CASE REPORT

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A Rare Malignancy in an Adolescent: Desmoplastic Small Round Cell Tumor

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ABSTRACT Desmoplastic small round cell tumor (DSRCT) is a very rare condition. The peritoneal cavity is most frequently involved. Abdominopelvic computed tomography findings include omental or serosal masses, a dominant tumor, tumor calcification, liver metastasis, abdominal lymphadenopathies, acid and intestinal obstruction. A complete surgical resection including systemic implants, systemic chemotherapy and radiotherapy, hyperthermic intraperitoneal chemotherapy and stem cell transplantation can be used in the treatment of the disease. Since this tumor is very rare, we could only find case reports or a few large numbered patient series in the literature. Here in this case report, we aim to discuss the imaging findings of an adolescent patient at the time of admission.

Keywords: Desmoplastic small round cell tumor; adolescent; multidetector computed tomography; lymphoma

Desmoplastic small round cell tumor (DSRCT) is a very rare condition usually seen in the twenties and thirties. It is 5 times more common in boys than girls. The tumor is rarely seen in the pleura, bone, and salivary glands, however the peritoneal cavity is the most frequent localization.[1-4] Approximately 450 cases with this disease have been reported in the pathology literature and less than 200 cases in the radiology literature since the first description in 1989 by Gerald and Rosai.[5]

Here, we aim to discuss the imaging findings of an adolescent patient with DSRCT at the time of admission and follow-up, which was sent to the radiology clinic with a preliminary diagnosis of lymphoma.

CASE REPORT

A 15-year-old male presented with abdominal pain and vomiting after meals. In the physical examination, a mass in the abdomen was palpated. The borders of the mass could not be clearly evaluated. There was a prominent defense in the abdomen. After that, the patient was administered ultrasonography (USG) and computed tomography (CT). Widespread wall thickening was observed in all small bowel segments which was more prominent in ileocecal region in the USG. Numerous different lesions were detected in the peritoneal area, having hypoechoic and heterogeneous internal structure, with calcific components. There were also similar lesions located around the perihepatic area (Figure 1a). In addition, a hypoechoic-lobulated contoured lesion was observed in the periportal area. These lesions were hypervascular on color imaging (Figure 1b, Figure 1c). That appearance was initially considered as a lymph node package. A dominant mass, approximately 10*8*6 cm was observed in the rectovesical pouch (Figure 1d). Contrast-enhanced abdominal CT was performed due to these findings. Numerous nodular lesions involving cystic and calcific areas were observed in the perihepatic peritoneal areas (Figure 2a, Figure 2b, Figure 2c). A large, lobulated contoured hypodense mass

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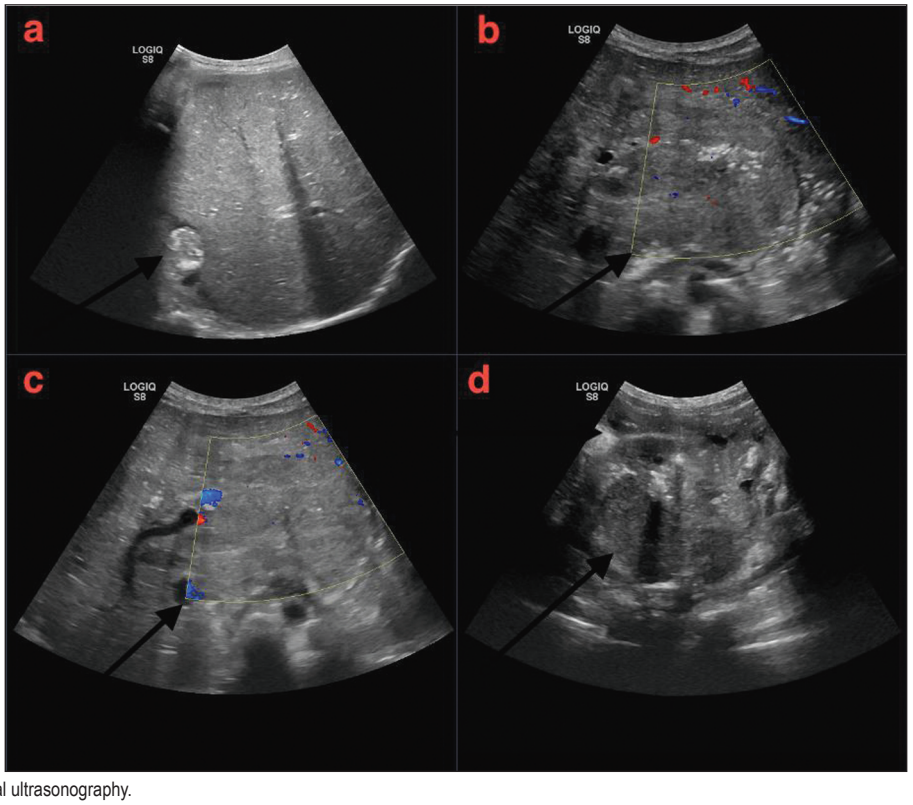

FIGURE 1: Abdominal ultrasonography.
A hypoechoic, lobulated contoured nodular lesion (marked with arrow) in perihepatic peritoneal area that has a calcific component (a). A large, hypoechoic, heterogeneous and lobulated contoured lesion, extending from the para-aortic area to the periportal area (marked with arrow). The lesion is hypervascular on color imaging (b, c). A 10*8*6 cm hypoechoic, heterogeneous lesion in the rectovesical pouch that has cystic and calsific compenents (marked with arrow) (d).

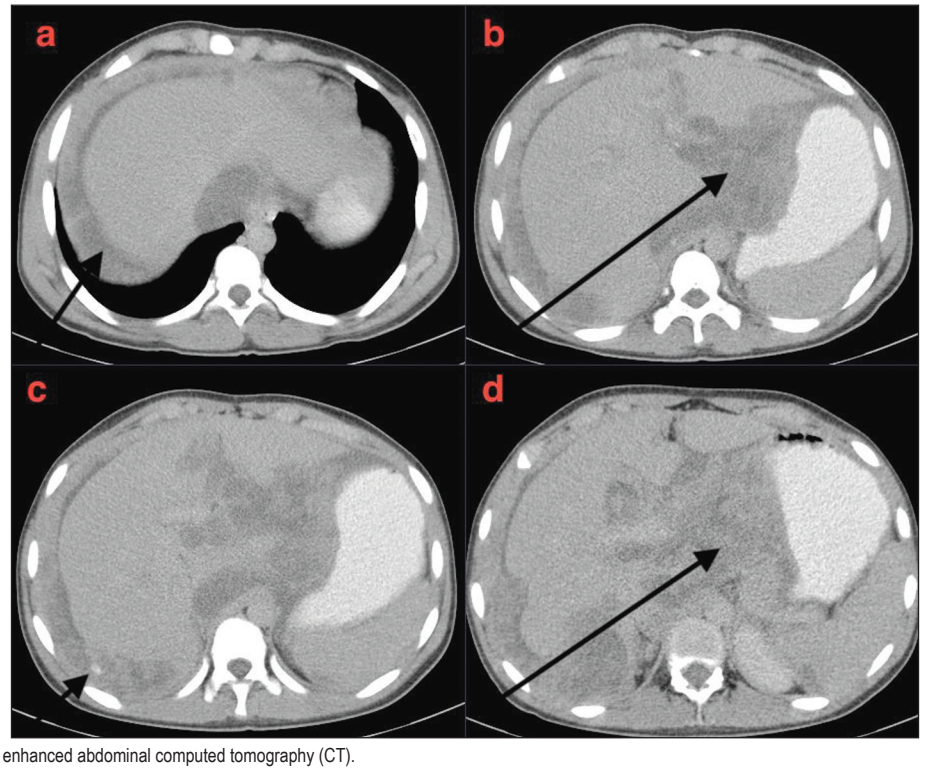

FIGURE 2: Contrast enhanced abdominal computed tomography (CT).
On axial abdomen CT images, there are multiple peritoneal hypodense implants in perihepatic area (a). Some of these have milimetric calcifications (marked with arrow) (c). On axial abdomen CT images, there is a large, lobulated contoured hypodense lesion extending from the paraaotic area to the periportal area (marked with arrow) (b, d).

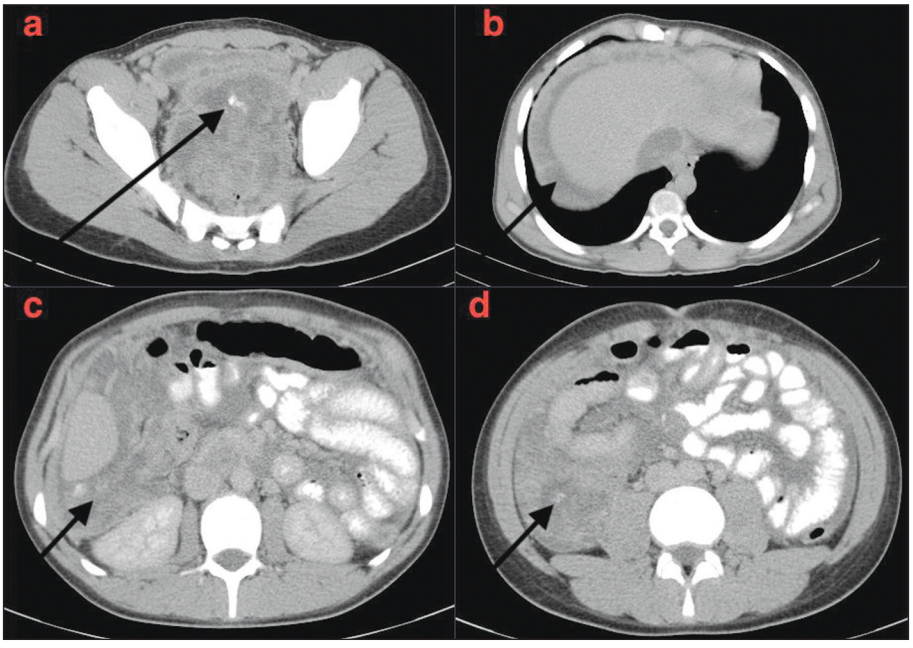

FIGURE 3: Contrast enhanced abdominal computed tomography (CT).
On axial abdomen CT images, there is a large, hypodense, heterogeneous lesion at the posterior of the bladder and the anterior of the rectum (marked with arrow). This lesion has cystic and calcific compenents (a). On axial abdomen CT images, there are hypodense peritoneal implants in the perihepatic area (marked with arrow) (b). On axial abdomen CT images, there are multipl hypodense, lobulated, intraperitoneal and retroperitoneal lesions that have cystic and calcific compenents (marked with arrow) (c, d).

was extending from the paraaotic area to the periportal area, that we evaluated as the lymph node package on the CT (Figure 2b, Figure 2d). In addition, a solid mass between the bladder and the rectum involving cystic and calsific areas was detected (Figure 3a). There were also large number of hypodense, lobular, intraperitoneal and retroperitoneal masses containing cystic and calcific areas within these lesions (Figure 3b, Figure 3c, Figure 3d). Fluorodeoxyglucose positron emission tomography/computed tomography (FDG PET/CT) was performed. These masses were hypermetabolic on FDG PET/CT with a maximum 13.8 SUV measurement (Figure 4). Microscopic examination of the biopsy revealed small round cell nests in the desmoplastic stroma in hematoxylin and eosin (HE) stained sections (Figure 5A, Figure 5B). The tumor cells were uniform, with hyperchromatic nuclei and scant cytoplasm. The desmoplastic stroma was consisted of spindle cells, variable collageneus and vascular structures. Immunohistochemically the tumor cells stained diffusely with pancytokeratin antibody, dot-like staining with vimentin and desmin antibodies. Their nuclei were stained with WT-1, INI-1 and FLI-1 antibodies (Figure 6A, Figure 6B, Figure 6C, Figure 6D). There was no immunoreactivity with CD99, myogenin, leukocyte common antigen (CD45), and alpha-smooth muscle actin, neuroendocrine markers (chromogranin A, synaptophysin, neuron spesific enolase, CD56). Accompanied by immunohistochemical findings, the diagnosis was desmoplastic small round cell tumor. After systemic chemotheraphy, bone marrow transplantation was done for the treatment but the patient died 3 months after the bone marrow transplantation.

The family of the patient had given an informed consent for publication of this case report.

DISCUSSION

DSRCT is a highly aggressive malignancy. Although various factors, have been accused for the etiology, is not fully known.[6] The incidence is from 0.2 to 0.5 people per million. It is characterized by a unique chromosome translocation that fails to suppress tumor growth in its pathogenesis.[7] It is classified as soft tissue sarcoma in small round cell tumors such as Ewing's sarcoma and primitive neuroectodermal tumor.[7] The largest series we have found in the literature is the relatively largest study with 94 patients

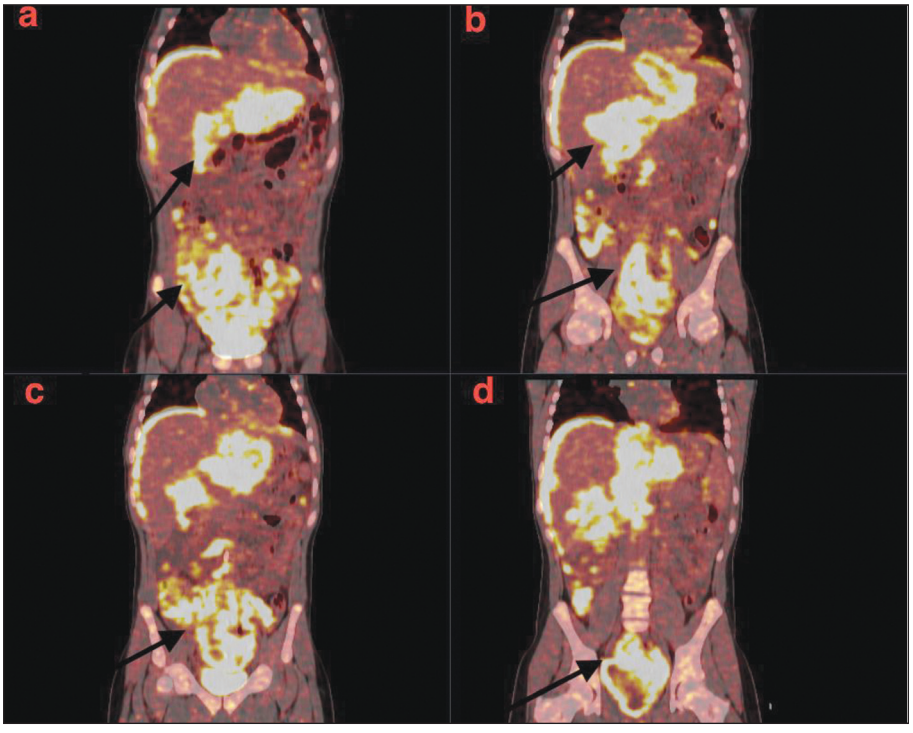

FIGURE 4: Fluorodeoxyglucose positron emission tomography/computed tomography (FDG PET/CT).
Lesions with increased pathological FDG metabolism in the aortakaval area (SUVmax: 13.8) (a, b), in intestinal area (SUVmax: 13.5) (a, b, c) and in the rectovesical pouch (SUV max: 12.7) (d) (marked with arrows).

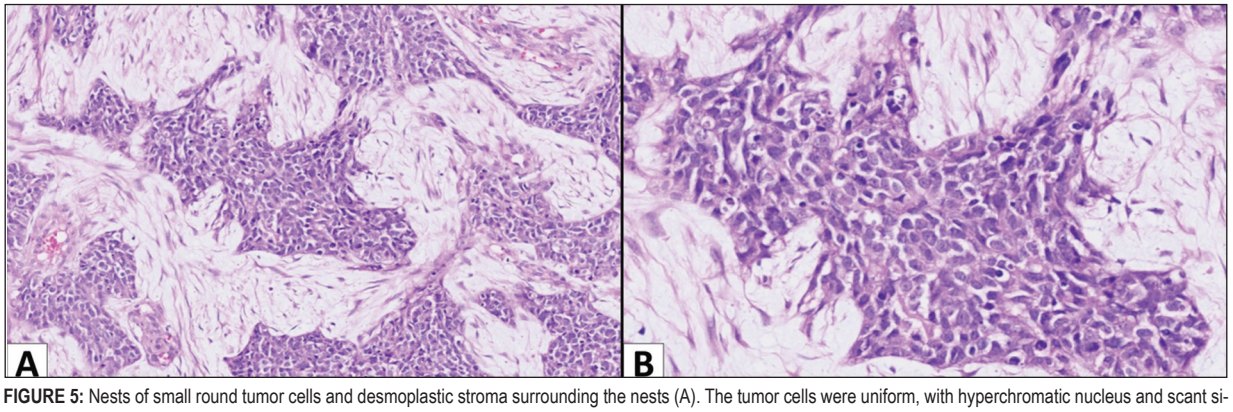

FIGURE 5: Nests of small round tumor cells and desmoplastic stroma surrounding the nests (A). The tumor cells were uniform, with hyperchromatic nucleus and scant sitoplasm (B). HE, x50, x100.

with abdominal DSRCT.[5] In DSRCT, the most frequently affected area is the peritoneal cavity by seeding. If the patient has more than one mass in the abdomen, there should be probably a large dominant mass accompanying the retrovesical or rectouterine region.[1,8]

Abdominopelvic CT findings include omental or serosal masses, a dominant tumor larger than 10 cm in size, tumor calcification, liver metastasis, abdominal lymphadenopathies, acid and intestinal obstruction. Tumor calcification has been reported between 22-100% in different studies in the literature.[5] In this patient, there were many lobulated contoured masses in the peritoneum, and there was a large mass accompanying them at the rectovesical region. Among these lesions, calcific foci were present especially in the dominant lesion at the rectovesical area and in the implants at the perihepatic region. In the dominant

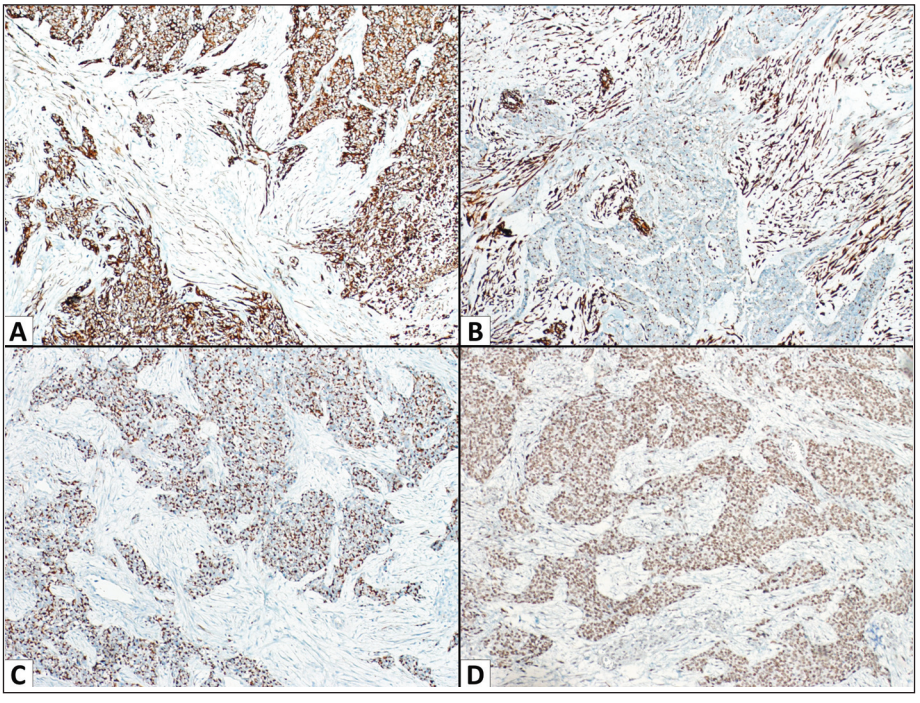

FIGURE 6: The tumor cells expressed pancytokeratin (A); stained with dot-like pattern for vimentin (B) and desmin (C), nuclear staining with INI-1 (D). x10.

mass of the patient, which was located at the posterior of the bladder, there was a significant increase in calcific foci after systemic chemotherapy. The increasing number of calcifications of tumor masses was noted in follow-up CT scans after radiotherapy in the study of Arora et al. and in some other studies has been reported in the literature.[9,10] Although it has been reported that metastases to the liver, lung, spleen or bones are often present at the time of diagnosis, there was no distant metastasis at the time of diagnosis in this patient.[11]

The differential diagnosis of the disease is broad and includes tumors such as rhabdomyosarcoma, germ cell tumors, neuroblastoma, lymphoma, soft tissue Ewing sarcoma and benign inflammatory processes.[5] Rhabdomyosarcoma usually occurs in the first 3 years of age, intraperitoneal and mesenteric masses seen in this tumor are smaller than DSRCT, calcification is very rare in the masses. Neuroblastoma is seen in the first years of life like rhabdomyosarcoma. Most of the neuroblastomas have calcifications, and the mass often contains necrotic and hemorrhagic foci. Sacrococcygeal teratomas are the most common germ cell tumor in the presacral region in childhood, but almost all of these tumors occur before the age of 2. Burkitt lymphoma is one of the most common malignant abdominal lesions in young adults. Burkitt lymphoma can originate from the terminal ileum, mesenteric lymph nodes and gastrointestinal tract, and can be seen in these areas as common intestinal wall thickening and peritoneal implants.[12] The most commonly involved area in Ewing sarcoma is the lumbosacral area. CT typically shows destruction of the bony cortex and a large soft tissue mass. Soft tissue masses are not typically associated with osseous metastases in DSRCTs.[13] Granulomatous infections such as tuberculosis may also be considered in differential diagnosis due to their omental cake forming feature. But peritoneal tuberculosis is also usually associated with large lymph nodes and bulky masses are uncommon in tuberculosis.[14] Considering all these differential diagnoses, we considered lymphoma in the first place with the patient's age and radiological features.

A complete surgical resection including peritoneal implants, systemic chemo-radiotherapy, hyperthermic intraperitoneal chemotherapy and stem cell transplantation can be used in the treatment of

the disease. However, despite all treatments, the prognosis is quite poor.[15] All of the treatment options above had been performed except hyperthermic intraperitoneal chemotherapy. The tumor relapsed 3 months after bone marrow transplantation and the patient died two years after the first diagnosis.

Until now, there are not many case reports about DSRCT, and most of them are in the pathology literature. We think that this case will contribute to the literature, since it includes both radiological and pathological features.

In conclusion, DSRCT is a rare tumor, most commonly presenting in adolescence and with significant radiologic features that can allow the radiologist to suggest the diagnosis. In particular, the presence of a dominant, heterogeneous solid mass in the retrovesical space, along with omental, mesenteric, or peritoneal surface masses, is a striking characteristic feature of DSRCT. Recognizing the imaging appearance of DSRCT plays an important role in the differential diagnosis and patient care, as this can expedite pathologic confirmation and commencing therapy.

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Güleç Mert Doğan, Ahmet Sığırcı, Merve Nur Güvenç;* ***Design:*** *Ahmet Sığırcı, Arzu Akyay, Sema Uğuralp;* ***Control/Supervision:*** *Güleç Mert Doğan, Sema Uğuralp, Ahmet Sığırcı;* ***Data Collection and/or Processing:*** *Güleç Mert Doğan, Sema Uğuralp, Ahmet Sığırcı;* ***Analysis and/or Interpretation:*** *Merve Nur Güvenç, Güleç Mert Doğan, Arzu Akyay;* ***Literature Review:*** *Güleç Mert Doğan, Merve Nur Güvenç;* ***Writing the Article:*** *Güleç Mert Doğan, Ahmet Sığırcı, Arzu Akyay;* ***Critical Review:*** *Ahmet Sığırcı, Arzu Akyay, Sema Uğuralp.*

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