CASE REPORT

ANORECTAL LEIOMYOMAS: REPORT OF TWO CASES WITH DIFFERENT ANATOMICAL PATTERNS AND LITERATURE REVIEW

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Gastrointestinal mesenchymal tumors comprise a rare group of gastrointestinal tract wall tumors that have long been a source of confusion and controversy, especially in terms of pathological classification, preoperative diagnosis, management strategies, and prognosis. This report describes the clinical manifestations and management of 2 rectal leiomyomas and reviews the pertinent literature. Case 1: A 44-year-old woman was admitted reporting a nodule in the right para-anal region for the previous 2 years. At proctological examination, a 4-cm diameter fibrous mass situated in the para-anal region that produced an arch under the smooth muscle on the right rectal wall just above the anorectal ring was noted. Computed tomography and magnetic resonance imaging of the abdomen and pelvis showed the lesion and detected no other abnormalities. Surgical treatment consisted of wide local resection of the tumor through a para-anal incision, with no attempts to perform lymphadenectomy. Case 2: A 40-year-old male patient was admitted reporting constant anal pain for 4 months. He presented a 3-cm submucosal nodule at the anterior rectal wall just above the dentate line. After 2 inconclusive preoperative biopsies, transanal resection of the tumor was performed. Histological analysis of the specimen showed a benign leiomyoma. A review of the literature is presented, emphasizing some clinical and therapeutic aspects of this unusual rectal tumor.

KEY WORDS: **Gastrointestinal stromal tumors. Leiomyoma. Leiomyosarcoma. Rectum. Literature.**

Gastrointestinal mesenchymal tumors are nonepithelial lesions that exhibit an immature proliferation of epithelioid or spindle cells from the gastrointestinal tract muscle layer. Historically, these tumors have been called benign (leiomyoma) or malignant (leiomyosarcomas). More recently, pathologists have begun to shift from these terms to *gastrointestinal stromal tumors* (GIST).

Stromal tumors may occur in any muscle layer segment of the digestive tract, such as smooth muscle tissue (muscularis mucosa or muscularis propria), nervous tissue originating in the myenteric plexus, or mesenchymal primitive cells. Thus, GIST are histologically classified into 4 types: smooth muscle, neural, mixed, and undifferentiated. Additionally, the smooth muscle terminology may still be used when cellular differentiation is clearly evidenced.[5]

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Pathological and prognostic classifications separate leiomyomas, low-grade leiomyosarcomas, and high-grade leiomyosarcomas.[3] Histological guidelines for highly malignant tumors do exist, including size (greater than 5 cm), mitotic rate (greater than 10 per 10 high-powered fields), necrosis number, increased vascularity, and cellular atypia. Low-grade lesions may have only 1 mitosis per 10 high-powered fields and be smaller than 5 cm in size.[18,24]

Regarding their topographic distribution, GIST prevail in upper

gastrointestinal regions. The stomach and small bowel are more frequently affected by these tumors, and the rectum is estimated to account for 7% to 11% of all gastrointestinal smooth muscle tumors.[18,21] Kim et al.[8] reported 19 patients with GIST in Georgia (USA) as follows: 12 gastric, 2 duodenal, 3 jejunal, and 2 rectal. Among 24 surgically treated patients during the last 10 years in Milano (Italy), Chiara et al.[2] observed 6 gastric leiomyomas, 1 ileal leiomyoma, 4 gastric leiomyosarcomas, 1 esophageal leiomyosarcoma, 4 ileal leiomyosarcomas, 2 rectal leiomyosarcomas, and 6 gastric leiomyoblastomas. In Taiwan, Chou et al.[3] reported 80 gastrointestinal smooth muscle tumors that were surgically removed between 1986 and 1992 as follows: 1 esophageal, 32 gastric, 33 intestinal, 2 colonic, and 12 rectal.

The incidence of rectal GIST is very low. Many years ago, it was estimated that 1 leiomyoma may be found in every 2000 or more rectal tumors.[10] Ten years later, Zerilli et al.[25] estimated this incidence to be around 0.15 and 0.3% of colorectal malignant neoplasms. Leiomyomas located in the anal canal and sphincter are the rarest ones. In Brazil, papers focusing on smooth muscle tumors of the rectum have been rarely published.[9,16,18,20]

Leiomyomas have a highly variable clinical course, and the therapeutic strategy is still controversial.[8] Due to their submucosal origin, these tumors are often asymptomatic at initial stages. When present, symptoms are similar to those observed in common anorectal diseases, namely, local discomfort or pain (related or not to defecation), sensation of a foreign body, change in bowel habits, and rectal bleeding.[25]

Leiomyomas occur mainly in patients between 40 and 50 years. Clinical diagnosis depends on awareness of these lesions, digital rectal examination, proctoscopy, and tissue biopsy. A great majority of rectal smooth muscle and stromal tumors are GIST with variations ranging from minimal indolent tumors to overt sarcomas.[12] While differential histological diagnosis between benign and malignant forms is a dilemma, treatment should always be surgical.

Preoperative diagnosis is difficult to achieve because a biopsy is often valueless, since it does not involve the entire tumor mass.[4,14,19] When diagnosis provided by biopsy does not show the malignant nature of the lesion, its histological features can be assessed only after complete local excision.[17]

The present paper reports 2 cases of rectal leiomyoma, giving emphasis to their clinical manifestations, diagnosis, and management; a review of the pertinent literature is also presented.

CASE REPORTS

Case 1

A 44-year-old-woman was admitted reporting a slow-growing nodule at the right para-anal region for the previous 2 years. The tumor was painless, and the patient had no bleeding, change in bowel habits, or weight loss. She reported a family history of colon (aunt) and breast cancer (mother). General physical examination was unremarkable. Upon proctologic assessment, a delimited mobile fibrous mass situated at the right para-anal region was noted. Although this mass did not invade the rectum, it was palpable just contiguously to the right rectal wall; on digital examination, it was felt like a little bulge just above the anorectal ring.

Magnetic resonance images showed a slight parietal thickening of the right distal rectum and a 14.5 x 7.0 x 6.0 cm solid, well-defined mass in the ischioanal space extending up to the gluteus region. Other abdominal structures had no changes (Figure 1). Computed tomography (CT) findings were absolutely similar to those already described by magnetic resonance.

Under epidural anesthesia, surgery was performed in the lithotomy position. Considering the location of the mass, a shallow para-anal incision was made radially on the right side of the anal margin. Dissection of the subcutaneous fat provided tumor visualization, and progressive liberation of surrounding tissues was made in order to achieve the superior margins of the tumor close to the sciatic tuberosity. After resection, the wound was repaired by primary closure (Figure 2).

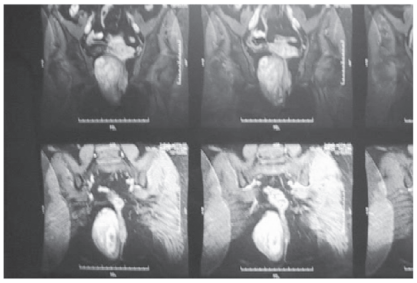

Figure 1 - Magnetic resonance showing a pararectal mass (case 1).

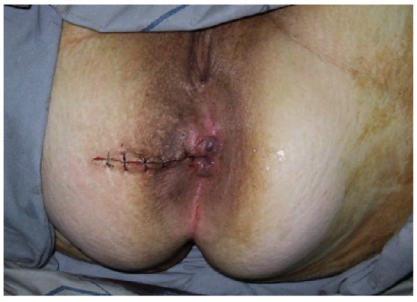

Figure 2 - Perineal wound closed after excision of the tumor (case 1).

The patient was released on the second postoperative day. Macroscopic and histological analysis revealed features of a benign leiomyoma (smooth muscle GIST) (Figure 3).

Case 2

The second patient was a 40-year-old man whose complaint was constant anal pain during the previous 4

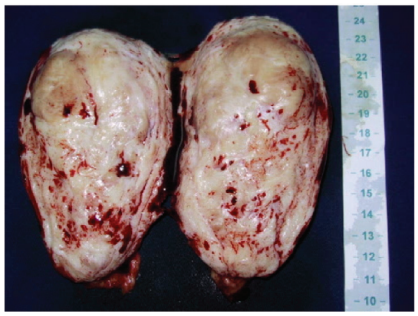

Figure 3 - Macroscopic vision of the opened mass (case 1).

months. At proctologic examination, a submucosal round nodule situated at the anterior rectal wall just above the dentate line was noted. The lesion had an approximately 3-cm diameter, and there was no mucosal ulceration. Two preoperative biopsies were inconclusive.

Tumor resection was performed under regional anesthesia with the patient positioned in the jack-knife position (Figure 4). A transanal excision was made with the aid of an anal device used for stapled hemorrhoidectomy. After a midline incision over the mucosa covering the tumor, 3 stitches were placed in the surrounding mucosa to facilitate the access to the tumor. The tumor was then carefully dissected and enucleated after liberation from the inner planes. The mucosa was then closed using absorbable and interrupted suture (Figure 5). Histological analysis of the specimen revealed a benign leiomyoma (smooth muscle GIST) (Figure 6).

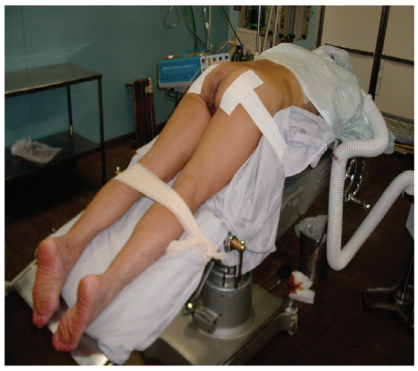

Figure 4 - Patient in Jack-knife position (case 2).

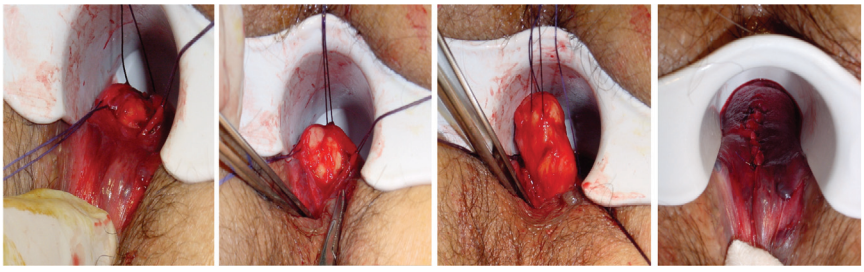

Figure 5 - Dissection, transanal excision of the tumor and mucosal closure (case 2).

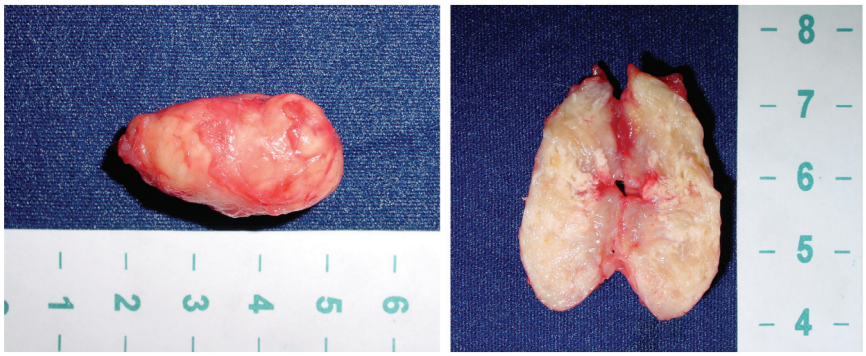

Figure 6 - Macroscopic aspects of the resected lesion (case 2).

DISCUSSION

Despite its low incidence and prevalence, rectal and anal leiomyomas have been discussed in case reports and review papers. These publications have usually dealt with the approaches of diagnostic tools and surgical management.

In a recent publication, Hatch and coworkers[5] reviewed all case reports about rectal and anal canal stromal tumors described in the world literature between 1881 and 1996. This review included 432 leiomyomas and 480 leiomyosarcomas. The review revealed that that leiomyomas predominately occur between 40 and 59 years of age. Our 2 patients were 40 and 44 years of age. In another review,[12] GIST were found to occur in adults with a median age of 60 years (range, 17-90 years) with a significant male predominance (71%).

Leiomyomas often remain asymptomatic until they have reached a fairly large size. The most common symptoms are bleeding, palpable mass, and rectal pain.[5] Patients usually presented clinical complaints for the previous 12 months, and those with leiomyomas tended to tolerate symptoms longer before attaining medical intervention.[4] Regarding the cases reported here, the woman presented no local pain or bleeding. Despite the long duration of symptoms, she only reported a slow-growing mass. This clinical picture of an asymptomatic mass has also been commonly observed in other reports.[10,19] Occasionally, patients will report bleeding (if the overlying mucosa ulcerates), constipation, pain, or a sense of fullness. Almost always, the chain of events leading to diagnosis starts when the tumor is discovered incidentally by digital examination or as a submucosal mass at rectoscopy.[10,25] Our male patient reported rectal pain and a sense of fullness.

Tumors may vary from small asymptomatic intramural nodules to large masses that bulge into pelvis, causing pain, rectal bleeding, or obstruction. Colonic and rectal leiomyomas often present as intraluminal

polypoid masses.[12] In a collective review of smooth muscle tumors of the rectum and anal canal, Hatch et al.[5] reported that intraluminal growth of both leiomyomas and leiomyosarcomas was more frequently seen than extraluminal or intramural patterns, and tumors were more likely to be found in the rectum than in the anus.

The 2 cases presented here had tumors in a close proximity to the rectum, and while the tumor in the female patient (case 1) presented as an extramural lesion, the man (case 2) had a lesion that could be characterized as an intramural tumor.

The majority of smooth muscle tumors appear as submucosal nodules, although a few of them have been described as polypoid. Ulceration of the overlying mucosa may occur in both leiomyomas and leiomyosarcomas. Stromal tumor dissemination occurs primarily by direct extension to adjacent organs. Hematogenous metastasis can reach the liver, lung, bones, and brain.[12] Although involvement of lymph nodes rarely occurs, it is associated with poor survival rates.[1]

Therefore, imaging techniques are useful for preoperative staging, since they can describe the relationships with the sphincters and urogenital tract, and they can detect metastatic spread to regional lymph nodes.[25] Complementary investigation, such as with CT, endorectal ultrasonography, and magnetic resonance imaging scan, strongly corroborates the diagnosis. Endorectal ultrasound can help to define the extent of disease and may be a useful adjunct in deciding about the appropriate surgical procedure.[6] During the treatment of our female patient, information obtained from physical assessment, CT scan, and magnetic resonance were sufficient and ruled out the need of endorectal ultrasonography.

Furthermore, radiological evaluation was very useful in defining operative strategy. In the first patient, the para-anal location of the tumor, as suggested by physical examination and confirmed by the magnetic resonance, allowed us to excise the tumor through a radial incision starting at the right anal margin. Thus, the tumor mass was easily found, dissected, and excised. During the treatment of the second patient, rectal assessment clearly showed a small submucosal and mobile tumor. These anatomical features suggested that surgery could be safely accomplished through a transanal approach.

The lack of reliable criteria for malignancy is the main problem the surgeon faces when selecting the operative procedure. Although most of the 150 leiomyomas of the rectum reported since 1872 were not larger than 5 cm, Le Borgne et al.[11] described 3 rectal leiomyomas measuring more than 5 cm. Hatch et al.[5] found that rectal leiomyosarcomas tended to be larger than leiomyomas, as was also the case for these neoplasms in other gastrointestinal locations. Tumors with an original size larger than 5 cm are those that have shown the highest tendency to recur, mostly as sarcomas. Therefore, recurrent lesions should be treated radically from the beginning.[10] Additionally, smooth-muscle rectal tumors should be considered more dangerous than those in other locations in the gastrointestinal tract, since half are malignant and only one-fifth of patients who have sarcomas survive 5 years.

When evaluating the clinical symptoms of a patient, one must have in mind that bleeding, constipation, and weight loss are associated with a higher risk of malignancy.[5]

Preoperative diagnosis can be difficult, and the final diagnosis is often made at the time of surgical treatment, such as with the second patient reported here. Preoperative histological diagnosis is adequate in only 29% of cases.[2] Microscopic diagnosis and differentiation of malignant from benign features require a pathologist with special interest and expertise with these lesions. The ultimate proof of malignancy is therefore determined by recurrence of the tumor or metastasis. Since they grow within the intestinal wall, symptoms are usually few or late, leading to delay in diagnosis.[17]

Leiomyomas are relatively insensitive to adjuvant therapy. Therefore, their treatment is primarily surgical and should guarantee complete clearance of the tumor.[1]

The choice of surgical approach for a rectal lesion depends mainly on clinical and histopathological findings. Small and benign-appearing lesions for which histology has excluded malignancy should be treated by local excision with adequate margins, followed by periodic surveillance. With complete resection of the tumor, the clinical course is favorable, with very few local recurrences. Local excision of low rectal lesions may be accomplished by a conventional transanal excision, while upper tumors may be excised using either transanal endoscopic microsurgery or a posterior approach.[15,25]

Since our 2 patients had tumors with no gross or histological features of malignancy, their management through a local excision was considered adequate, and the patients were assigned to a follow-up program.

True rectal leiomyosarcomas are rare and account for less than 0.1% of all malignant tumors of the rectum. One estimate is that less than 300 cases have been reported so far, and anal lesions are even rarer.[23] They tend to occur between 50 to 69 years of age, and approximately 20% of rectal leiomyosarcomas reported from 1881 to 1996 had metastasized at diagnosis.

Several therapeutic modalities may be involved in the treatment of leiomyosarcomas of the rectum, including radical resection, local excision, and

even nonoperative therapy.[7, 13] Chemotherapy and radiotherapy are generally not effective.[24] Although there is no clear evidence that adjuvant therapy influences overall survival, further trials are needed to establish its exact role, since good results have been reported in selected patients, with disease-free interval prolongation.[17, 23]

Radical surgery is indicated for local recurrence of benign tumors, and treatment of larger (greater than 5 cm) and malignant rectal myomas should be very aggressive from the onset (low anterior resection or abdominoperineal resection), although there is a questionable rate of survival improvement.[11,14] According to Chou et al.,[3] leiomyosarcomas usually measure more than 10 cm, and the significant factors affecting survival rates at univariate analysis are maleness, size greater than 5 cm, inadequate resection, and advanced-stage and high-grade disease.

Although the most common therapy for large and low differentiated leiomyosarcomas of the rectum is radical resection through abdominoperineal resection or low anterior resection, the need for radical resection of this extent has been questioned with regard to treatment of high-graded tumors smaller than 2.5 cm. Some authors recommend a wide local excision; however, in this case a high local-recurrence rate has to be expected. This is the reason for a more radical treatment of all leiomyosarcomas of the rectum.

Vorobyov et al.[22] reported their experience with the treatment of 36 rectal leiomyomas from 1972 to 1990 in Moscow. There were 13 male (36 %) and 23 female (64 %) patients, and median age was 52.1 years. Electroexcision of tumors measuring less than 1 cm was performed endoscopically in 12 patients. Leiomyomas measuring 2.5 to 5 cm were removed through the transanal approach in 10 patients. Six patients underwent excision of the tumor through the pararectal approach, whereas leiomyomas located in the rectovaginal wall were removed through the vagina in 1 patient. Abdominoperineal extirpation and abdominoanal resection of the rectum was performed in 7 patients with tumors measuring 8 to 20 cm. Recurrences were noted in 9 patients after transanal, pararectal, or transvaginal excision of leiomyomas. In 7 of them, malignant transformation of the tumor occurred at terms ranging from 9 months to 9.5 years.

The local recurrence rate for resectable leiomyosarcomas was more than 80%, exceeding the propensity of leiomyosarcomas in other areas of the gastrointestinal tract to recur.[5] In the study performed by Miettinen et al.,[12] 70% of patients with tumors > 5 cm with more than 5 mitosis/50 high power fields (HPF) (n = 31) died of disease, whereas only 1 tumor <2 cm with < 5 mitosis/50 HPF (n = 21) recurred, and none caused death. Long latency was common between the primary operation and recurrences and metastases; either one occurring in 60 of 111 patients with follow-up (54%).

Therefore, extended follow-up is required, because long-term recurrences seem to be also possible in cases involving low-grade lesions.[2] Biologic behavior also varies with location; colonic tumors are generally less aggressive when compared to rectal ones. Rectal tumors manifest greater recurrence and dissemination rates, even after wide resections with curative purposes.[5]

According to Witzigmann et al.,[24] the prognosis for rectal leiomyosarcomas is generally poor. Survival rates vary from 20% to 25% in 5 years.[1] Although size, histological grade, and local staging play an important role, complete resection is considered the most significant favorable prognostic factor. Regarding anal leiomyosarcomas, the evaluation of prognosis is hampered because its rarity.

RESUMO

Campos FG e col. Leiomiomas anorretais: descrição de dois casos com características anatômicas diferentes e revisão da literatura. **Rev. Hosp. Clín. Fac. Med. S. Paulo 59**(5):296-301, 2004.

Os tumores mesenquimais gastrointestinais constituem um grupo raro de neoplasias que têm sido fonte de confusão e controvérsia, especialmente quanto à classificação patológica, diagnóstico pré-operatório, manuseio e prognóstico. O presente artigo descreve as manifestações clínicas e o tratamento de dois pacientes com leiomioma retal e revê a literatura pertinente. Caso 1: Uma mulher de 44 anos foi admitida referindo um nódulo na região paranal direita nos últimos 2 anos. Ao exame físico notou-se uma massa fibrosa de 4 centímetros de diâmetro situada na região paranal que produzia um discreto abaulamento na musculatura lisa da parede retal, logo acima do anel ano-retal. As imagens de tomografia computadorizada e ressonância magnética do abdômen e pelve confirmaram a lesão e não detectaram outras anormalidades. O tratamento cirúrgico consistiu de ressecção alargada do tumor através de uma incisão paranal, sem se realizar linfadenectomia. Caso 2: Outro paciente com 40 anos foi admitido com história de dor anal constante há 4 meses. Este homem

apresentava nódulo submucoso de 3 cm na parede retal anterior, logo acima da linha pectínea. Após duas biópsias inconclusivas, realizou-se a ressecção transanal do tumor. A análise histológica do espécime demonstrou tratar-se de um leiomioma benigno. Uma breve revisão da literatura é apresentada, enfatizando alguns aspectos clínicos e terapêuticos deste tumor retal pouco comum.

UNITERMOS: **Tumores estromais gastrointestinais. Leiomioma. Leiomiossarcoma. Reto. Literatura.**

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