International Surgery Journal
Aregala P et al. Int Surg J. 2025 Aug;12(8):1423-1427
http://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: https://dx.doi.org/10.18203/2349-2902.isj20252305

Case Report

Intra-abdominal multifocal desmoplastic small round cell tumor: a case report of a rare neoplasm

Praneeth Aregala[1]*, Sreehari Gowda[1], Sachin Marda[1], Manjula Vijayanand Kelkeri[2], T. M. Jyoshna[1]

[1]Department of Oncology, Yashoda Super Specialty Hospitals, Somajiguda, Hyderabad, Telangana, India
[2]Department of Histopathology, Yashoda Super Specialty Hospitals, Somajiguda, Hyderabad, Telangana, India

Received: 19 June 2025
Revised: 15 July 2025
Accepted: 21 July 2025

***Correspondence:**
Dr. Praneeth Aregala,
E-mail: pranethraju.a@gmail.com

ABSTRACT

Desmoplastic small round cell tumour (DSRCT) is a rare, highly aggressive malignancy predominantly affecting adolescents and young adults. We report a case of multifocal DSRCT in a 31-years-old male who presented with complaints of abdominal pain, early satiety, weight loss for two months. Whole body 18FDG-PETCT scan revealed FDG avid lesions in the left hypochondrium and lumbar region abutting the stomach, splenic surface, right costophrenic and abdominopelvic lymph nodes, bilateral iliac fossa and in the pelvis. Ultrasound-guided biopsy of the left hypochondriac lesion done. Based on histopathological and immunohistochemical investigations, it was diagnosed to be a DSRCT with multifocal presentation. Patient received adjuvant chemotherapy with VAC/IE, post chemotherapy tumour response was suboptimal, so patient underwent cytoreductive surgery and HIPEC therapy. Despite advances in multimodal therapy, outcomes remain poor since the majority of patients present disease recurrence and die within three years. The dismal survival makes DSRCT an orphan disease with an urgent need for new drugs.

Keywords: CRS, Cytoreductive surgery, DSRCT, Desmoplastic small round cell tumor, HIPEC, Hyperthermic intraperitoneal chemotherapy

INTRODUCTION

The rare and aggressive mesenchymal cancer known as desmoplastic small round cell tumor (DSRCT) has distinctive chromosomal translocation characteristics.[1] Gerard and Rosai et al, provided the initial description of DSRCT in 1989, categorizing it according to the expression of various markers. With a median age of 20, DSRCT primarily affects young people.

When DSRCT is diagnosed, metastases are found and the most common symptom is abdominal discomfort.[2] In order to confirm specific chromosomal translocations, the main techniques now employed to diagnose DSRCT are histopathological, immunohistochemical and cytogenetic analyses.[3] We report a new, uncommon instance of abdominal DSRCT. This study aimed to explain the microscopic pattern and cytological features of DSRCT as well as our experience with diagnosis and treatment.

CASE REPORT

A 31-years-old male patient presented with complaints of abdominal pain occasionally, early satiety and weight loss to our hospital in May 2023.On per abdominal examination, approximately 15×8 cm firm immobile, non-tender swelling located in left hypochondrium reaching up to midline with poor superior boarders, rest of the abdomen was relatively normal. Patient underwent 18F-FDG PET-CT, which showed a large soft tissue mass lesion of size 13.8×6.8×10.2 cms (SUV max 17.8) involving left hypochondrium and lumbar region and it is

abutting and indenting upon body of the stomach along greater curvature, displacing and compressing transverse colon with indistinct fat planes, loss of fat planes with spleen, body of pancreas. A lesion pf size 6.5×4.6 cms with SUV max 11.6 present over the surface of spleen and largest pelvic lesion measures 3.4×3.3 cms with SUV max 12.8. FDG avid mildly enlarged retro pancreatic, portocaval, aortocaval, paraaortic, mesenteric, left external iliac lymph nodes noted, largest measuring 12 x 14 mm (SUV max 12.7). FDG avid serosal surface soft tissue density lesions in the bilateral iliac fossa and pelvis involving ileal loop, sigmoid colon and rectum, largest measuring 32×23 mm (SUV max 16.4). Mildly FDG avid small omento-peritoneal nodules noted in the left lumbar region measuring 6 mm.

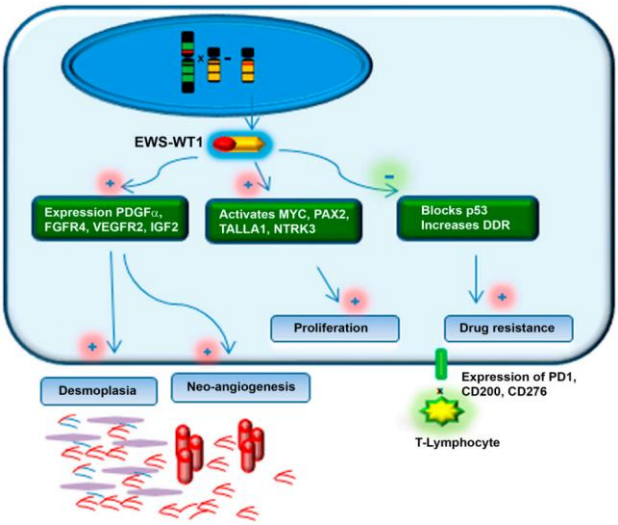

Figure 1: Schematic representation of EWS–WT1 fusion protein mechanism of action in desmoplastic small round cell tumor. Increase in tyrosine-kinase receptor expression, modulation of DNA replication proteins, activation of DNA-Damage Repair (DRR) machinery resulting in proliferation, desmoplasia, neo-angiogenesis and drug resistance.

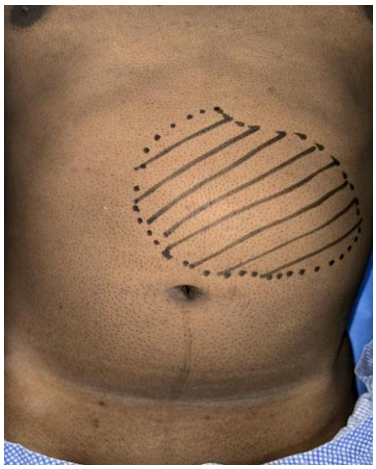

Figure 2: Mass mainly occupying left hypochondrium and epigastric region with ill-defined margins.

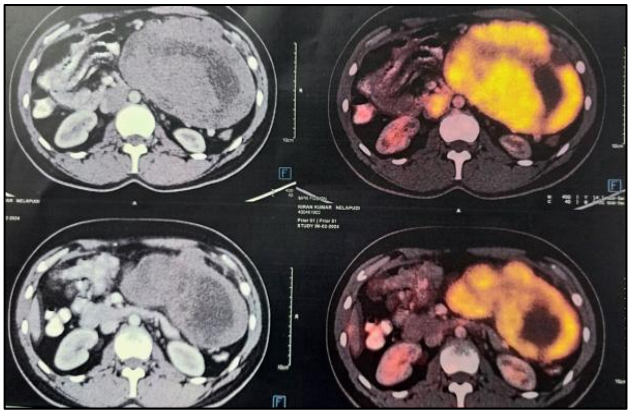

Figure 3: PET CT image showing bulky growth with ill-defined posterior margins of stomach.

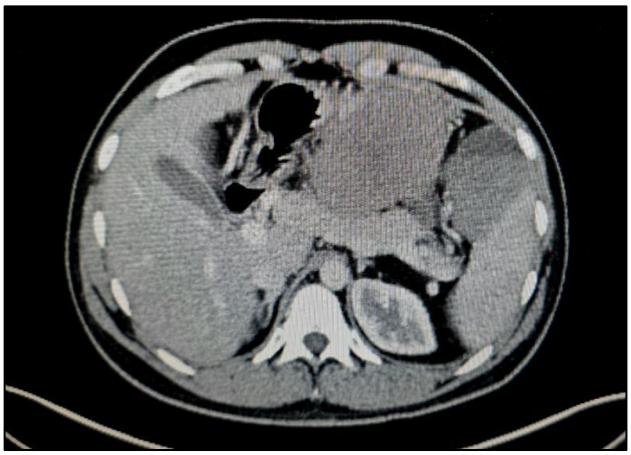

Figure 4: CT film showing lesion involving spleen and posterior stomach.

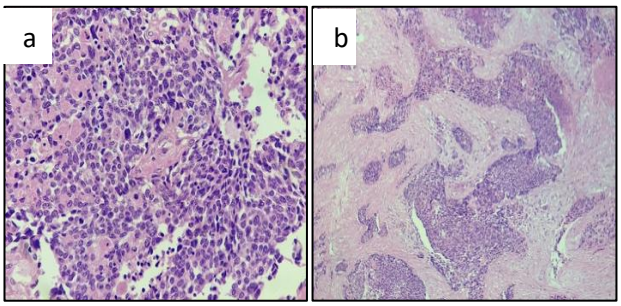

Figure 5 (a and b): Histological findings of DSRCT, showing sheets of round to oval cell with mitotic figures.

Ultrasound guided biopsy done which showed microscopically fragments of an epithelioid to spindle cell tumor composed of syncytial sheets, clusters and papillaroid arrangement of cells with hyperchromatic nuclei, mild anisonucleosis with focal areas of necrosis and hemorrhage. The cell clusters are interspersed with dense hyalinized and fibro collagenous bands, Mitosis in inconspicuous. On IHC, tumor cells are diffusely positive for PanCK desmin and CD99 while negative for SMA,

myogenin, NKX2.2, WT1, SALLA, ckit, Dogl and synaptophysin. Next Generation Sequencing revealed EWSR1-WT1 gene rearrangement at 22q12, confirming the diagnosis of a DSRCT. Tumor board decision was taken and planned for chemoport placement and VAC (vinorelbine, actinomycin, cyclophosphamide) regimen. A total of 17 cycles VAC and IE (ifosfamide and etoposide) maintain therapy.

Patient was reassessed with PETCT in September 2023, which showed very partial response to hypochondrial lesion, increase in splenic lesion size and new onset interaortocaval nodes. So patient was planned for cytoreductive surgery and HIPEC therapy. On October 1st 2024, patient underwent surgery with resection of tumor, distal pancreatectomy, splenectomy and all visible tumor deposits excision followed by Hyperthermic intraperitoneal chemotherapy with cisplatin (100 mg/m^2) over 90 min at 42°C. Post-operatively patient was discharged in hemodynamically stable condition on Pod-8. Patient was on follow up, as of now no evidence of recurrence.

Table 1: Memorial Sloan Kettering cancer center image-based risk stratification.

Risk	Ascites	Liver lesions	Estimated 5-Year OS (95% CI)
Intermediate	No	No	61% (40–76%)
High	Either ascites or liver lesions		16% (6–29%)
Very high	Yes	Yes	8% (1–29%)

DISCUSSION

DSRCT is thought to derive from the serosal surface of the abdominal cavity, including the peritoneum. The tumour site is responsible for the non-specific signs and symptoms. From highest to lowest frequency, the following are typical signs and symptoms that a patient might have: back pain, constipation, abdominal mass, abdominal pain and dyspepsia. At diagnosis, however, most patients have widespread metastases to intra- and extra-abdominal organs, such as the liver, pancreas, kidneys and lungs, in the skull rarely.[4]

It was named DSRCT by pathologists after finding that in tumour tissue the undifferentiated small round cell clusters were surrounded by a great deal of desmoplasia. DSRCT typically presents from 5–50 years of age with a mean age of 22 years. 85% to 90% of patients are male, but the female-to-male ratio is slightly higher in younger patients (i.e., ≤20 years of age at the time of diagnosis).[8]

Jordan et al, proposed a staging system for DSRCT where stage 1 would include patients with localized disease, confined to one or two abdominal sites, stage 2 would include extensive peritoneal disease, stage 3 would include peritoneal disease and liver metastases, and stage 4 would include patients with disease that has spread outside the abdominal cavity, including the lymph nodes.[5]

Molecular profile of Desmoplastic small round cell tumour

Although the precise pathophysiology of DSRCT is unknown, the hallmark event that causes the upregulation of several growth factor genes (particularly PDGFRα and vascular endothelial growth factor) and transcriptional factors linked to tumorigenesis is the chromosomal translocation t (11; 22)(p13; q12), which results in the fusion of the Ewing sarcoma RNA-binding protein 1 (EWSR1) and Wilms tumor suppressor (WT1) genes.[6] This translocation helps distinguish DSRCT from other small round cell neoplasms such mesothelioma, small cell carcinoma and Ewing's sarcoma. It is pathognomonic for DSRCT.[7] Histologically, cohesive nests of tiny, ovoid cells that stain blue amid a thick desmoplastic, collagenous stroma are the hallmark feature of DSRCTs.

It is important to note that the tumor cells are positive for mesenchymal markers (desmin, vimentin), epithelial markers (cytokeratin, EMA) and neural markers (neuron-specific enolase, CD57), thereby displaying a unique triphenotype IHC profile.

More recently, six DSRCT patients' genomic analyses identified 137 somatic mutations linked to biological processes: The mesenchymal-epithelial reverse transition and DNA damage response (DDR) network and mesenchymal-epithelial reverse transition/epithelial-mesenchymal transition (MErT/EMT), highlighting the significance of these mechanisms in drug resistance, tumour heterogeneity and aggressiveness. DSRCT and tumours belonging to the Ewing Sarcoma (ES) family have several characteristics. The EWS translocation is present in the majority of these malignancies. However, the elevated expression of Androgen Receptor (AR) is a significant molecular aberration that sets DSRCT apart from ES.[9,10]

Clinical presentation and diagnosis

DSRCT is more likely when several peritoneal implants are evident on CT or MRI. Most often, the first organ metastasis occurs in the liver. The pleura, the mediastinum and the lungs are the next most commonly discovered areas. Large intra-abdominal soft-tissue masses including omental and serosal surfaces, the absence of a defined organ of origin and extensive tumor implants are the most characteristic signs. The usual clinical presentation consists of palpable lumps, ascites, altered bowel habits or obstruction, significant weight loss, hydronephrosis and urine problems and persistent but vague abdominal pain.

Following chemotherapy, these volumetric changes might not be apparent right once and DSRCT is less likely to show morphologic response because of the stromal

composition of the lesions. Fluorodeoxyglucose positron emission tomography may therefore be helpful in post-treatment evaluation for the additional benefit of evaluating for distant metastases and assisting in disease staging.[11] An image-based risk categorization strategy based on the presence of ascites and/or serosal or parenchymal liver abnormalities at the time of diagnosis was more recently developed by the Memorial Sloan Kettering Cancer Centre (MSKCC).[12] Patients are divided into three risk groups using this risk stratification technique based on whether they were diagnosed with ascites or liver metastases.

At a specialized sarcoma centre, cases should be considered in a multidisciplinary meeting to develop a treatment strategy following a thorough diagnosis and evaluation. Similar treatment should be taken into consideration, even though DSRCT in children and young adults is frequently described independently in the literature.[13]

Induction chemotherapy

Systemic chemotherapy is typically used as the first line of treatment because of the high percentage of metastatic disease and the chemosensitivity of DSRCT. The regimens used include those designed for Ewing sarcoma, which are typically the P6 regimen (cyclophosphamide, doxorubicin, vincristine, ifosfamide and etoposide) or VAIA regimen (vincristine, dactinomycin, doxorubicin and ifosfamide) or those for soft tissue sarcoma, which contain ifosfamide and doxorubicin. doxorubicin and ifosfamide). When paired with the lack of extra-abdominal metastases and a low peritoneal tumour burden, the volumetric response to systemic chemotherapy serves as an indicator of tumour biology and helps identify appropriate surgical candidates.

Chemotherapy regimens used in various trials are equivalent to the standard Ewing's sarcoma method due to the parallels in molecular research. Kushner et al, used the P6 regimen, which comprised seven chemotherapy cycles with cyclophosphamide, doxorubicin, vincristine (HD-CAV), etoposide and ifosfamide. Surgery, radiation and myeloablative chemotherapy with autologous stem cell transplantation (ASCT) were sometimes used after these treatments and the outcomes were positive.[14]

Surgery

Although complete cytoreductive surgery (CRS) is the standard therapy, the microscopic residual disease is often present. Hence, Hyperthermic Intraperitoneal Chemotherapy (HIPEC) has been examined in some studies as an adjunct strategy but with conflicting results. A recent prospective cohort study of 20 patients showed that CRS+HIPEC prolongs Relapse Free Survival (RFS) (14.9 vs. 4.5 months, p=0.0012) and Overall Survival (OS) (44.3 vs. 12.5 months, p=0.0013) when compared to other tumours.[15] But the conflicting reports have also emerged previously.

In two retrospective studies, maximum surgical debulking has been shown to increase survival. They also discovered that improving survival and palliation are two areas where a multimodality strategy is quite helpful.[16] In a retrospective analysis, Subbiah et al, published the largest series of DSRCT patients (187 patients). In spite of the low cure rates, they showed that radiotherapy, surgery, intraperitoneal chemotherapy, removal of the primary tumor and its metastases and being younger than 30 years old all increased survival.[17]

Novel treatment methods

There is growing evidence that suggests the mTOR pathway may play a role in DSRCT. Although they haven't been confirmed yet, several case reports have indicated modest improvements in temporal progression when using mTOR inhibitors such temsirolimus and sirolimus. Given that the disease primarily affects young boys, targeting androgen receptors is yet another approach that has been investigated to determine whether testosterone plays a part in the disease process.[18] Other possible targets are being assessed, such as the Deoxyribonucleic acid (DNA) damage repair route and the c-met pathway. In preclinical research, immune checkpoint inhibitors and NTRK inhibitors have also demonstrated some effectiveness.

Differential diagnosis

The differential diagnosis of DSRCTs can be made with a spectrum of other round cell neoplasms, which includes ES, rhabdomyosarcoma, small cell carcinoma and mesothelioma. DSRCT is distinguished from the other neoplasms by the presence of EWS-WT1 translocation, ideally performed by PCR or FISH or by immunohistochemistry staining if the former are not available. Nowadays, large fusion panels using RT-PCR are able to help in differentiating small round cell sarcomas from the ES family and new entities are being recognized such as tumors harboring CIC-DUX4, BCOR-CCNB3 and CIC-FOX04 fusions.[19]

CONCLUSION

DSRCT is an uncommon condition that frequently affects children or adolescents with specific radiological characteristics. Molecular genetics, immunohistology-chemistry and morphology can all be combined to effectively identify DSRCT. Even with intensive care, the results are still subpar. To enhance patient prognosis, multimodal therapy, identification and clinical vigilance are necessary. DSRCT patients that respond well to initial therapy later experience intraperitoneal and extraperitoneal recurrences, which means that the prognosis is still dismal. Although trimodality treatment (induction chemotherapy, CRS with HIPEC and WART)

was associated with a median OS of 60 months, the median DFS was just 10 months.[13]

To increase knowledge of this rare disease and explore further the role of HIPEC, increasing patient numbers for data collection is essential. This highlights the importance of the International DSRCT registry/retrospective database, initiated by the Transatlantic Australasian Retroperitoneal Sarcoma Working Group (TARPSWG).

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Aregala P, Gowda S, Marda S, Kelkeri MV, Jyoshna TM. Intra-abdominal multifocal desmoplastic small round cell tumor: a case report of a rare neoplasm. Int Surg J 2025;12:1423-7.