Solitary schwannoma of the rectum mimicking rectal cancer.
Report of a case and review of the literature

Michele Tedeschi, Filippa Cuccia, Emanuele Angarano, Domenico Piscitelli*, Gianluigi Gigante, Donato F. Altomare

University “Aldo Moro” of Bari, Bari, Italy
Department of Emergency and Organ Transplantation
*Department of Pathology

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A case of a submucosal tumor bulging into the rectum in an old female patient is reported. It proved to be a very rare case of rectal schwannoma, whose differential diagnosis is difficult to obtain preoperatively and optimal treatment and prognosis still uncertain. The mass was removed by means of an ultrasound device after an open anterior rectotomy. The operation was uneventful and the patient is disease free 18 months after.

KEY WORDS: Schwannoma recti, Surgery, Prognosis, GIST.

Introduction

Gastrointestinal nerve tumours are uncommon stromal tumours (accounting for only 0.1% of benign tumours) of the gastrointestinal tract were firstly described and defined by Herrera in 1989. Schwannomas are benign tumours belonging to this group, and may develop in any anatomical region. Schwannomas have been reported to account for only 3% of all gastrointestinal mesenchymal tumours and particularly rare in the rectum. They have been reported to occur in patients with a median age of 50 to 60 years with a prevalence in females with a size ranging from 0.5 to 11 cm. There is evidence that benign schwannomas arise from neuron-supporting cells in the myoenteric plexus as site-specific modified Schwann cells, or at any rate that the cells involved in the development of these tumours are schwannian in origin. Macroscopically schwannomas present as solid, round, capsuled tumours with hard consistency and greyish colour. In rare cases they can undergo cystic degeneration, necrosis, and calcifications, but these features are more frequent in schwannomas of the soft tissues, such as those originating in the retroperitoneum.

According to the histological features of the tumor cells, 3 different variants of colorectal schwannomas have been identified, namely spindle cells, epithelioid and plexiform cells. Using a different classification, it is possible to describe two histological types of schwannomas: Antoni type A with densely packed spindle cells (Verocay bodies) and Antoni type B with loosely organized spindle cells (absence Verocay bodies) in myxoid stroma. Preoperative imaging tests including abdominal x-rays, transanal 3D US, MRI and CT scans can help the identification and definition of these tumors but the true diagnosis is almost always histological. Differential diagnosis needs to be made between Schwannomas, GIST and desmoids, that are the most frequent mesenchymal tumours involving the gastrointestinal tract.
Herein we present a case of an 80-year-old woman who underwent surgical removal of a rectal mass proved to be a Schwannoma of the rectum.

**Case history**

An 80-years-old woman underwent colonoscopy with the supposed diagnosis of rectal cancer because of recurrent rectal bleeding, constipation with incomplete defecation and tenesmus. At endoscopy, a rounded polyloid lesion covered by normotrophic mucosa, broad-based plant which seemed to originate from the rectal submucosa was identified at 13 cm from the anal verge. A subsequent abdominal CT scan confirmed the presence of a expansive process mimicking a solid polyloid submucosal mass of about 4 cm in diameter which largely restricted the rectal lumen. No secondary lesions were identified. Physical examination including anal digital exploration was negative. Also blood tests were normal, including tumor markers (TPA: 28.1, CA 125: 18.08, Ca 19.9: 3.35, CEA: 2,25).

This frail old woman had a BMI of 19 and was classified as ASA III for the anaesthesiologic risk because of hypertension, chronic heart insufficiency and depressive disorder.

After laparotomy the rectum was fully mobilized from the sacrum along the holy plane using harmonic scalpel device without vascular disruption. The anterior wall of the rectum was opened and a 4 cm polypoid neoplasm covered by normal mucosa of the posterior wall of middle rectum was identified (Fig. 1).

The tumor was removed by bipolar scissors and the posterior rectal wall reconstructed by interrupted vycril 3/0 stitches. The anterior rectal wall was then closed by transverse single layer vicryl suture.

The postoperative course was uneventful and the patient was discharged 10 days after surgery. At two year follow up the patient was asymptomatic.

**Macroscopic findings and histology**

Show (Fig. 2) the schwannoma as a polypoid mass of 4 cm with smooth surface focally ulcerated. The cut surface looked solid, smooth, tan-yellow coloured. No cystic change, hemorrhage and calcification were present. This benign encapsulated tumor was composed of cells with the histological and immunophenotype features of Schwann cells. The neoplasia arose in the submucosa of rectum, focally ulcerated the mucosa and was provided by thick fibrous capsule with aggregates of small lymphocytes along the perimeter. It consisted of elongate spindle-shaped nuclei and abundant pink cytoplasm, aligned and compactly disposed in broad bundles (like Antoni type A), no mitosis or cellular atypia were evident. The cells were uniformly and heavy immunopositive for S-100 protein (Fig. 3).
Discussion

The incidence of submucosal schwannomas has been reported to range between 2% to 6% of all submucosal tumours of the intestine. Among the case reports of benign schwannomas in the digestive tract published in the literature, the esophagus and the stomach are the most frequent sites involved, followed by the small intestine, the appendix and Meckel's diverticulum. Schwannoma of the colon is rare and only few reports of this tumour localized in the rectum were found in a literature search. For the retrievable literature a table showing patients features, tumor size, and disease interval free time is reported (Table I). Other intra-abdominal sites, such as the great omentum, lesser sac, liver and the biliary tree are even rare. The correct diagnosis of schwannoma of the rectum is difficult to get at an early stage since it generally grows very slowly and asymptomatic, but sometimes cause rectal bleeding, anemia, bowel occlusion, difficult defecation, mild fever, asthenia and pain. Furthermore preoperative diagnosis is always difficult because any imaging test is not typical and even biopsy specimens taken at endoscopy are meaningless because of the submucosal growth of the tumor.

At histology schwannoma cells show a strong, diffuse expression of vimentin and protein S-100. Rarely schwannomas can express CD-34 but never CD-117, SMA or desmin, and this is important to distinguish schwannomas from other mesenchymal tumours such as GIST, that express CD-117 as well as CD-34, and tumours originating from the smooth muscle cells, that express SMA and desmin. According to Arai et al., a positive immunoreactivity to S-100 protein and Leu 7 antigen indicates a schwannian nature of the tumor. Although schwannomas are considered to be benign tumours, the risk of recurrence cannot be overlooked. In a study conducted by Lauwers, 30% of patients developed a local recurrence and in the large group of 303 patients reported by Gupta et al., 2% of them developed distant metastases.

An aggressive behaviour is generally correlated with the size of the tumor and the extent of mitotic activity, in fact masses larger than 10 cm. and/or with a mitotic count of more than 5-10/HPF are generally correlated with a high risk of local recurrence and distant metastases, and have poor prognosis. To better standardize the terminology and avoid confusion the WHO coined for these tumors the term MPNST (malignant peripheral nerve sheath tumors).

The optimal treatment for schwannomas is complete surgical excision ensuring negative margins, since it is usually impossible to distinguish these tumours from other mesenchymal tumours of the gastrointestinal tract like GISTs but a close follow-up based on magnetic resonance is however recommended. Minimally invasive modalities are justified by the benign nature of the tumor and can be performed safely by transanal endoscopic microsurgery local excision when the tumor does not infiltrate the mesorectum (like in our case), by intersphincteric resection in case of very large tumors.

Table 1 - Review of the case reports of rectal schwannomas in the literature

<table>
<thead>
<tr>
<th>Authors/Year</th>
<th>Sex/Age</th>
<th>Symptoms</th>
<th>Size (cm)</th>
<th>Operation</th>
<th>Disease-free time</th>
</tr>
</thead>
<tbody>
<tr>
<td>Qasi /1996 (1)</td>
<td>F/29</td>
<td>Dyspareunia; constipation</td>
<td>5</td>
<td>NK</td>
<td>24 months</td>
</tr>
<tr>
<td>Kakizoe/1998 (2)</td>
<td>M/67</td>
<td>No</td>
<td>1,5</td>
<td>RE-TEM</td>
<td>9 months</td>
</tr>
<tr>
<td>Maciejewski/2000 (3)</td>
<td>F/67</td>
<td>Abdominal pain, flatulence</td>
<td>10</td>
<td>ATE</td>
<td>144 months</td>
</tr>
<tr>
<td>Miettinen/2001 (4)</td>
<td>M/82</td>
<td>NK</td>
<td>0,5</td>
<td>Endoscopic polypectomy</td>
<td>211 months</td>
</tr>
<tr>
<td>Bhardway/2002 (5)</td>
<td>M/35</td>
<td>Difficulty in defecation</td>
<td>NK</td>
<td>NK</td>
<td>24 months</td>
</tr>
<tr>
<td>Mulchandani/2005 (6)</td>
<td>M/70</td>
<td>Difficulty in defecation</td>
<td>12</td>
<td>Proctolectomy</td>
<td>6 months</td>
</tr>
<tr>
<td>Pescatori/2005 (7)</td>
<td>M/28</td>
<td>NK</td>
<td>NK</td>
<td>PIE</td>
<td>8 months</td>
</tr>
<tr>
<td>Lec/2006 (8)</td>
<td>F/61</td>
<td>Narrow calipered stool</td>
<td>4</td>
<td>Transanal excision</td>
<td>NK</td>
</tr>
<tr>
<td>Hsu/2010 (9)</td>
<td>F/88</td>
<td>Difficulty in defecation, Abdominal distention</td>
<td>4,3</td>
<td>PIE</td>
<td>9 months</td>
</tr>
<tr>
<td>Our patient/2010</td>
<td>F/80</td>
<td>Constipation, tenesmus, Recurrent rectal bleeding</td>
<td>4</td>
<td>transrectal excision</td>
<td>18 months</td>
</tr>
</tbody>
</table>

NK= not know; RE-TEM= rectal expander-assisted transanal endoscopic microsurgery; ATE= abdomino-transsacral exploration; PIE= perianal intersphincteric excision.
low anal location, or by laparoscopic approach in cases of retrorectal Schwannomas. In our case, due to the position in the posterior rectal wall at 13 cm from the anal verge, the uncertainty of the nature of the tumor and the poor clinical condition of the patient, an abdominal approach with an anterior rectotomy was preferred to remove the tumor instead of a rectal resection, with a short operating time, low risk of post-operative complications, and preserving the possibility of a second look in case of malignant nature of the neoplasm. The long term prognosis of this tumor has never been well documented but recurrences are uncommon and surgery is usually the curative treatment.

Conclusions

Schwannomas of the rectum are extremely rare but nevertheless this diagnosis needs to be considered in the differential diagnosis of rectal tumors in the presence of submucosal growing tumors. The treatment of choice is complete surgical excision ensuring negative margins, but wide resection is not necessary. A correct preoperative diagnosis is difficult to obtain, and is generally achieved by histologic and immunohistochemical examination of the surgical specimen.

Riassunto

Si riporta un caso di un tumore sottomucoso del retto riscontrato in una anziana paziente di sesso femminile. È stato provato che si trattava di un raro caso di Schwannoma del retto la cui diagnosi differenziale è difficile da ottenere in fase pre-operatoria e necessita sempre della conferma istologica e immunoistochemica. La massa è stata rimossa con l’utilizzo di un bisturi ad ultrasuoni dopo aver eseguito una rettotomia aperta anteriore e al follow-up a 18 mesi la paziente è libera da malattia. Tuttandosi di una patologia a comportamento biologico solitamente benigno, una chirurgia demolitiva non appare giustificata

References