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ABOUT COVER

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AIMS AND SCOPE

The primary aim of World Journal of Clinical Cases (WJCC, World J Clin Cases) is to provide scholars and readers from various fields of clinical medicine with a platform to publish high-quality clinical research articles and communicate their research findings online.

WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

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CASE REPORT

Extragastrointestinal stromal tumors with diffuse membranous distribution with bleeding: A case report

Jian-Duo Xu, Zheng Wang, Qian Zhou, Ning Meng, Shu-Mei Zhang, Nan Liu

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Abstract

BACKGROUND

Extragastrointestinal stromal tumors (EGIST) and gastrointestinal stromal tumors are of similar pathological type and form. Here we report a rare case of EGIST diffusely distributed in membranous tissue in abdominal cavity, the feature of which included diffuse tumors at membranous tissue in entire abdominal cavity and spontaneous bleeding of the tumors.

CASE SUMMARY

The patient was a 71-year man and hospitalized due to continuous pain at lower abdomen for more than 10 days. Upon physical examination, the patient had flat and tough abdomen with mild pressing pain at lower abdomen, no obvious abdominal mass was touchable, and shifting dullness was positive. Positron emission tomography-computed tomography (CT) showed that in his peritoneal cavity, there were multiple nodules of various sizes, seroperitoneum, multiple enlarged lymph nodes in abdominal/pelvic cavity and right external ilium as well as pulmonary nodules. Plain CT scanning at epigastrium/hypogastrium/pelvic cavity + enhanced three-dimensional reconstruction revealed multiple soft tissue nodules in abdominal/pelvic cavity, peritoneum and right groin. Tumor marker of carbohydrate antigen 125 was 808 U/mL, diffuse tuberous tumor was seen in abdominal/pelvic cavity during operation with hematocelia, and postoperative pathological examination confirmed EGIST. Imatinib was administered with better therapeutic effect.

CONCLUSION

Gene testing showed breast cancer susceptibility gene 1 interacting protein C-terminal



helicase 1 and KIT genovariation, and the patient was treated with imatinib follow-up visit found that his clinical symptoms disappeared and the tumor load alleviated obviously via imageological examination.

Key Words: Diffuse tumor in abdominal cavity; Extragastrointestinal stromal tumors; Gastrointestinal stromal tumors; Malignant extragastrointestinal stromal tumors; Diffusely membranous metastasis

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Core Tip: Extragastrointestinal stromal tumors (EGIST) are less common in comparison with gastrointestinal stromal tumors. EGIST tumors in the membranous tissue are rare and tumors independently developing in membrane tissues such as greater omentum, mesentery or peritoneum are occasionally reported. Diffuse membranous tumor in entire abdominal cavity is extremely rare worldwide. We report a case of EGIST with the tumors like cobblestone being diffusely distributed in entire abdominal cavity accompanied by spontaneous bleeding. The tumors may not be fully resected by surgery, hence we made biopsy, pathological examination and gene detection to determine the therapy. Treatment with imatinib achieved better outcome.

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INTRODUCTION

Extragastrointestinal stromal tumors (EGIST) are less common compared with gastrointestinal stromal tumors (GIST). The tumor developing in membranous tissue is rare and independently developing in membrane tissues such as greater omentum, mesentery or peritoneum has been occasionally reported. Diffuse membranous tumor in entire abdominal cavity is extremely rare worldwide. We here report a case of EGIST with the tumors like cobblestone being diffusely distributed in entire abdominal cavity accompanied by spontaneous bleeding. The tumors may not be fully resected by surgery, hence we made biopsy, pathological examination and gene detection to determine the treatment strategy. The patient was treated with imatinib, and achieved better outcome.

CASE PRESENTATION

Chief complaints

The patient was a 71-year man, with hypertensive disease for 6 months, the highest pressure being 200/110 mmHg, and he has been taking felodipine orally to control blood pressure.

History of present illness

The patient had continuous mild pain at lower abdomen about 10 days prior to hospitalization, with regular bowel movement, and loss of appetite and weight for 5 kg.

History of past illness

He had a history of smoking for 50 years, 60 cigarettes/day, without drinking or exposing to radioactive substance.

Personal and family history

His family history was unremarkable.

Physical examination

Upon physical examination, the patient had normal abdominal appearance, tough abdomen with mild pressing pain at lower abdomen, no rebound tenderness or muscular tension, no obvious abdominal mass was touchable, shifting dullness was positive, with normal bowel sound.

Laboratory examinations

Tumor marker of carbohydrate antigen 125 was 808 U/mL. Gene detection showed breast cancer susceptibility gene 1 interacting protein C-terminal helicase 1 (BRIP1) and KIT genovariation, and the patient was thus sensitive to imatinib. Immunohistochemical analysis resulted in the following results: Calretinin (-), D2-40 (-), CK5/6 (-), WT-1 (+), CD117 (+), Dog-1 (+), SHA (diffused weakly +), Desmin (-), S-100 (-), SOX-10 (-), P16 (-), CD34 (-), and Ki-67 (30% approximately).



Imaging examinations

Plain CT scanning at epigastrium/hypogastrium/pelvic cavity + enhanced three-dimensional reconstruction revealed multiple soft tissue nodules in abdominal/pelvic cavity, peritoneum and right groin (Figure 1). Positron emission tomography-computed tomography showed thickened peritoneum with abnormal hypermetabolism, multiple mass or nodular soft tissue density images in abdominal/pelvic cavity and peritoneum (Figure 2). On enhanced scanning, the lesion was enhanced at a mild to moderate level; multiple hypermetabolic lymph nodes in abdominal/pelvic cavity and right external ilium; and seroperitoneum were observed.



Figure 1 Abdominal computed tomography scanning. It shows that the tumor is of heavy load and diffused in the abdominal cavity.



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Figure 2 Positron emission tomography-computed tomography. It shows that the peritoneum is thickened with abnormal hypermetabolism, multiple mass or nodular soft tissue density images in abdominal/pelvic cavity and peritoneum; *via* enhancement scanning, the lesion is enhanced in mild to moderate level; multiple hypermetabolic lymph nodes in abdominal/pelvic cavity and right external ilium; and seroperitoneum.

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FINAL DIAGNOSIS

Based on pathological examination, a diagnosis of EGIST was considered (Figure 3).



Figure 3 Pathological examination. Based on pathological examination, extragastrointestinal stromal tumors of high risk was considered.

TREATMENT

Based on gene detection results, the patient was treated with imatinib.

OUTCOME AND FOLLOW-UP

Follow-up visit found that his clinical symptoms disappeared and the tumor load alleviated obviously *via* imageological examination (Figure 4).



Figure 4 Abdominal computed tomography after treatment. It shows the tumor load is alleviated obviously with good therapeutic effect.

DISCUSSION

EGIST is rarer in comparison with GIST. GIST is diagnosed due to bleeding and obstruction while EGIST usually presents pain and palpable lump. In this case, the patient was hospitalized due to abdominal pain; upon physical examination, the entire abdomen was tough without solitary lump, but accompanied with spontaneous bleeding in abdominal cavity (Figure 5). The patient had symptoms of both GIST and EGIST. EGIST is commonly seen at vulva[1,2], vagina[3,4], retroperitoneum[5], ovary, posterior gastric, testis[6], greater omentum[7,8], pancreas[9,10], prostate[11-13], bladder[14], mesentery[15,16], liver[17], rectum[18,19], rectosacral space[20], esophagus[21], etc. But this case is rare where diffuse membranous tissue tumors existed in abdominal cavity. In addition, EGIST may have spontaneous rupture[22], just as this case, as shown in Figure 6 for tumor rupture and bleeding. Some EGIST cases are characterized by strong invasion and have unfavorable prognosis[23]. Uzunoglu and Tosun[24] have reported 135 EGIST cases between 2007 and 2020 and showed that elderly or female patients had worse prognosis. In our case, the patient was an old man with strong tumor

invasiveness; however, conservative medicating treatment showed a better treatment effect.



Figure 5 Preoperative abdominal computed tomography. Surgical exploration shows that the masses in abdominal cavity had spontaneous necrosis and bleeding.



Figure 6 Tumor rupture and bleeding. During operation, diffuse nodules like cobblestone in abdominal cavity with hematocelia were seen. The nodules were in various sizes, approximately 0.5-1.5 cm, diffusely distributed in membrane tissues such as peritoneum, greater omentum and mesentery. The first picture on the left showed purple black nodules bleeding and bright red blood not coagulated.

Compared with GIST, EGIST has negative c-KIT mutation and positive PDGFRA in histopathological examination and immunohistochemical analysis. Whatever the tumor location is, DOG1 remains a preferred biomarker and CN34 and CD117 are deemed as auxiliary examination[25]. The gene detection for this case showed BRIP1 and KIT genovariation, BRIP1 (point mutation, exon11; nucleotide variation c.1567A > G; amino acid variation p.T523A; abundance 34.45%), KIT (insertion/deletion variation, exon11; nucleotide variation c.1672_1677del; amino acid variation p.K558_V559del; abundance 34.94%). The detection showed that the patient was sensitive to imatinib. After treatment with imatinib, his clinical symptoms disappeared gradually and the abdominal CT reexamination showed that the imageological indexes had obviously improved. This case is different from previous ones in clinical symptoms, imageological manifestations and gene detection results, which have indicated that EGIST has various manifestations, thus comprehensive diagnostics should be performed.

CONCLUSION

This case of EGIST is rarely seen in clinical practice. The patient was a 71-year man with continuous hypogastralgia. During laparoscopic surgery, several botryoid nodules of various sizes were found in the abdominal cavity. Surgical



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treatment was impossible. The patient underwent tumor biopsy, and histopathological examination, which showed positive CD117, and gene detection showed BRIP1 and KIT genovariation. After treatment with imatinib, his clinical symptoms disappeared and the abdominal CT reexamination showed that the imageological indexes had obviously improved. In this case, we made an overall evaluation on tumor characteristics in the aspects of imageological examination, gene detection, immunohistochemical analysis and pathological feature, and clinical manifestation difference, which verifies that imatinib treatment for EGIST with diffuse membranous distribution has better therapeutic effect than surgical treatment.

FOOTNOTES

Author contributions: Xu JD designed the research; Wang Z performed the research; Meng N, Zhou Q, Zhang SM, and Liu N analyzed the data and wrote the manuscript; all authors have read and approved the final manuscript.

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