Ileal gastrointestinal stromal tumors mimicking gynecological masses: a single institutional experience

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Introduction

Gastrointestinal stromal tumor (GIST) is the most common mesenchimal neoplasm of the gastrointestinal tract. The small intestine is the second predominant site of origin of GIST (1). This neoplasia has a wide spectrum of clinical presentations and rarely can be diagnosed as an asymptomatic pelvic mass accidentally detected by transvaginal ultrasound and thus wrongly diagnosed as a gynaecologic tumor in female patients. To date, in literature 35 cases of ileal GIST mimicking gynecologic masses have been described (2-10). **The aim of this study is to describe the clinical and trans-vaginal sonographic appearance of an ileal GIST mimicking a gynecologic tumors** in five patients referred to our Institution, in order to better recognize this rare clinical presentation.

Patients and methods

We retrospectively reviewed 64 female patients with ileal GIST, collected since 2001 to 2017 in our GIST database. Among those 64 female patients with ileal GIST, we selected 5 patients presented with a pelvic mass and referred to a gynecological consultation at first. **Characteristics of patients are listed in Table 1.**

Results

Clinically, one patient was asymptomatic and the mass was occasionally found with a routine transvaginal ultrasound, two presented with abdominal pain, one with melena and one with a left inguinal swelling. In all cases CA 125 was negative or mildly increased (< 170 U/ml). **At trans-vaginal ultrasound exam all the lesions were predominant solid, dishomogeneous and hypoechoic (Figure 1A and 1B).** The mean diameter was 68 mm. In four cases the lesions were described as "lobulated masses". Cystic areas due to necrosis were described in one patient. In all cases **no acoustic shadows** were observed. In three cases the mobility towards other pelvic structures was reduced and in four cases **the masses were localized in the right pelvis mimicking right ovarian lesions**. All cases showed a **high vascularization at Power Doppler (Fig 2A and 2 B)**. A laparotomic ileal resection was performed in all cases. Hysterectomy and bilateral salpingo-oophorectomy were performed in 2 cases and left salpingo-oophorectomy in one. The histologic exam revealed an ileal GIST with a mitotic index ranging from <5/50 HPF to 19/50 HPF (median: 5/50 HPF) with bilateral ovarian metastases in one patient with an advanced disease (11).

Conclusion

In our experience the clinical presentation of ileal GIST was heterogeneous and nonspecific and does not permit a differential diagnosis between GIST and ovarian tumor. GIST shows transvaginal sonographic peculiar features. The mass appears solid, irregular, hypo-echoic with small anechoic parts, without acoustic shadows, highly vascularized and frequently dislocated in the right pelvis. Normal or mild serum levels of CA 125 (< 170 U/ml) and absence of free fluid in the pouch of Douglas can be considered ancillary features for suspicion of GIST. Lastly a gynecologic evaluation should be proposed in patients with a history of GIST because of the possibility of metastases to the genital tract (11).

References

1.Biasco G, Velo D, Angriman I, Astorino M, Baldan A, Baseggio M, et al. Gastrointestinal stromal tumors: report of an audit and review of the literature. Eur J Cancer Prev. 2009 Apr;18(2):106-16.
2.Dayan E, Zhu H, Copperman AB, Lewis S, Kamath A. Gastrointestinal stromal tumor presenting as a right adnexal mass with histopathologic correlation. Clin Imaging 2017;44:97-100
3.Ijeri SK, Rathod PS, Kundargi R, Pallavi VR, Shobha K, Shankaranand, Vijay CR, Uma Devi K, Bafna UD. Gastrointestinal Stromal Tumor Mimicking as Ovarian Tumor in Gynaecologic Oncology. Indian J Surg Oncol 2016;7:56-61.
4.Karaca N, Akpak YK, Tatar Z, Batmaz G, Erken A. Gastrointestinal Stromal Tumor: May Mimic Adnexal Mass. Glob J Health Sci 2015;8:20-6
5.Muñoz M, Ramirez PT, Echeverri C, Alvarez LG, Palomino MA, Pareja LR. Gastrointestinal stromal tumors as an incidental finding in patients with a presumptive diagnosis of ovarian cancer. J Gynecol Oncol 2012;23:48-52.
6.Teoh WC, Teo SY, Ong CL. Gastrointestinal stromal tumors presenting as gynecological masses: usefulness of multidetector computed tomography. Ultrasound Obstet Gynecol. 2011;37:107-9.
7.Angioli R, Battista C, Muzii L, Terracina GM, Cafà EV, Sereni MI, Montera R, Plotti F, Rabitti C, Panici PB. A gastrointestinal stromal tumor presenting as a pelvic mass: A case report. Oncol Rep 2009;21:899-902
8.Pinto V, Ingravallo G, Cicinelli E, Pintucci A, Sambati GS, Marinaccio M, D'Addario V. Gastrointestinal stromal tumors mimicking gynecological masses on ultrasound: a report of two cases. Ultrasound Obstet Gynecol 2007;30:359-61.
9.Morimura Y, Yamashita N, Koyama N, Ohzeki T, Takayama T, Fujimori K, Sato A. Gastrointestinal stromal tumor mimicking gynecological disease. Fukushima J Med Sci 2006;52:21-8.
10.Zigheboim I, Henao G, Kunda A, Gutierrez C, Edwards C. Gastrointestinal stromal tumor presenting as a pelvic mass. Gynecol Oncol 2003;91:630-5
11.De Leo A., Nannini M., Dondi G., Santini D., Urbini M., Gruppioni E., De Iaco P., Perrone A.M., Pantaleo M.A. Unusual bilateral ovarian metastases from ileal gastrointestinal stromal tumor (GIST): a case report. BMC Cancer. 2018 Mar 16;18(1):301

Fig.1 A: Trans vaginal ultrasound image of the right pelvic mass (Case 3)

Fig. 1 B: Study of the vascularization of the pelvic mass using transvaginal Doppler ultrasonography (Case 3)

Fig.2 A: Trans vaginal ultrasound image of the right pelvic mass (Case 4).

Fig. 2 B: Study of the vascularization of the pelvic mass using transvaginal Doppler ultrasonography (Case 4)

Table 1: Patients' characteristics

Patient	Age	Clinical presentation	Surgical Management	Disease status	Size (cm)	Mitotic index	Molecular analysis	Follow-up status
Case 1	31	Melena, anemia	Ileal resection	Localized	7,5	5/50 HPF	exon 11 KIT [c.1657_1668del (p.Y553_Q556)]	Alive without disease (OS: 49 months)
Case 2	77	Left inguinal swelling	Ileal resection hysterectomy bilateral salpingo oophorectomy asportation of peritoneal lumps	Advanced	17	9/50HPF	exon 9 KIT (c.1509_1510insGCCTAT)	Dead
Case 3	53	Abdominal pain	Ileal resection	Localized	8	5/50HPF	KIT/PDGFR WT	Alive without disease (OS: 16 months)
Case 4	76	Abdominal pain	Ileal resection, left salpingo-oophorectomy omentectomy	Localized	6,5	<5/50HPF	exon 11 KIT [p. V559D (c.1676T>A)]	Alive without disease (OS: 12 months)
Case 5	50	None	Ileal resection hysterectomy bilateral salingo oophorectomy douglas pouch and pelvic peritonectomy	Advanced	7	19/50HPF	exon 11 KIT [p.M552_W557 del (c-1653_1670del)]	Alive without disease (OS: 7 months)