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A Rare Sigmoid-Colon Schwannoma in a Premenopausal Woman: A Case Report

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Abstract. ***Background.*** Schwannomas of the gastrointestinal tract are a rare type of spindle cell tumor of peripheral nerve. Commonly, schwannomas are discovered incidentally, as they are usually asymptomatic.

Case. 46-year-old female patient, suffering from secondary amenorrhea and nonspecific intermittent pelvic pain associated with constipation. During gynecological visit an ultrasonographic systematic transvaginal examination was performed. At the sigmoid-rectal level an intraluminal solid mass was described and an urgent colonoscopy was prescribed. Endoscopic submucosal dissection was performed with en-bloc resection. On immunohistochemical analysis, S100 was strongly positive in tumor cells. Finally, a benign schwannoma of the sigmoid colon was diagnosed.

Conclusion. Our case highlights the importance of an adequate transvaginal pelvic examination with the evaluation of all pelvic organs. It could be challenging to make diagnosis in an early stage on asymptomatic patients.

Keywords: transvaginal ultrasound, rectal mass, sigmoid mass, premenopause, surgical procedures.

Reta sigmoidinės gaubtinės žarnos švanoma moteriai prieš menopauzę: atvejo aprašymas

Santrauka. ***Kontekstas.*** Virškinamojo trakto švanoma yra retas periferinio nervo verpstės ląstelių navikas. Dažniausiai švanomos aptinkamos atsitiktinai, nes jos dažniausiai būna besimptomės.

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Atvejis. 46 metų pacientė kenčia nuo antrinės amenorėjos ir nespecifinio protarpinio dubens skausmo, susijusio su vidurių užkietėjimu. Ginekologo vizito metu jai buvo atliktas ultragarsinis sisteminis transvaginalinis tyrimas. Sigmoidiniu tiesiosios žarnos lygiu buvo aptiktas intraluminalinis kietas darinys ir paskirta skubi kolonoskopija. Atlikta endoskopinė submukozinė disekcija su en-bloc naviko rezekcija. Imunohistocheminės analizės metu S-100 buvo stipriai teigiamas naviko ląstelėse. Galiausiai diagnozuota gerybinė sigmoidinės gaubtinės žarnos švanoma.

Išvada. Remiantis šiuo atveju norima pabrėžti tinkamo transvaginalinio dubens tyrimo svarbą, vertinant visus dubens organus. Ankstyvojoje ligos stadijoje nustatyti diagnozę pacientams, nesant aiškių ligos simptomų, gali būti sudėtinga.

Raktiniai žodžiai: transvaginalinis ultragarsas; tiesiosios žarnos masė; sigmoidinė masė; premenopauzė; chirurginės procedūros.

Introduction

Schwannomas are an uncommon type of spindle cell tumor of peripheral nerve in adults, which originate from schwann cells.

Gastrointestinal tract schwannomas occur in the stomach (83%), small bowel (12%) and colon-rectum (5%) [1]. Gastrointestinal schwannomas occur at similar rates in men and women with a mean age of 60–65 years [3]. Commonly, schwannomas are discovered incidentally on screening endoscopy or during abdominal imaging and the diagnosis is made on definitive pathologic examination of operative specimen [4]. On immunohistology, they stain positive for S100.

The differential diagnosis of this entity includes all mesenchymal or neuro-ectodermal neoplasms, in decreasing frequency, gastrointestinal stromal tumors (GISTs), leiomyomas, leiomyosarcomas, neurofibromas, ganglioneuromas, paragangliomas, lipomas, granular cell tumors, and glomus tumors [3].

Rectal schwannoma may rarely cause symptoms such as obstruction, bleeding, and tenesmus, however usually it is asymptomatic. Although, schwannomas are benign tumors, the possibility of malignant degeneration does exist and is directly related to the tumor dimensions. Radical surgical treatment is the gold standard in all cases, since the results of both chemotherapy and radiotherapy remain uncertain [7]. We report a case of sigmoid colon schwannoma incidentally observed at transvaginal ultrasonography and discuss the clinical features of the disease.

A systematic review of the literature was performed to explore the symptoms, the treatment and the prognosis of rectal swhannomas. Furthermore, we highlight the utility of pelvic ultrasound in the early diagnosis of this disease.

Case report

A 46-year-old caucasian, female patient, BMI 26.5 kg/cm^2, was referred to our Ultrasonographic Gynecological Center on April 2022, suffering from secondary amenorrhea from September 2021 and nonspecific intermittent pelvic pain associated with constipation. An early menopause was excluded after consulting the following hormonal exams: follicle-stimulating hormone (FSH) 5.84 UI/L; luteinizing hormone (LH) 2.5 UI/L; prolactine (PRL) 17.2 ng/mL, progesteron 0.15 ng/dl, 17-b-estradiol 65 ng/l; thyroid-stimulating hormone (TSH) 1.25 μU/ml. Her clinical history included menarche at age of 16-year-old, regular menstruation, one cesarean section and an appendicectomy. Last gynecological visit with Pap smear test was performed in September 2021. No comorbidities were reported.

At our center, an ultrasonographic systematic transvaginal examination was performed. Firstly, the anterior compartment was evaluated with a normal bladder and ureters. Regarding the evaluation of the uterus an endometrial homogeneous hyperechoic echostructure thickening of 15 mm

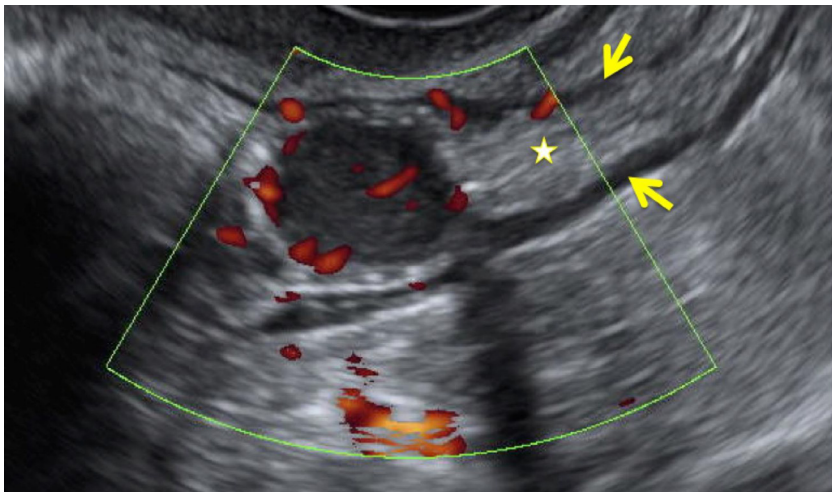

Fig. 1. Ultrasonographic transvaginal imagine: intraluminal solid mass, with increased peripheral and central vascularity of 12x11 mm.

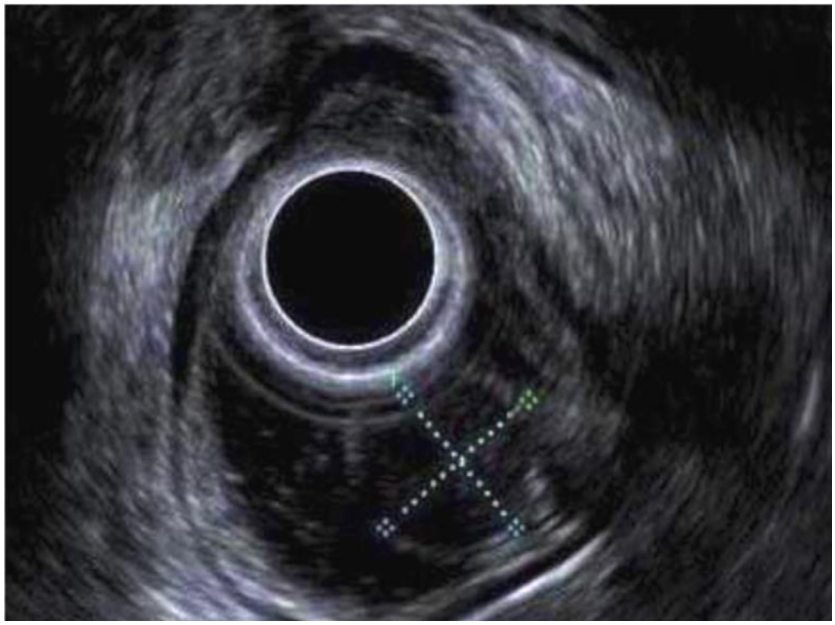

Fig. 2. Endoscopic ultrasonography findings of rectal neuroendocrine. Presence of a 12x12mm ovalar hypoechoic lesion, originating from the submucosa, in contact with the muscularis propria.

was observed, incompatible with her secondary amenorrhea. A present posterior sliding sign, and absent anterior one was observed, probably due to the cesarean section. Bilaterally, ovaries resulted mobile with poor follicular activity, compatible with the age of the patient. On the left ovary, a follicular unilocular thin-walled cyst with anechoic contents of 30 mm was observed. Finally, during the evaluation of the posterior compartment, at the sigmoid-rectal level we observed an intraluminal solid mass, with increased peripheral and central vascularity of 12x11 mm (Figure 1). Our first hypothesis was a rectal polyp or a neuroendocrine tumor (NET) and an urgent colonoscopy was prescribed.

At colonoscopy, performed on May, a submucosal mass covered by normal mucosa sized about 1 cm in the sigmoid colon was confirmed. Transanal endosonography was performed, confirming the presence of a 12x12mm ovalar hypoechoic lesion, originating from the submucosa, in contact with the muscularis propria (Figure 2). Therefore, endoscopic submucosal dissection was performed with en-bloc resection. The specimen was pinned on corkboard and fixed in 10% formalin. At macroscopic examination, a well-demarcated lesion without encapsulation was observed (Figure 3). At histology, hematoxylin and eosin stain showed that the tumor was composed of spindle cells with

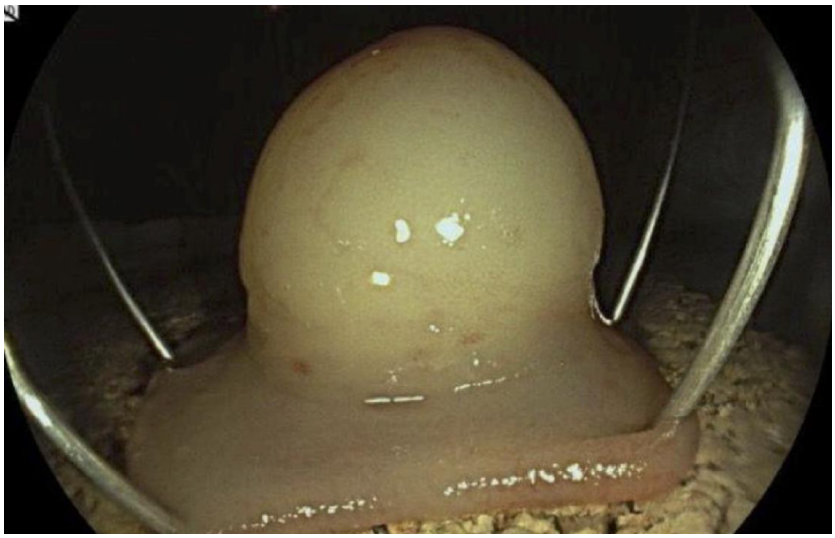

Fig. 3. Macroscopic specimen: submucosal mass covered by normal mucosa sized about 1 cm in the sigmoid colon.

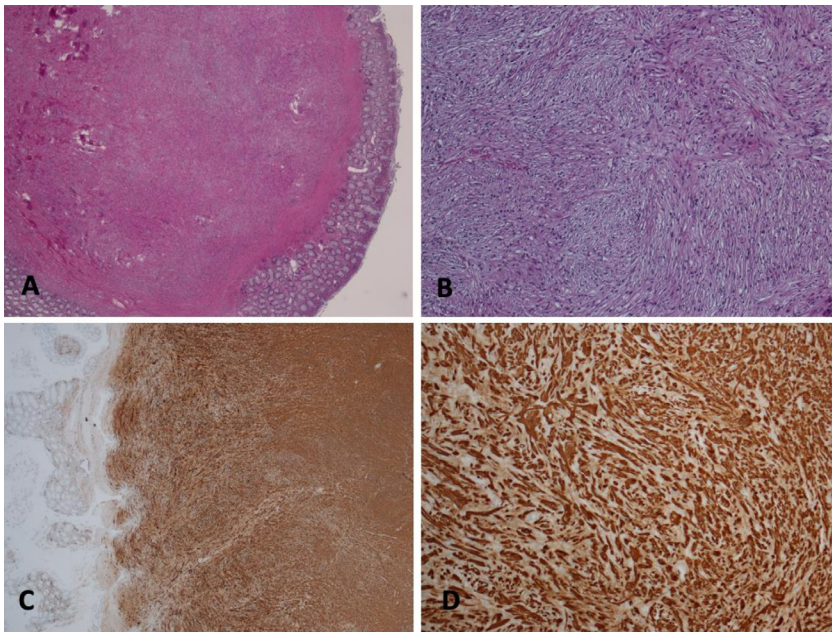

Fig. 4. Microscopic section:
A Colonic epithelioid schwannoma is composed of uniform polygonal tumor cells forming a trabecular pattern.
B The tumor cells illustrate strong immunoreactivity to S100 protein.
C The tumor was composed of multiple discontiguous nodules of S100 positive protein.
D Low-power appearance of the distinctive growth pattern of the tumor following haematoxylin and eosin staining.

low nuclear atypia and low proliferation index Ki 67 1%. The resection margins were free from tumor. On immunohistochemical analysis, the tumor cells illustrate strong immunoreactivity to S100 protein; otherwise, CD 117 (C-KIT), CD34, and Smooth Muscle Actin (SMA) were negative (Figure 4). Finally, the diagnosis was a benign schwannoma of the sigmoid colon. The patient was discharged after 3 days without postoperative complications. After oncological consultation, no postoperative adjuvant therapy was reputed to be needed.

The patient has signed a consent form.

Discussion

Schwannomas of the gastrointestinal tract are rare, particularly in the colon and rectum [8]. Colonic schwannomas are distributed equally among males and females and generally are diagnosed after the sixth decade of the life [8]. The tumors usually manifest themselves as intraluminal polyps or masses and can present with gastrointestinal bleeding from ulceration, colonic obstruction, or abdominal pain

[8]. Due to the rarity of symptomatic tumors, the majority of schwannomas are incidentally diagnosed after the operation. In particular, in a review of 41 cases of schwannomas of the large intestine in Japan only 10% of cases were identified pre-operatively [9]. Despite all efforts done preoperatively, the differential diagnosis of these tumors from other malignant stromal tumors is mostly possible after histological examination of the resected specimen. Computerized scans, barium enemas, and magnetic resonance imaging may reveal these tumors as encapsulated masses arising from the colon mucosa, but there is still a lack of set criteria to differentiate benign from malignant stromal tumors. If available, endosonographic features of irregular extraluminal margins, cystic spaces, and lymph node with malignant pattern help differentiate benign stromal cell tumors from the malignant tumors [10]. Schwannoma cells are immune reactive to S100 protein, vimentin and negative to Smooth Muscle Actin (SMA) and CD117 (KIT). Smooth muscle tumors have desmin and SMA reactivity. Gastrointestinal Stromal Tumor (GIST) with KIT mutation/expression show immunopositivity to CD117 and CD34 [6]. Histologically these tumor types also differ from GISTs because prominent lymphoid cuff and diffuse lymphoid infiltration, impression of cellular heterogeneity, focal nuclear atypia, and microtrabecular architectural pattern are only present in shwannomas [6]. These tumors are generally considered benign and radical surgical removal with free margins remains the first line of the treatment. A differential diagnosis with leiomyomas, whose histologic and immunohistochemical features are those of well-differentiated smooth muscle cells, is also needed (6). Metastatic melanoma is another S100 protein positive tumor that can go in differential diagnosis with shwannomas. In case of S100 protein positive malignant tumor with spindle or epithelioid features, the possibility of malignant melanoma should always be primarily considered, and other more specific markers should be examined, such as HMB45, tyrosinase, and microphthalmia transcription factor. Indeed, these markers, quite specific for melanoma, are not present in any of the colonic schwannomas [6]. Differently, intestinal polyps usually are not considered in the differential diagnosis of shwannomas, as the endoscopic appearance is quite typical, as these arise from dysplasia of epithelial cells.

In our case report a 46 year-old women was diagnosed with a colonic lesion by a transvaginal ultrasound examinations, which reported an intraluminal recto-sigmoid lesion treated with endoscopic submucosal dissection and finally characterized as a shwannoma. In our review, including only women in fertile age, only 3 cases were treated endoscopically. In literature, most cases undergo aggressive surgical intervention because of lack of preoperative diagnosis.

The main limitation of our case report refers to the limited possibility of generalizing the validity of the study. Furthermore, as a case report, it focuses on rare or unusual case, thus their use and interpretation must be carefully considered, within a much broader context.

The challenge for the future should be to perform a preoperative noninvasive diagnosis therefore allowing more conservative treatments such as endoscopic resections. Our case highlights the importance of an adequate transvaginal pelvic examination and the evaluation of all pelvic organs from the anterior to the posterior compartment. Nowadays, transvaginal ultrasonographic examination should have as target the evaluation of all pelvic organs and not just the uterus and the ovaries.

Conclusion

Our case illustrates an example of a transvaginal ultrasonographic early stage diagnosis, in a premenopausal women, of a rare rectal lesion, schwannoma, treated through endoscopic resection; otherwise an extensive surgery would have been requested. Nowadays, transvaginal ultrasonographic examination should have as target the evaluation of all pelvic organs and not just the uterus and the ovaries. Thus, a preoperative noninvasive and early stage diagnosis could allow a conservative treatment such as in our case, an endoscopic resection.

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