged in multiple directions.¹⁵ The immunohistochemical study is characterized by CD34 marker positivity, ranging from 46% to 100% and with CD117 (c-kit) positivity in all cases.¹⁶

It is difficult to predict the degree of malignancy for GIST. Hsu et al.³ 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.¹⁷

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.¹⁸

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

Neoadjuvant or adjuvant radiotherapy and chemotherapy does not provide good results.¹⁹ 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.^{10,17,20} 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

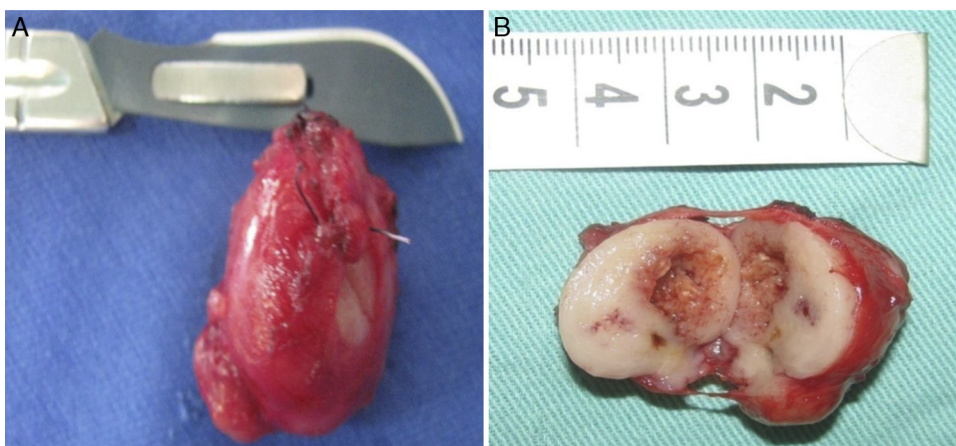


Fig. 3 – Surgical specimen. (A) Rectal nodule; (B) longitudinal section.

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 multivision, 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

- Efron DT, Lillemoe KD. Current management of gastrointestinal stromal tumors. *Adv Surg.* 2005;39:193-221.
- Gupta M, Sheppard BC, Corless CL, MacDonell KR, Blanke CD, Billingsley KG. Outcome following surgical therapy for gastrointestinal stromal tumor. *J Gastrointest Surg.* 2006;10:1099-105.
- Hsu K-H, Yang T-M, Shan Y-S, Lin P-W. Tumor size is a major determinant of recurrence in patients with resectable gastrointestinal stromal tumor. *Am J Surg.* 2007;194:148-52.
- Miettinen M, Lasota J. Gastrointestinal stromal tumors: pathology and prognosis at different sites. *Semin Diagn Pathol.* 2006;23:70-83.
- Tran T, Davila JA, El-Serag HB. The epidemiology of malignant gastrointestinal stromal tumors: an analysis of 1,458 cases from 1992 to 2000. *Am J Gastroenterol.* 2005;100:162-8.
- Shinomura Y, Kinoshita K, Tsutsui S, Hirota S. Pathophysiology, diagnosis, and treatment of gastrointestinal stromal tumors. *J Gastroenterol.* 2005;40:775-80.
- Miettinen M, Sobin LH, Lasota J. Gastrointestinal stromal tumors of the stomach: a clinicopathologic, immunohistochemical, and molecular genetic study of 1765 cases with long-term follow-up. *Am J Surg Pathol.* 2005;29:52-68.
- Sanchez VZ. GIST rectal. *Rev Esp Enferm Dig Madrid.* 2008;100.
- Dani R. *Gastroenterologia essencial.* 3 ed. Rio de Janeiro: Guanabara Koogan; 2006.
- Li A, Nordberg ML, Herrera GA. Gastrointestinal stromal tumor: current concepts and controversies. *Pathol Case Rev.* 2002;7:226-33.
- Huilgol R, Young CJ, Fracs S. The GIST: case reports of a gastrointestinal stromal tumour and a leiomyoma of the anorectum. *ANZ J Surg.* 2003;73:167-9.
- Dragested J, Gammelgaard J. Endoluminal ultrasonic scanning in the evaluation of rectal cancer: a preliminary report of 13 cases. *Gastrointest Radiol.* 1983;8:367-9.
- Hildrebrand U, Fiefel G. Preoperative staging of rectal cancer by intrarectal and management of squamous-cell carcinoma of the anal canal. *Dis Colon Rectum.* 2002;45:16-22.
- Tarantino D, Bernstein MA. Endoanal ultrasound in the staging ultrasound. *Dis Colon Rectum.* 1989;28:42-6.
- Linhares E, Valadão M. Atualização em GIST. *Rev Col Bras Cir Rio de Janeiro.* 2006;33:51-4.
- Greenson JK. Gastrointestinal stromal tumour and other mesenchymal lesions of the gut. *Mod Pathol.* 2003;16:366-75.
- Franquemont DW. Differentiation and risk assessment of gastrointestinal stromal tumours. *Am J Clin Pathol.* 1995;103:41-7.
- Nishimura J, Nakajima K, Omori T, Takahashi T, Nishitani A, Ito T, et al. Surgical strategy for gastric gastrointestinal stromal tumors: laparoscopic vs. open resection. *Surg Endosc.* 2007;21(6):875-8.
- Harrison ML, Goldstein D. Management of metastatic gastrointestinal stromal tumour in the Glivec era: a practical case-based approach. *Intern Med J.* 2006;36:367-77.
- Langer C, Gunawan B, Schüller P, Huber W, Füzesi L, Becker H. Prognostic factors influencing surgical management and outcome of gastrointestinal stromal tumour. *Br J Surg.* 2003;90:332-9.

21. Baker H, Schweiger W, Cerwenka H, Mischinger HJ. Use of anal endosonography in diagnosis of endometriosis of the external anal sphincter. *Dis Colon Rectum*. 1991;42:680-2.
22. Blatchford GJ. Anal ultrasonography in the evaluation of fecal incontinence. *Sem Colon Rectal Surg*. 1995;6:99-104.
23. Kleinübing H Jr, Jannini JF, Malafaia O, Brenner S, Pinho TM. Transperineal ultrasonography: new method to image the anorectal region. *Dis Colon Rectum*. 2000;43:1572-4.
24. Cheong DM, Noguerras JJ, Wexner SD, Jagelman DG. Anal endosonography for recurrent anal fistulas: image enhancement with hydrogen peroxide. *Dis Colon Rectum*. 1993;36:1158-60.
25. Christensen AF, Nielsen MB, Engelholm SA, Roed H, Svendsen LB, Christensen H. Three-dimensional anal endosonography may improve staging of anal cancer compared with two-dimensional endosonography. *Dis Colon Rectum*. 2004;47:341-5.
26. Gold DM, Bartram CI, Halligan S, Humphries KN, Kamm MA, Kmiot WA. Three-dimensional endoanal sonography in assessing anal canal injury. *Br J Surg*. 1999;86:365-70.
27. Goldman S, Norming U, Svensson C. Transanorectal ultrasonography in the staging of anal epidermoid carcinoma. *Int J Colorectal Dis*. 1991;6:152-7.
28. Herzog U, Boss M, Spichtin HP. Endoanal ultrasonography in the follow-up of anal carcinoma. *Surg Endosc*. 1994;8:1186-9.
29. Hildebrant U, Feifel G. Preoperative staging of rectal cancer by intrarectal ultrasound. *Dis Colon Rectum*. 1985;28:42-6.
30. Hunerbein M, Pegios W, Rau B, Vogl TJ, Felix R, Schlag PM. Prospective comparison of endorectal ultrasound, three-dimensional endorectal ultrasound and endorectal MRI in the preoperative evaluation of rectal tumors. Preliminary results. *Surg Endosc*. 2000;14:1005-8.
31. Katsura Y, Yamada K, Ishizawa T, Yoshinaka H, Shimazu H. Endorectal ultrasonography for the assessment of wall invasion and lymphnode metastasis in rectal cancer. *Dis Colon Rectum*. 1992;35:362-8.
32. Kim JC, Cho YK, Kim SY, Park SK, Lee MG. Comparative study of three-dimensional and conventional endorectal ultrasonography used in rectal cancer staging. *Surg Endosc*. 2002;16:1280-5.
33. Regadas SMM, Regadas FSP, Rodrigues LV, Nogueira MAA, Regadas RP. Ultrasono anorretal. Indicações e resultados preliminares. *Rev Bras Coloproctol*. 1999;19:5-10.
34. Sentovich SM, Blatchford GJ, Falk PM, Thorson AG, Christensen MA. Transrectal ultrasound of rectal tumors. *Am J Surg*. 1993;166:638-42.
35. Schröder J, Löhnert M, Doniec JM, Dohrmann P. Endoluminal ultrasound diagnosis and operative management of rectal endometriosis. *Dis Colon Rectum*. 1997;40:614-7.
36. Tarantino D, Bernstein MA. Endoanal ultrasound in the staging and management of squamous-cell carcinoma of the anal canal: potential implications of a new ultrasound staging system. *Dis Colon Rectum*. 2002;45:16-22.
37. Williams JG. Anal ultrasonography in the diagnosis and management of abscess and fistula disease of the anorectum. *Sem Colon Rectal Surg*. 1995;6:105-13.
38. Yang YK, Wexner SD, Noguerras J, Jagelman DG. The role of anal ultrasound in the assessment of benign anorectal disease. *Coloproctology*. 1993;5:260-4.
39. Rosch T, Lorenz R, Classen M. Ultrasonography in the evaluation of colon and rectal disease. *Gastrointest Endosc*. 1990;36:S33-9.
40. Tio TL, Weijers O, Hulsman F, Jonkers L, Collins E, Sie LH, et al. Endosonography of colorectal diseases. *Endoscopy*. 1992;24 Suppl. 1:309-14.
41. Schroder J, Lohnert M, Doniec JM, Dohrmann P. Endoluminal ultrasound diagnosis and operative management of rectal endometriosis. *Dis Colon Rectum*. 1997;40:614-7.
42. Tjandra JJ, Milson JW, Stolfi VW, Lavery I, Oakley J, Church J, et al. Endoluminal ultrasound defines anatomy of the anal canal and pelvic floor. *Dis Colon Rectum*. 1992;35:465-70.
43. Bahr A, Parades V, Gadonneix P, Etiennay I, Salet-Lize ED, Villet R, et al. Endorectal ultrasonography in predicting rectal wall infiltration in patients with deep pelvic endometriosis: a modern tool for an ancient disease. *Dis Colon Rectum*. 2006;49:869-75.
44. Kleinübing H Jr, Jannini JF, Malafaia O, Brenner S, Pinho M. Transperineal ultrasonography: new method to image the anorectal region. *Dis Colon Rectum*. 2000;43:1572-4.
45. Piloni V. Dynamic imaging of pelvic floor with transperineal sonography. *Tech Coloproctol*. 2001;5:103-5.
46. Beer-Gabel M, Teshler M, Barzilai N, Lurie Y, Malnick S, Bass D, et al. Dynamic transperineal ultrasound in diagnosis of pelvic floor disorders – a pilot study. *Dis Colon Rectum*. 2002;45:239-48.
47. Beer-Gabel M, Teshler M, Schechtman E, Zbar AP. Dynamic transperineal ultrasound vs. defecography in patients with evacuatory difficulty: a pilot study. *Int J Colorectal Dis*. 2004;19:60-7.
48. Adams DR, Blatchford GJ, Lin KM, Ternent CA, Thorson AG, Christensen MA. Use of preoperative ultrasound staging for treatment of rectal cancer. *Dis Colon Rectum*. 1999;42:159-66.
49. Karaus M, Neuhaus P, Wiedenmann B. Diagnosis of enteroceles by dynamic anorectal endosonography. *Dis Colon Rectum*. 2000;43:1683-8.
50. Gavioli M, Bagni A, Piccagli I, Fundaro S, Natalini G. Useful of endorectal ultrasound after preoperative radiotherapy in rectal cancer. *Dis Colon Rectum*. 2000;43:1075-83.
51. Buchanan GN, Bartram CI, Williams AB, Halligan S, Cohen CRG. Value of hydrogen peroxide enhancement of three-dimensional endoanal ultrasound in fistula-in-ano. *Dis Colon Rectum*. 2005;48:141-7.
52. Regadas SMM, Regadas FSP, Rodrigues LV, Silva FR, Lima DMR, Regadas-Filho FSP. Importância do Ultra-som Tridimensional na Avaliação Anorretal. *Arq Gastroenterol*. 2005;42:226-32.
53. Dam C, Lindebjerg J, Rafaelsen SR. MRI of rectal stromal tumour. *Ugeskr Laeger*. 2012;174:1826-7.
54. Murad-Regadas SM, Regadas FSP, Rodrigues LV, Escalante RD, Silva FRS, Lima DMR, et al. Ecodefecografia Tridimensional Dinâmica. Nova Técnica para Avaliação da Síndrome da Defecação Obstruída (SDO). *Rev bras Coloproct*. 2006;26:168-77.
55. Hussain T, Salamat A, Farooq MA, Hassan F, Hafeez M. Indications for endoscopic ultrasound and diagnosis on fine-needle aspiration and cytology. *Coll Physicians Surg Pak*. 2009;19:223-7.
56. Stewart LK, McGee J, Wilson SR. Transperineal and transvaginal sonography of perianal inflammatory disease. *AJR Am J Roentgenol*. 2001;177:627-32.