Case Report

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Clear Cell Sarcoma-Like Tumor of the Gastrointestinal Tract with Peritoneal Metastasis in a Young Adult: A Case Report with Literature Review

젊은 성인에서 복막 전이를 동반한 위장관의 투명 세포 육종 유사 종양: 증례 보고 및 문헌 고찰

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Clear cell sarcoma-like tumor of the gastrointestinal tract (CCSLTGT) is a rare malignant mesenchymal tumor of the gastrointestinal (GI) tract with a high probability of local recurrence and distant metastasis in young adults. To the best of our knowledge, only seven case reports have described the imaging findings of a CCSLTGT originating from in the small intestine in English literatures so far. Therefore, we report the imaging findings of a CCSLTGT that occurred in the small intestine of a 22-year-old female and summarize the imaging findings of the previous reports.

Index terms Clear Cell Sarcoma; Small Intestine; Neoplasms; Computed Tomography, X-Ray; Differential Diagnosis

INTRODUCTION

Clear cell sarcoma-like tumor of the gastrointestinal tract (CCSLTGT), also referred to as malignant gastrointestinal neuroectodermal tumor is a rare malignant mesenchymal tumor of the gastrointestinal (GI) tract, which arises from primitive neural crest cells that are related to the autonomous nervous system in the bowel wall (1). Zambrano et al. (2) first described an osteoclast-rich tumor of the GI tract that resembled clear cell sarcoma (CCS) on pathological examination. However, due to the lack of melanocytic markers in CCS, Stock-

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man et al. (3) designated this as a CCS-like tumor. To the best of our knowledge, there have been only seven case reports including imaging findings of small-bowel CCSLTGT. Herein we describe a case of CCSLTGT with peritoneal metastasis in a young female.

CASE REPORT

A 22-year-old female presented with epigastric pain, nausea, and weight loss of 15 kg over 6 months. The patient had no medical or surgical history and exhibited no remarkable findings on physical examination. The initial laboratory exam showed mild hyponatremia, and the complete blood cell count was normal.

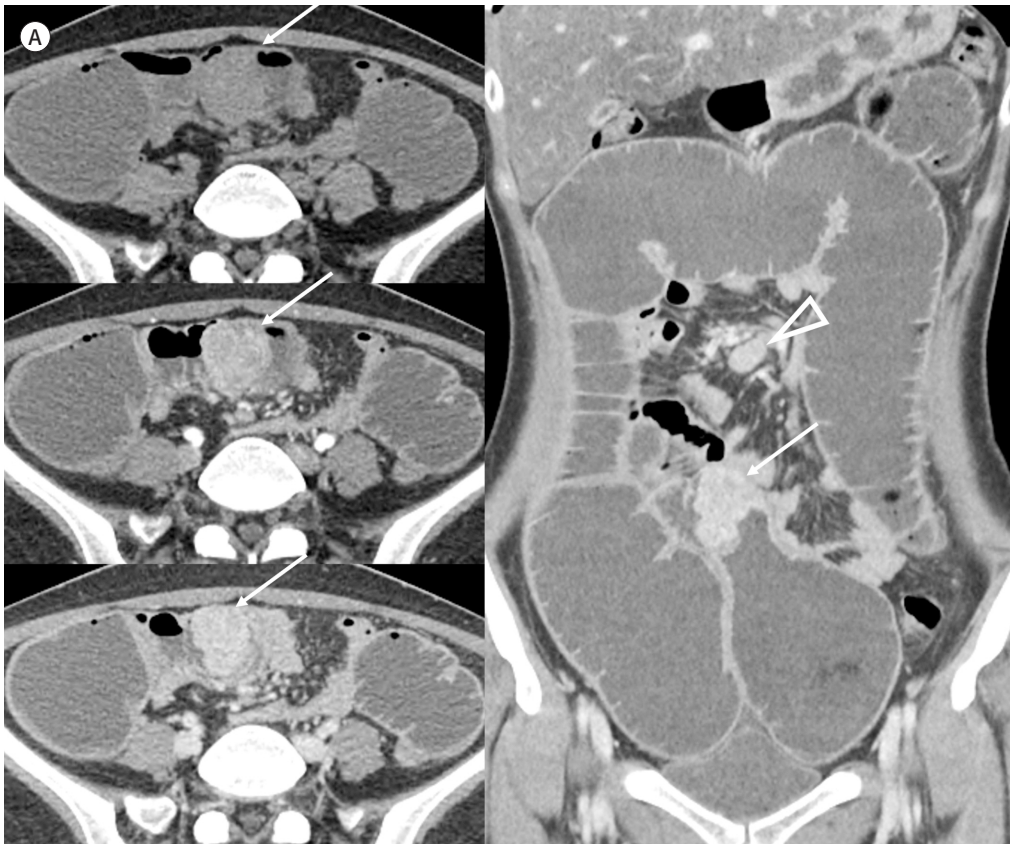
CT revealed a lobulated contour mass lesion sized approximately 3.3 cm × 2.7 cm × 3.6 cm in the mid-abdomen with proximal bowel loop distension. The mass was protruding into the intraluminal area of the small bowel loop, but also showed an exophytic appearance outside of the bowel lumen with a lobulated outer margin. On the pre-contrast image, the mass showed similar attenuation to the adjacent muscles (approximately 45 Hounsfield units [HU]) without any calcification or hemorrhage. A contrast-enhanced scan in the arterial phase showed heterogeneous strong enhancement of approximately 110 HU, and in the delayed phase, the mass showed progressive enhancement of approximately 125 HU (Fig. 1A). There was no internal necrotic or cystic change in the mass. No desmoplastic reaction was observed. Multiple enlarged mesenteric lymph nodes were observed adjacent to the tumor with maximum long diameter 2.5 cm, suggesting metastatic lymphadenopathy (Fig. 1A). Although the tumor was protruding into the intraluminal area and showed an extraluminal exophytic appearance, the epicenter of the tumor was in the intraluminal area of the small intestine. Hence, tumors of small bowel origin such as neuroendocrine tumor (NET), gastrointestinal stromal tumor (GIST), adenocarcinoma, and lymphoma were considered as differential diagnoses, rather than tumors of mesenteric origin.

The patient underwent resection of the small-bowel mass and lymph node dissection. In the operative field, the mass was observed in the jejunum 1 m below the Treitz ligament and was approximately 5 cm × 5 cm in size. Gross examination showed a mass constricting the small intestine, and tumor cells formed a mass under the mucosal layer extending from the serosa to the submucosa. The tumor was strongly positive for the S100 protein (Fig. 1B) in the absence of melanocytic markers (HMB45 and Melan-A), and fluorescence in situ hybridization showed the EWSR1 breakpoint region on chromosome 22q12. These pathological findings were consistent with CCSLTGT, and seven out of 43 lymph nodes were pathologically proven to have metastasized. Therefore, the final pathological stage was pT4N2Mx.

A fluorine-18 fluorodeoxyglucose (F-18 FDG) PET/CT was performed postoperatively to evaluate distant metastases and it showed a hypermetabolic soft tissue lesion in the left paracolic gutter, suggesting peritoneal metastasis (Fig. 1C). Otherwise, no other distant metastases were observed. Preoperative CT revealed a nodular enhancing lesion sized approximately 1.1 cm in the left paracolic gutter, which corresponded to a hypermetabolic lesion on the F-18 FDG PET/CT scan.

The patient did not undergo chemotherapy, and a CT scan obtained 1 month after surgery showed a slight increase in the size of the peritoneal metastasis in the left paracolic gutter and

Fig. 1. Clear cell sarcoma-like tumor of the gastrointestinal tract in a 22-year-old female presenting with epigastric pain and weight loss.
A. The non-enhanced axial CT image (left upper) shows a lobulated contour soft tissue mass (arrows) without hemorrhage or calcification. Heterogeneous strong enhancement (110 HU) in the arterial phase (left middle) and progressive enhancement (125 HU) in the delayed phase (left lower) are observed. On the coronal CT image (right), the tumor is mainly located in the intraluminal area of the small intestine, but also shows extraluminal extension (arrow). An enlarged mesenteric lymph node (arrowhead) is also observed.
HU = Hounsfield unit



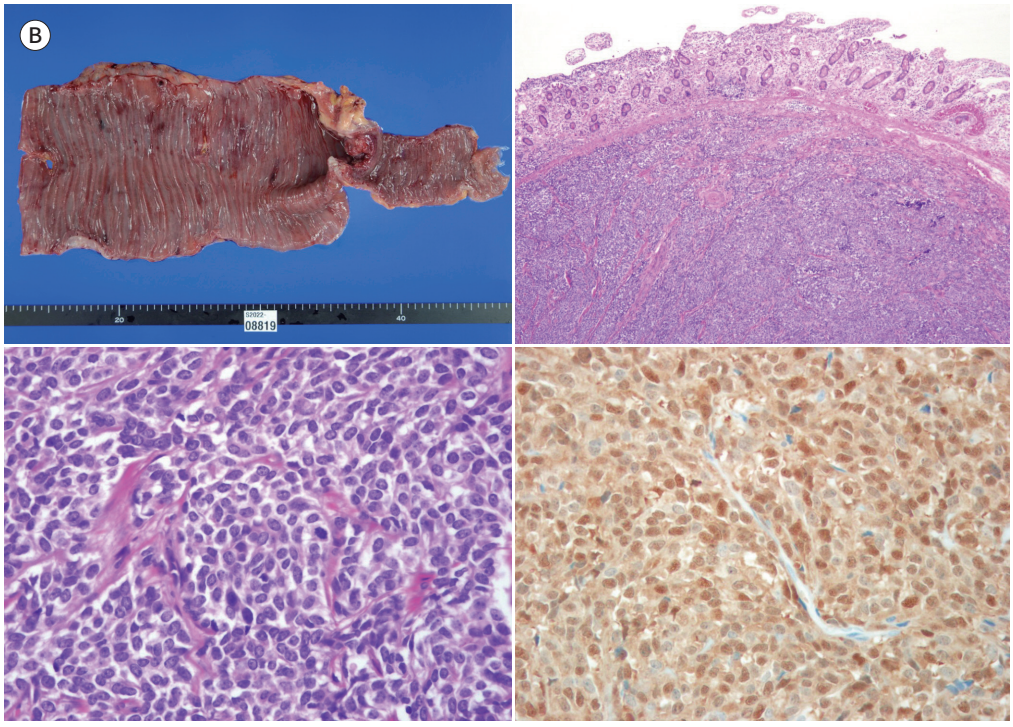
other nodular enhancing lesions in the cul-de-sac. A CT scan at the 4-month follow-up revealed a new ill-defined hypo-attenuated lesion in S7 of the liver, suggesting hepatic metastasis. Peritoneal and lymph node metastases had also progressed since the last examination (Fig. 1D).

This study was approved by the Institutional Review Board of our institution and the requirement for informed consent was waived (IRB No. 2022-11-021).

DISCUSSION

CCSLTGT is a rare malignant soft tissue tumor of the GI tract, which occurs most commonly in young adults. More than 60% of the patients are aged below 45 years, with a median age of 35 years at diagnosis (1). Patients usually present with abdominal pain, vomiting, abdominal distension, weight loss, and anemia. The etiology of CCSLTGT is unknown, but some case reports have suggested that a history of radiotherapy may be related to the pathogenesis of the disease (4). CCSLTGT occurs most commonly in the small bowel wall, but can also oc-

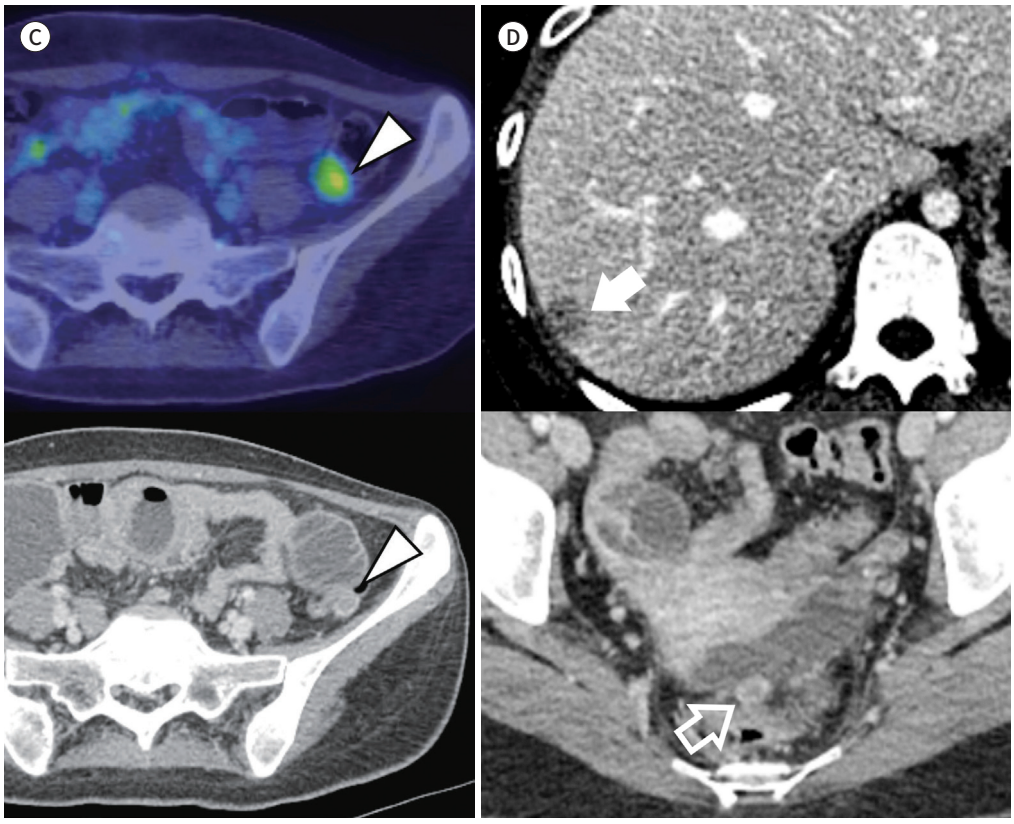
Fig. 1. Clear cell sarcoma-like tumor of the gastrointestinal tract in a 22-year-old female presenting with epigastric pain and weight loss.
B. Gross examination shows a mass constricting the small intestine (upper left). Histopathological findings indicate that the tumor cells have formed a mass under the mucosal layer (H&E stain, × 40) (upper right) and infiltrating tumor cells have a clear cytoplasm and are arranged in a pseudo-alveolar architecture (H&E stain, × 400) (lower left) with strong positivity for the S100 protein (S100, × 400) (lower right).
H&E = hematoxylin and eosin



cur in the stomach, colon, esophagus, and anal canal (1). Since a mass originating from the small bowel cannot be detected easily via GI endoscopic examination, CT is a particularly important modality to diagnose CCSLTGT.

To the best of our knowledge, only seven papers have described the radiological findings of a CCSLTGT in the small intestine (Table 1) (1, 5-9). The patients' ages ranged from 16 to 69 years, and the mean and median ages were 35.5 and 31.0 years, respectively. No definite gender predominance was noted (female = 4, male = 4). Half of the cases involved the ileum and the remaining ones involved the jejunum. There were six reports describing tumor morphology, among them, four patients showed bowel wall thickening (1, 5, 6, 7) and the others presented with an exophytic appearance or intraluminal protruding tumor (8, 9). Four reports illustrated the enhancement pattern of the tumor, and three patients showed mild to moderate progressive enhancement (5, 8, 9). However, one case report described a strong arterial enhancing mass with delayed progressive enhancement, similar to that observed in the present case (6). Most of the patients had liver or lymph node metastases at diagnosis. However, only our patient showed a peritoneal metastasis. Thus, if a small-bowel tumor shows asymmetric wall thickening with or without intraluminal protrusions or exophytic appearance and progressive enhancement pattern with lymph node metastasis in young adults, CCSLTGT should be considered as a differential diagnosis. Although the imaging findings of CCSLTGT were nonspecif-

Fig. 1. Clear cell sarcoma-like tumor of the gastrointestinal tract in a 22-year-old female presenting with epigastric pain and weight loss.
C. F-18 FDG PET/CT (upper) reveals moderate FDG uptake (maximum standardized uptake value = 3.0) by the nodular lesion in the left paracolic gutter (arrowhead), and preoperative axial CT image (lower) shows a small enhancing nodular lesion in the same area (arrowhead), suggesting peritoneal metastasis.
D. CT scan at the 4-month follow-up shows a hepatic metastasis (arrow) in S7 of the liver (upper) and a progressed peritoneal metastasis (arrow) in the cul-de-sac (lower).
FDG = fluorodeoxyglucose



ic, this report is the first to organize the imaging findings described in the existing literature.

Differential diagnosis should include NET, GIST, adenocarcinoma, and lymphoma. Since our patient showed a strongly enhancing small bowel mass, and NET is the most common primary small bowel malignancy; NET was primarily considered as the differential diagnosis. However, the patient did not exhibit a desmoplastic reaction and the tumor did not contain calcification, which commonly appears in NET (1). Moreover, small-bowel GIST usually appears as an endoluminal or exophytic small bowel mass. Therefore, it can be considered a differential diagnosis. However, GIST does not commonly show lymph node metastasis, and a large mass may be accompanied by various types of necrosis, hemorrhage, calcification, and ulceration (1). Unlike small-bowel GIST, our patient showed a mass with irregular margins and multiple lymph node metastasis and did not exhibit hemorrhage, calcification, or necrosis. Intestinal adenocarcinoma shows irregular intestinal wall thickening or localized intraluminal protruding soft tissue masses with mild to moderate enhancement (8). These findings are not clearly different from the imaging findings of a CCSLTGT. However, intestinal adenocarcinoma often occurs during the sixth or seventh decade of life (1). Lastly, most of the

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Table 1. Radiologic Information of 8 Patients with Clear Cell Sarcoma-Like Tumor of the Gastrointestinal Tract Originating in the Small Bowel

Case	Age (Years)	Sex	Location	Size (cm)	CT Findings	Metastasis at Diagnosis	Reference
1	30	Female	Jejunum	Not provided	- Irregularly thickened bowel loop with extraluminal growth of the mass - Mild to moderate heterogeneous progressive enhancement	Liver	5
2	16	Male	Ileum	4.1 × 2.4	- Intraluminal tumor - Arterial mild, delayed moderate homogeneous progressive enhancement	Lymph node	8
3	32	Male	Ileum	4.7 × 3.3	- Exophytic mass with probable intraluminal extension - Mild homogeneous enhancement	Not provided	9
4	33	Male	Ileum	2.5 × 0.9	- Segmental wall thickening - Arterial strong, delayed progressive enhancement	Lymph node	6
5	56	Female	Jejunum	Not provided	- Circumferential obstructive mass	Not provided	1
6	26	Female	Ileum	Not provided	- Heterogeneously enhancing mass	Not provided	1
7	69	Male	Jejunum	Not provided	- Local intestinal wall thickening	Lymph node	7
8	22	Female	Jejunum	3.3 × 2.7 × 3.6	- Intraluminal and extraluminal tumor - Progressive homogeneous strong enhancement	Lymph node Peritoneum	Present case

small intestinal lymphomas show diffuse, long, and uniform thickening of the intestinal wall or polypoid masses with mild to moderate homogeneous enhancement (8). On the other hand, our patient showed strong arterial enhancement with a mass protruding into the bowel lumen. Therefore, the diagnosis of lymphoma was less likely.

CCSLTGTs are extremely aggressive malignant tumors with a high probability of local recurrence and distant metastasis. Approximately 50% of the patients die within 3 years of diagnosis (3). It metastasizes mainly to the regional lymph nodes, liver, peritoneum, lungs, and bones. Radical resection and wide lymphadenectomy are the basic treatment options. Generally, conventional Adriamycin-based chemotherapy used in other non-small round cell soft tissue sarcomas is not effective, and there are no reports describing the efficacy of postoperative radiotherapy and chemotherapy (8). Therefore, further research should be conducted to improve the prognosis of this disease.

In conclusion, although CCSLTGT has low incidence, it is a malignant tumor that occurs at an early age, with an exceedingly high probability of metastasis. Therefore, radiologists need to consider this rare disease during the initial diagnosis of a progressively enhancing small-bowel tumor in young adults to ensure optimal management.

Author Contributions

Writing—original draft, P.S.Y.; and writing—review & editing, all authors.

Conflicts of Interest

The authors have no potential conflicts of interest to disclose.

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젊은 성인에서 복막 전이를 동반한 위장관의 투명 세포 육종 유사 종양: 증례 보고 및 문헌 고찰

박소연 · 서정욱*

위장관의 투명 세포 육종 유사 종양은 젊은 성인에서 국소 재발 및 원격 전이 가능성이 높은 드문 악성 중간엽 기원 위장관 종양이다. 저자들이 아는한 현재까지 영어로 보고된 소장에서 발생한 위장관의 투명 세포 육종 유사 종양의 영상 소견이 포함된 사례 보고서는 7예 뿐이다. 이에 저자들은 22세 여성 환자의 소장에서 발생한 위장관의 투명 세포 육종 유사 종양의 영상 소견을 보고하고 발표된 논문들의 영상 소견을 정리해 보고자 한다.

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