CASE REPORT

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A Rare and Aggressive Abdominopelvic Tumor: A Case of Desmoplastic Small Round Cell Tumor in a Young Male

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ABSTRACT

Desmoplastic small round cell tumor (DSRCT) is a rare and aggressive mesenchymal tumor primarily affecting young adults, commonly originating in the abdomen and pelvic region. Here, we present the case of a 21-year-old male referred to our facility due to abnormal kidney function tests and a detected abdominal mass. The patient's laboratory results did not reveal any findings suggestive of a specific disease. The patient had experienced back pain for a month, which was managed initially with analgesics. Upon admission, hypertension was noted, and investigations revealed elevated urea, creatinine, uric acid, and lactate dehydrogenase levels. Abdominal ultrasound disclosed a hypoechoic lesion in the bladder pelvis, further confirmed by computed tomography (CT) scans, which showed lung and abdominal metastases. Positron emission tomography/CT scan highlighted hypermetabolic lymph nodes and skeletal lesions. Biopsy confirmed DSRCT with a Ki-67 proliferation index of 15-20%. The patient commenced VAC-IE chemotherapy. Key aspects of interest include the young age, the rapid metastatic spread, and the unexpected diagnosis. This case underscores the diagnostic challenges and aggressive nature of DSRCT, necessitating comprehensive management strategies. Our purpose is to describe the challenges and experiences in the diagnosis of DSRCT.

Keywords: Abdominopelvic tumor, Case report, Desmoplastic small round cell tumor, Young male

INTRODUCTION

Desmoplastic small round cell tumor (DSRCT) is an exceptionally rare and aggressive mesenchymal malignancy that primarily affects adolescents and young adults with a predilection for males [1,2]. The incidence of DSRCT is exceedingly low, with fewer than 500 cases reported in the medical literature since its initial description in 1989, highlighting its status as an orphan disease that poses significant diagnostic and therapeutic challenges [3]. The clinical significance of DSRCT lies in its aggressive behavior, propensity for widespread metastasis, and poor prognosis, necessitating a high index of suspicion and a multidisciplinary approach to management. Due to its insidious onset and non-specific symptomatology, such as abdominal pain, distension, or incidentally discovered palpable masses, DSRCT often poses a diagnostic challenge, frequently resulting in delayed diagnosis, which can significantly impact treatment outcomes and overall survival.

The genetic hallmark of DSRCT is the reciprocal translocation t(11;22)(p13;q12), which results in the fusion of the *EWSR1* gene on chromosome 22 with the *WT1* gene on chromosome 11 [4]. This translocation leads to the formation of the EWSR1-WT1 fusion protein, a chimeric transcription factor that dysregulates the expression of target genes involved in cellular proliferation, differentiation, and apoptosis, thereby driving the pathogenesis of DSRCT [4]. While the EWSR1-WT1 translocation is highly specific for DSRCT, its detection through molecular diagnostic techniques such as fluorescence *in situ* hybridization or reverse transcription polymerase chain reaction is crucial for confirming the diagnosis, particularly in cases with ambiguous histopathological features [4]. It is critical to differentiate DSRCT from other small round blue cell tumors.

CASE REPORT

A 21-year-old male was referred to our facility due to abnormalities in kidney function tests and an incidental finding of an abdominopelvic mass on ultrasound. The patient reported experiencing progressive lower back pain over the past month, which was initially managed with over-

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the-counter analgesics. Subsequently, he developed nausea and vomiting, prompting further evaluation. Laboratory investigations revealed elevated creatinine, urea, uric acid, and lactate dehydrogenase levels.

On admission, the patient's blood pressure was elevated, requiring antihypertensive treatment. Abdominal ultrasound showed a hypoechoic mass, (11×9 cm) in the posterior pelvis of the bladder. Further imaging with thoracic and abdominal computed tomography (CT) scans demonstrated multiple lung nodules consistent with metastatic disease, as well as heterogeneous soft tissue masses in the abdominal cavity. Positron emission tomography/CT confirmed hypermetabolic lymphadenopathy in the mediastinum, abdomen, and pelvis, along with skeletal metastases and hypometabolic ascitic fluid. A biopsy of the abdominal mass was performed, revealing small round blue cells embedded in a desmoplastic stroma, consistent with DSRCT. Immunohistochemical analysis showed positivity for desmin and EMA, and negativity for CK7, CK20, myogenin, MyoD1, and p63. The Ki-67 proliferation index was approximately 15-20% (Table 1, Figures 1, 2).

The patient, who received a pathological diagnosis of DSRCT based on biopsy results was referred to the medical oncology department. The patient developed post-renal acute kidney injury (AKI) due to pressure from the mass during follow-up. The AKI condition was corrected by placing a nephrostomy. It was considered that the existing hypertensive condition might be related to renal artery compression. The VAC-IE chemotherapy protocol was initiated. After the first cycle of the VAC-IE regimen, the patient developed a deterioration in general condition; therefore, the treatment was continued with the VAC regimen. After the third cycle, the patient was reported to have stable disease on an interim evaluation, and the treatment was extended to the sixth cycle. At the end of the 6th cycle, the patient is still being monitored with stable disease. Follow-up appointments are ongoing.

DISCUSSION

The current case presents widespread metastasis upon initial diagnosis, which contrasts with some reported cases, where the disease is localized or regional [5]. Early diagnosis and management are of importance, given the aggressive nature of DSRCT and the poor prognosis associated with metastatic disease [6]. Given the diagnostic complexity and the need for specialized expertise in managing this rare malignancy, a multidisciplinary approach involving oncologists, surgeons, and radiologists is essential for optimizing patient outcomes.

DSRCT can be confused with other abdominal tumors such as lymphoma, rhabdomyosarcoma, neuroblastoma, primitive neuroectodermal tumor, small cell mesothelioma, Ewing's sarcoma, and Wilm's tumor, especially in young patients [7]. DSRCT is typified by a proliferation of small, round, hyperchromatic neoplastic cells encased within a densely fibrotic, desmoplastic stromal component [8]. Immunohistochemical staining is critical for confirming the diagnosis and differentiating DSRCT from

Table 1. The patient's laboratory results at presentation

Urea	32 mg/dL (7-20 mg/dL)
Creatinine	1.85 mg/dL (0.6-1.3 mg/dL)
Uric acid	8.6 mg/dL (3.5-7.2 mg/dL)
Total protein	69 g/L (64-83 g/L)
Albumin	44 g/L (3550 g/L)
Aspartate aminotransferase (AST)	22 U/L (10-40 U/L)
Alanine aminotransferase (ALT)	24 U/L (7-56 U/L)
Alkaline phosphatase (ALP)	80 U/L (40-130 U/L)
Gamma-glutamyl transferase (GGT)	32 U/L (9-48 U/L)
Lactate dehydrogenase (LDH)	304 U/L (125-250 U/L)
International normalized ratio (INR)	1.4 (0.8-1.2)
White blood cell count (WBC)	9.07x10^9/L (3.9-10.2 x10^9/L)
Neutrophil count	7.15x10^9/L (1.5-7.5 x10^9/L)
Hemoglobin	12.5 g/dL [13.5-17.5 g/dL (man), 12.0-15.5 g/dL (woman)]
Platelet count	337 x10^9/L (150-450 x10^9/L)
Carcinoembryonic antigen (CEA)	0.7 ng/mL (<3 ng/mL)
Cancer antigen 19-9 (CA 19-9)	10.7 U/mL (<37 U/mL)
Alpha-fetoprotein (AFP)	<1.3 µg/L (<10 µg/L)
Erythrocyte sedimentation rate (ESR)	35 mm/hour [<20 mm/hour (man), <30 mm/hour (woman)]
C-reactive protein (CRP)	41.10 mg/L (<5 mg/L)

other tumors [9]. DSRCT typically shows positivity for desmin, WT1, and epithelial markers such as epithelial membrane antigen and cytokeratin. In most cases, DSRCTs resemble disseminated carcinomatoses in their clinical manifestation as well as cytomorphologically, even in young adult patients [9]. Fibrillary stromal fragment, clinical setting, and adjunctive immunocytochemical staining are most helpful for avoiding misdiagnosis [9].

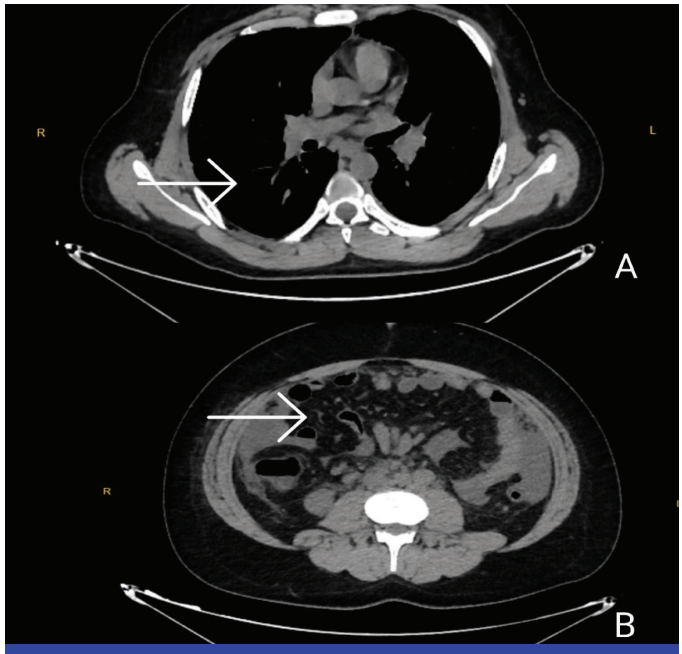

Figure 1. (A, B) The patient's thorax-abdomen CT scan arrows indicate metastases

CT: Computed tomography

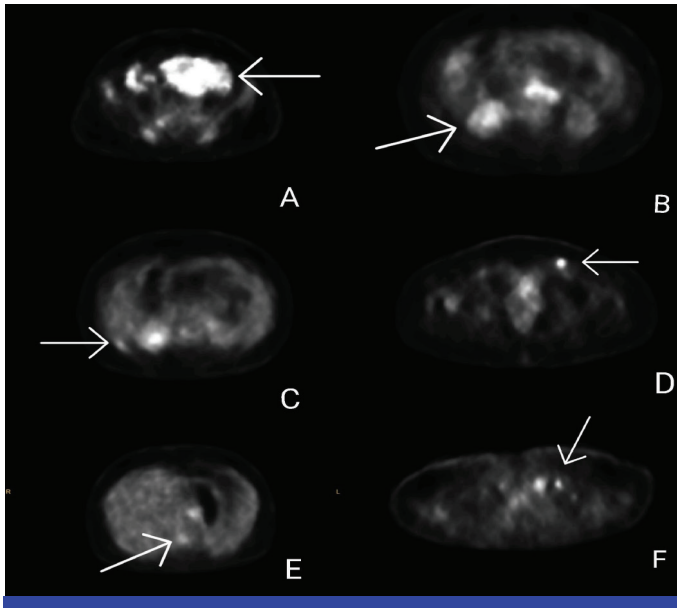

Figure 2. (A-F) The patient's PET/CT scan arrows indicate hypermetabolic lesions

PET: Positron emission tomography, CT: Computed tomography

Multimodal treatment strategies, including intensive combination chemotherapy, aggressive surgical resection, and adjuvant radiotherapy, have been the mainstay of DSRCT management and are associated with improved overall survival, according to recent studies. Recent studies have highlighted the role of Hyperthermic Intraperitoneal Chemotherapy and whole-abdomen radiotherapy in improving local disease control in patients with DSRCT [2,3]. The role of tyrosine kinase inhibitors such as pazopanib and sunitinib, which target platelet-derived growth factor receptor alpha (PDGFRα) and vascular endothelial growth factor (VEGF), has been explored in clinical trials, with some evidence of modest activity in patients with advanced DSRCT [10]. Clinical trials have assessed the efficacy of mTOR inhibitors in advanced and recurrent disease; some research trials in soft-tissue sarcoma and Ewing sarcoma include DSRCT patients, but few studies have been tailored to the specific clinical needs and underlying cytogenetic abnormalities characterizing this disease, such as the typical *EWSR1-WT1* gene fusion [2]. The presence of the EWSR1-WT1 translocation, which results in the upregulation of PDGFRα and VEGF, underscores the potential of targeted therapies in this disease [10]. Early clinical trials with IGF-1R inhibitors and PD-1/PD-L1 checkpoint inhibitors have shown promise, but further studies are needed to establish their efficacy in DSRCT [2]. The prognosis for DSRCT remains poor, with overall survival rates ranging from only 15% to 30% at five years, according to recent studies [7]. DSRCT is frequently diagnosed at an advanced stage, with widespread metastasis to the liver, lungs, and bones. The role of targeted therapies and immunotherapy in DSRCT is an area of ongoing research, and clinical trials are needed to evaluate the efficacy of these agents.

DSRCT remains a diagnostic challenge, particularly in young male patients in whom more common malignancies such as lymphoma and germ cell tumors are initially considered. This case highlights the importance of broad differential diagnoses when evaluating patients with an abdominopelvic mass, as uncommon malignancies can present similarly to more prevalent ones. The unexpected diagnosis of DSRCT underscores the necessity of a thorough diagnostic approach, including histopathological and molecular analyses, to accurately classify rare tumors. Early diagnosis and comprehensive treatment planning, incorporating multimodal therapies such as chemotherapy, surgery, and radiation therapy, remain essential for improving survival outcomes. As advancements in targeted therapies and immunotherapy continue to evolve, further research and clinical trials are warranted to explore novel therapeutic strategies that may enhance prognosis in patients with DSRCT.

Ethics

Informed Consent: Informed consent was obtained from the patient for publication of this case report, including any accompanying images and data.

Footnotes

Authorship Contributions

Surgical and Medical Practices: Y.S.T., E.S.S., A.B., B.E., En.S.Ş., Concept: Y.S.T., A.B., B.E., Design: Y.S.T., Data Collection or Processing: Y.S.T., E.S.S., A.B., Analysis or Interpretation: Y.S.T., E.S.S., Literature Search: Y.S.T., E.S.S., B.E., Writing: Y.S.T., E.S.S., A.B., B.E., En.S.Ş.

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