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Desmoplastic small round cell tumor: A case report in a young Filipino woman and review of literature

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Abstract:

Desmoplastic small round cell tumor (DSRCT) is a rare and highly aggressive soft-tissue sarcoma that commonly presents late in the disease due to its nonspecific symptoms. Due to its rarity, there is still no standardized treatment regimen. A multimodal approach remains to be the cornerstone of treatment-complete cytoreductive surgery, chemotherapy, and radiotherapy. Despite advances in treatment, use of targeted and immunotherapy, an improved survival rate remains to be elusive. This paper presents a case of DSRCT in a 21-year-old Filipino patient as well as a review of the related literature of reported DSRCT cases in females and the emerging therapies for the disease.

Keywords:

Cell, desmoplastic, round, small, tumor desmoplastic small round cell tumor

Case Report

A 21-year-old nulligravid presented with a palpable abdominal mass and progressive abdominal enlargement. An abdominal computed tomography (CT) scan revealed multiple, heterogeneously enhancing lobulated masses in the abdominal and pelvic regions, the largest confluent focus was 13.3 cm × 13.3 cm × 13.7 cm, and nodular thickening of the peritoneum and omentum with few enhancing nodularities. Tumor markers showed elevated Lactate dehydrogenase (LDH) (598.4 U/L) and CA 125 (222.5 U/mL). She was advised to undergo surgery but did not comply.

Five months later, she returned to the emergency department with right lower quadrant pain with no other accompanying symptoms. Physical examination revealed direct tenderness over a palpable 20.0 cm × 18.0 cm × 12.0 cm irregular, solid mass. Rectal examination revealed a 5.0 cm × 5.0 cm solid, irregular mass compressing on the anterior rectal wall, 6.0 cm from the anal verge. The patient was admitted with a probable diagnosis of ovarian new growths, bilateral, with probable peritoneal carcinomatosis and rupture.

Emergency exploratory laparotomy revealed 1.7 L of serosanguinous ascitic fluid and a 21.0 cm × 18.0 cm × 9.0 cm solid mesenteric mass [Figure 1], matted to both small and large intestines, with omental implants. The right ovary was converted into a 12.0 cm × 10.0 cm × 7.0 cm solid, vascular mass adherent to the sigmoid colon and to the posterior parietal peritoneum. The left ovary was 15.0 cm × 11.2 cm × 4.0 cm and bilobulated [Figure 2]. The uterus had implants, the largest being 3.0 cm × 2.0 cm, and the sigmoid colon was studded with multiple tumor implants. There were multiple nodular implants on the cul de sac, with an aggregate measurement of 7.0 cm × 6.0 cm × 4.0 cm. Intraoperative diagnosis was ovarian new growth, malignant, and stage IIIC (omentum).

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The procedure included peritoneal fluid cytology, evacuation of ascites, left salpingooophorectomy, biopsy of the omental node and sigmoid implant, and insertion of a Jackson-Pratt drain.

Histopathology of the left ovary and sigmoid implant showed infiltrative nests with central necrosis [Figure 3]. The tumors tested positive for pancytokeratin, CK7, Epithelial membrane antigen (EMA), DESMIN, CD56, WT1, and inhibin [Figure 4], while negative for CK20, synaptophysin, p40, p53, SOX10, p16, and S100. Ewing sarcoma breakpoint region 1 (EWSR1) fluorescence *in situ* hybridization (FISH) analysis showed a t (11;22) chromosomal translocation [Figure 5], confirming the diagnosis of desmoplastic small round cell tumor (DSRCT).

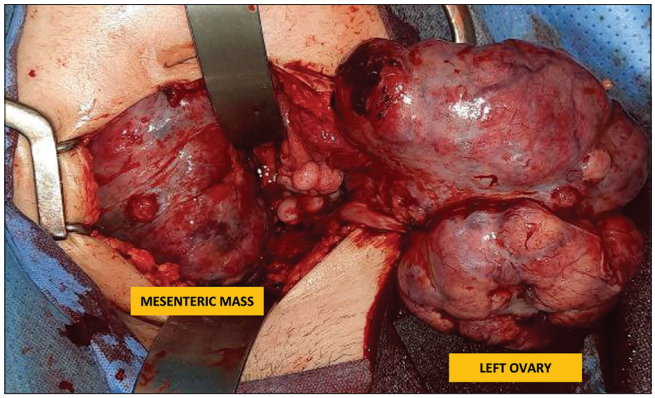

Figure 1: Intraoperative picture of a 21 cm × 18 cm × 9 cm solid, irregular, vascular mesenteric mass adherent and matted to the small and large intestines. Intimately related to it is the left ovary which was converted to 15.0 cm × 11.2 cm × 4.0 cm solid, bilobular mass

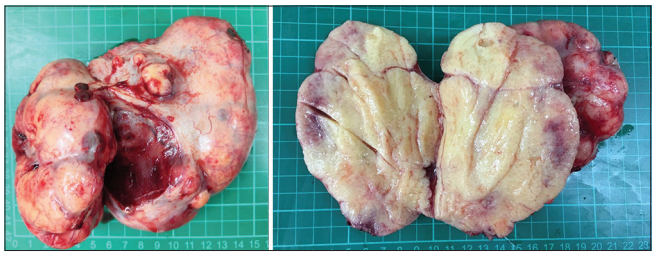

Figure 2: The left ovary is converted to a bilobed solid mass with smooth gray external surface measuring 15.0 cm × 11.2 cm × 4.0 cm with cream to cream-yellow cut surfaces. The attached fallopian tube measures 5.5 cm in length and 0.7 cm in diameter and has a smooth tan-brown glistening serosa

She was referred to medical oncology for comanagement, with plans to start chemotherapy using the P6 regimen (vincristine, doxorubicin, cyclophosphamide alternating with etoposide and ifosfamide). Unfortunately, the patient succumbed 4 months after diagnosis, before the initiation of chemotherapy.

Case Discussion

DSRCT is a rare and aggressive soft-tissue sarcoma first described in 1989.[1] Its incidence is approximately 0.2–0.5 cases per million.[2] It primarily affects adolescents and young adults, with a peak incidence at 20–25 years old.[2] Males are more commonly affected, with a male-to-female ratio of 4:1.[2] However, an increasing number of cases among females have been documented, with 37 patients reported in the literature [Table 1]. This index case is the 38th patient.[2,28]

Clinical manifestation

The symptoms of DSRCT are often nonspecific, including abdominal discomfort, abdominal enlargement, alterations in bowel and urinary habits, and unintentional weight loss, as observed in our patient. These symptoms typically present when the tumor has already infiltrated the peritoneum. Over 90% of cases originate within the abdominal cavity, commonly affecting organs such as the ovaries, pancreas, kidneys, urinary bladder, and testes.[2] Our patient presented with multiple abdominopelvic masses with involvement of the ovaries. This is supported by the 37 cases reviewed [Table 1]. Thirty-four of these, including this case, presented with masses that originate from the abdomen [Table 1]. Extraperitoneal metastases (EPM) are common and occur in up to 60% of patients at diagnosis.[2] The liver (35%–50%), lungs, pleura, and mediastinum are the most frequent sites of EPM.[2] Despite advances in treatment, prognosis remains poor, with a 5-year overall survival (OS) rate of 15%–30%.[1,28] Median survival ranges from 28 to 60 months, with median disease-free survival of 10–15.5 months.[29] Unfortunately, our

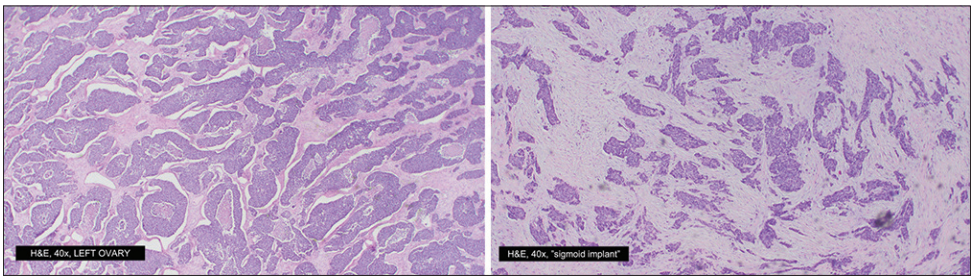

Figure 3: Scanning view of the tumor in the left ovary showing infiltrative nests, variable in size and often seen with central necrosis. Same morphology was seen in the sigmoid implant

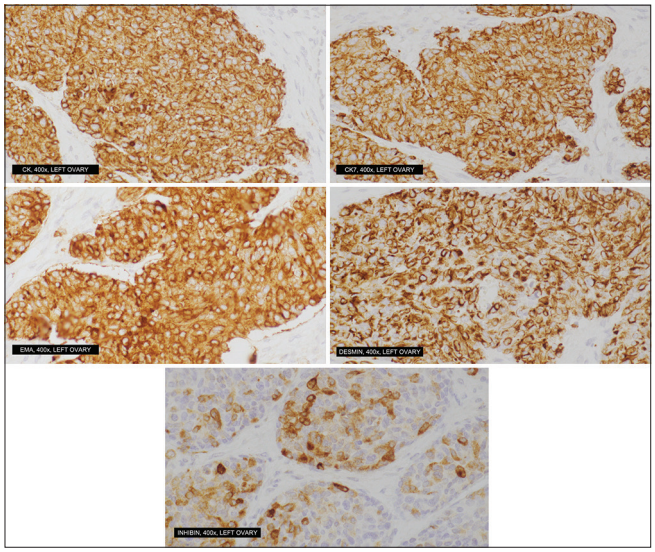

Figure 4: Immunohistochemistry studies showing the tumor tested positive for pancytokeratin, cytokeratin 7, EMA, desmin, and inhibin

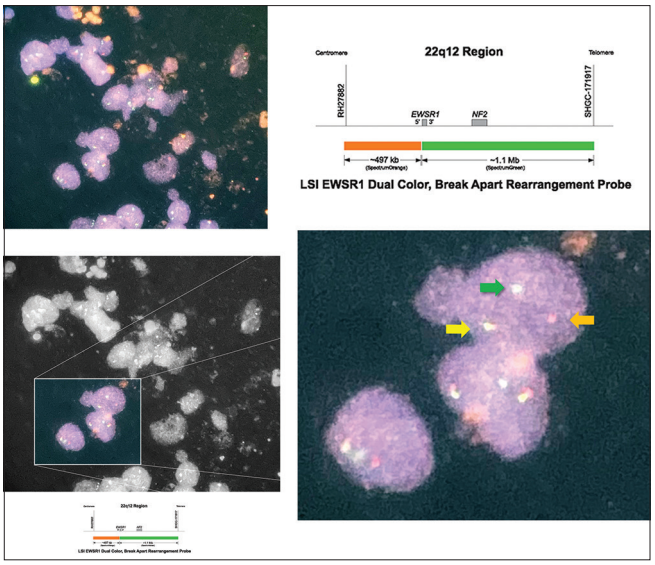

Figure 5: Fluorescence *in situ* hybridization study (FISH) with EWSR1 to demonstrate EWSR1 gene arrangement was done. An EWSR1 break apart probe was used. A normal gene is seen as a yellow signal due to the proximity of the orange and yellow probe on the normal fused gene. There is loss of the fused gene (yellow) as pointed by the green and orange arrows. EWSR1 FISH results are compatible with desmoplastic small round cell tumor

patient's clinical course reflected these statistics, having succumbed to the disease just 10 months after the onset of symptoms.

Diagnosis

The exact histogenesis of the disease remains uncertain, although it is postulated to originate from primitive mesothelial cells.[2] Histologically, DSRCT is characterized by uniform small, round, or ovoid tumor cells arranged in nests, with hyperchromatic nuclei and unclear cell borders. The stroma is desmoplastic, and immunohistochemistry shows coexpression of epithelial, mesenchymal, myogenic, and neural markers. The definitive diagnosis relies on the identification of the t (11;22)(p13;q12) chromosomal translocation, which generates the EWSR1-WT1 fusion gene. FISH analysis is critical in confirming the diagnosis, as demonstrated in this case [Figure 3]. Unfortunately, most published cases did not specify how the definitive diagnosis of DSRCT was arrived at.

Imaging studies such as ultrasound, CT, and magnetic resonance imaging (MRI) are nonspecific. On ultrasound, DSRCT appears as hypoechoic, lobulated, heterogeneous masses, whereas CT shows multifocal masses with poorly defined boundaries and uneven enhancement.[2] On MRI, T1-weighted images show mildly hypointense or isointense lesions, and T2-weighted images show heterogeneous high-intensity lesions.[2] Positron emission tomography scans are useful for detecting distant metastasis.[2]

Staging

There is no universally accepted staging system for DSRCT. Hayes-Jordan *et al*. proposed a staging system, which is under investigation at MD Anderson Cancer Center.[1] The proposed system includes:[1]

- Stage 1: Localized disease, confined to one or two sites in the abdomen or one site elsewhere
- Stage 2: Extensive peritoneal disease
- Stage 3: Liver metastasis with peritoneal disease
- Stage 4: Disease outside the abdominal cavity, including in the lymph nodes.
- Based on this staging, our patient would be classified as Stage 2.

Treatments

Treatment for DSRCT is largely based on protocols used for Ewing sarcoma, given their similar oncogenic pathways involving the EWS gene fusion.[30] Due to the rarity of DSRCT, no standardized treatment protocols exist. Multimodal therapy is commonly used, including cytoreductive surgery (CRS), alkylator-based chemotherapy, and radiotherapy.[30,31] Some centers also use hyperthermic intraperitoneal chemotherapy (HIPEC) at the time of CRS to treat microscopic residual disease. However, the role of HIPEC in improving survival is still under debate. There are also numerous novel therapies (e.g. immunotherapy) being used in refractory, recurrent, or metastatic DSRCT, but their effectiveness in improving progression-free survival (PFS) and OS remains to be unclear.

Surgery

As in other ovarian malignancies, optimal cytoreduction is essential for curative treatment. Removal of >90% of the tumor and resection of any residual disease >1.0 cm is

Table 1: Reported cases of desmoplastic small round cell tumor in women

Author (year)	Age (years)	Tumor location	Treatment	Outcome
Young *et al*. (1992)[3]	15	Pelvis (ovary)	Surgical debulking with multi-agent chemotherapy	DOD at 4 months
Young *et al*. (1992)[3]	15	Pelvis (ovary)	Surgical debulking, no chemotherapy	Secondary debulking at 7 months
Young *et al*. (1992)[3]	14	Pelvis (ovary)	Surgical debulking	None
Zaloudek *et al*. (1995)[4]	22	Pelvis (ovary)	Surgical debulking with bleomycin/E/P	DOD at 18 months
Fukunaga *et al*. (1996)[5]	60	Intraabdominal	Surgical debulking, Cis/5-FU	PR, then PD. AWD at 36 months
Ordoñez and Sahin (1998)[6]	34	Pelvis	Surgical debulking, Cis/E × 6 cy	CR. Recurred 6 months later, unresponsive to further chemotherapy. DOD at 19 months
De Lena *et al*. (1998)[7]	23	Intraabdominal	Surgical debulking, alternating three regimens: Cis/E/doxorubicin/bleomycin; gemcitabine/ifosfamide /dacarbazine; methotrexate/5-FU/folinic acid × 9 cy	Alive NED at 15 months
Slomovitz *et al*. (2000)[8]	11	Pelvis (ovary)	Surgical debulking, VDCy/IE and myeloablative chemotherapy	None
Elhajj *et al*. (2002)[9]	27	Intraabdominal and pelvis	Surgical debulking, Etop/Cis d1-3×3 cy	SD. PD after 3 months, unresponsive to further chemotherapy. DOD at 42 months
Parker *et al*. (2002)[10]	23	Pelvis	Surgical debulking, Cis/P × 4 cy	PD. No further treatment
Church *et al*. (2006)[11]	23	Intraabdominal and pelvis	Cis d1/doxorubicin d1-3 alternating with Cis d1/P + ifosfamide d1-5 × 6 cy	Resolution of symptoms for 1 year. PD unresponsive to further chemotherapy. DOD at 27 months
Church *et al*. (2006)[11]	29	Intraabdominal and pelvis	Cis d1/E d1-3/bleomycin d2, 8, 15 alternating with Cis + ifosfamid d1-5/P d1 × 6 cy	CR after 4 cy. Recurrence 6 weeks after treatment. DOD within 12 months
Khalbuss *et al*. (2006)[12]	19	Pelvis	Radiation and chemotherapy- unknown type	Rapidly PD. DOD at 3 months
Livaditi *et al*. (2006)[13]	13	Intraabdominal	IRS-38 + surgical debulking + blood stem cell transplantation	PFS: 9 months OS: 11 months
Bland (2007)[14]	33	Intraabdominal and pelvis	Surgical debulking, Cy/actinomycin/V alternating with ifosfamide /E × 1 cy	Rapidly PD. DOD at 3 months
Fang *et al*. (2008)[15]	13	Pelvis (ovary)	Surgical debulking, bleomycin/E/P, radiotherapy	DOD at 21 months
Fang *et al*. (2008)[15]	23	Pelvis (ovary)	Surgical debulking, myeloablative chemotherapy	Alive at 7 months
Kim *et al*. (2009)[16]	26	Intraabdominal	VDC/IE	PFS: NR OS: 9 months
Engohan-Aloghe *et al*. (2009)[17]	21	Pelvis (ovary)	Surgical debulking, unknown chemotherapy	Alive at 7 months
Ota *et al*. (2010)[18]	26	Pelvis (ovary)	Surgical debulking, VDCy/IE	DOD at 23 months
Ota *et al*. (2010)[18]	19	Pelvis (ovary)	Surgical debulking, bleomycin/E/P	DOD at 21 months
Nakayama *et al*. (2013)[19]	6	Pelvis (ovary)	Surgical debulking, VDCy/IE	DOD at 28 months
Nakayama *et al*. (2013)[19]	28	Pelvis (ovary)	Neoadjuvant IE/VAdCy, surgical debulking	DOD at 40 months
Nakayama *et al*. (2013)[19]	17	Pelvis (ovary)	Surgical debulking, IE/V adriamycin Cy with interval debulking followed by radiation therapy	Alive at 11 months
Xie *et al*. (2015)[20]	30	Pelvis (ovary)	Surgical debulking, V/adriamycin/Cy	Alive at 15 months
Al-Ibraheemi *et al*. (2018)[21]	14	Shoulder	Resection	N/A
	24	Neck	N/A	N/A
	31	Thigh	Chemotherapy + radiotherapy + resection	AWD
	32	Peritoneum	Resection	N/A
	43	Pelvis	N/A	N/A
	46	Uterus	Resection	N/A
Fagouri *et al*. (2018)[22]	16	Intraabdominal	Surgical debulking + doxorubicin/ifosfamid × 3 cy then PR + surgical debulking + docetaxel/gemcitabine/Cy × 2 cy then PD	PR then PD then DOD at 6 months
Nacef *et al*. (2019)[23]	46	Intraabdominal (omentum)	HD-CAV + surgical debulking	PFS: 1 months OS: NR
Atef *et al*. (2019)[24]	22	Pelvis	Surgical debulking + VAC-IE	N/A

Contd...

Table 1: Contd...

Author (year)	Age (years)	Tumor location	Treatment	Outcome
Altal *et al.* (2019)[25]	23	Liver lymph nodes	VDC	PFS: NR OS: 4 months
Zhou *et al.* (2021)[26]	27	Shoulder	VAC-IE	DOD at 11 months
Bambalan and Castro (2021)[27]	17	Intraabdominal (omentum) and pelvis (ovary)	Surgical debulking + VDCy/IE × 6 cy + second surgical debulking + EBRT 38 and HDR (total dose: 2100 cGy) + VIT × 2 cy + CP × 1 cy	PR, then PD. DOD at 36 months
Beltran and Toral (2022, unpublished)	21	Intraabdominal (mesentery) and pelvis (ovary)	Surgical debulking	DOD at 4 months

V: Vincristine, D: Doxorubicin, Cy: Cyclophosphamide, E: Etoposide, I: Irinotectan, T: Temozolamide, C: Carboplatin, P: Paclitaxel, HD-CAV: High dose cyclophosphamide, Adriamycin: vincristine, VDC: Vincristine: doxorubicin, cyclophosphamide, IRS 38: Oncovin, platino, adriamcyin, cyclophosphamide, VAC-IE: Vincristine, adriamycin, cyclophosphamide, with irinotecan and etoposide, CR: Complete response, PR: Partial response, PD: Progressive disease, AWD: Alive with disease, N/A: Not available, DOD: Dead of disease

critical for better outcomes. Any residual disease >1.0 cm resulted in a poor outcome according to Hayes-Jordan *et al*.[1] Loktev *et al* reported higher expression of androgen receptor in DSRCT, however, the use of receptor blocking agents remains controversial. However, due to the widely disseminated and adhesive nature of DSRCT, similar to the index case, complete resection is achievable in only 44%–65% of cases.[1,2,28] Of the 38 cases reported, including this case, 26 underwent primary debulking surgery, 7 received neoadjuvant therapy (chemotherapy or radiotherapy or both), and 5 were treated with chemotherapy alone [Table 1].

Hyperthermic intraperitoneal chemotherapy

HIPEC is used by some centers to manage microscopic residual disease following CRS. However, evidence regarding its survival benefit remains controversial. Some studies have shown no significant improvement in OS, particularly among patients with extra-peritoneal metastasis at the time of surgery.[1] A recent phase II study (NCT01277744) evaluating cisplatin-based HIPEC demonstrated improved 3-year OS in some patients, but its efficacy remains inconclusive, especially in those who undergo incomplete resections.[28] Among the 38 reviewed cases, none received HIPEC [Table 1]. In our case, the patient could have been a good candidate due to extensive peritoneal involvement and no EPMs, electrocorticography 1 performance status, and normal renal and hepatic function. However, the absence of histologic confirmation of DSRCT and failure to achieve complete cytoreduction precluded its use.

Chemotherapy

DSRCT is alkylator sensitive and dose responsive. The first-line regimen is the P6 protocol, as in Ewing sarcoma and metastatic neuroblastoma in children. It consists of high-dose cyclophosphamide, doxorubicin, and vincristine (HD-VDC) on cycles 1, 2, 3, and 6 given with cyclophosphamide (4200 mg/m^2), doxorubicin (75 mg/m^2), and vincristine (HD-VC) alternating with ifosfamide (9–12 mg/m^2) and etoposide (500 to 1000 mg/m^2) on cycles 4, 5, and 7.[1,2,28,31] Mello *et al* concluded that radiotherapy seems to improve survival after CRS, with better peritoneal PFS, PFS and OS.[29] This regimen, however, can be quite toxic.[1] Therefore, other Ewing sarcoma regimens [Table 2] are utilized to circumvent this toxicity.[2,28] Second-line chemotherapeutic agents for recurrent or persistent DSRCT include irinotecan 10mg/m^2/day (days 1–5 and 8–12) and temozolamide 100–125 mg/m^2/day (days 1–5) administered every 3 weeks.[28] Of the 38 cases reviewed, 29 patients received chemotherapy in either adjuvant (21), neoadjuvant (7), or primary treatment (5) setting [Table 1], with no standard regimen used. Due to its rarity, a standard treatment protocol is yet to be established.[1,2,28] However, available data suggest that patients experience the best outcomes with a multimodal approach. Neoadjuvant chemotherapy followed by surgery offers the highest survival benefit, with 3-year OS rates of 58%–62% and 5-year OS up to 45%.[1] Adjuvant chemotherapy after surgery also improves survival, with a reported 3-year OS of approximately 55%.[1] In contrast, chemotherapy alone, without surgical resection, is associated with poor prognosis, yielding 3-year OS below 30% and 5-year OS often under 15%.[1] Therefore, integrating chemotherapy with surgery remains the most effective strategy for improving survival in DSRCT.

Radiation

This condition requires consolidative adjuvant therapy to control intraperitoneal progression. WART has also been part of the multimodality treatment strategy. It is given at a median dose of 30 Gy with daily fractions of 1.5–1.55 Gy. In areas of residual tumor, boost is given up to 45–50 Gy. However, reports have consistently shown grade 3–4 acute hematologic and gastrointestinal toxicity.[1,2,28,31,33] Because of this, some have advocated the use of intensity modulated radiation therapy (IMRT) to reduce the toxicity associated with WART.[1,2,28,31,33] IMRT has shown to improve dose distribution, limiting radiation doses to organs at risk of toxicity, thus giving it a better toxicity profile.[1,2,28,31,33] There is no significant difference in OS between WART or IMRT, and though both have shown efficacy in locoregional

Table 2: Commonly used chemotherapeutic regimens in desmoplastic small round cell tumor

Protocol	Drug	Dose
First line		
VIDE	Vincristine	1.5 mg/m^2
	Ifosfamide	3000 mg/m^2
	Doxorubicin (Adriamycin)	20 mg/m^2
	Etoposide	150 mg/m^2
P6	Cyclophosphamide	4200 mg/m^2
	Doxorubicin	75 mg/m^2
	Vincristine	1 mg/m^2
	Ifosfamide	9–12 g/m^2
	Etoposide	500–1000 mg/m^2
VAIA	Ifosfamide	2000 mg/m^2
	Adriamycin	30 mg/m^2
	Vincristine	1.5 mg/m^2
	Actinomycin D	0.5 mg/m^2
VDC/IE	Vincristine	1.5 mg/m^2
	Doxorubicin	37,5 mg/m,^{2}
	Cyclophosphamide	1200 mg/m^2
	Ifosfamide	1800 mg/m^2
	Etoposide	100 mg/m^2
VDI	Vincristine	1.5 mg/m^2
	Doxorubicin	37.5 mg/m^2
	Ifosfamide	1800 mg/m^2
PAVEP	Doxorubicin	20 mg/m^2
	Ifosfamide	3000 mg/m^2
	Etoposide	150 mg/m^2
	Cyclophosphamide	300 mg/m^2
	Etoposide	75 mg/m^2
	Doxorubicin	40 mg/m^2
	Cisplatin	100 mg/m^2
Second line		
I/T	Irinotecan/Temozolamid	NR
T/C	Topotecan/Cyclophosmphamide	NR
GD	Gemcitabine	1000 mg/m^2
	Docetaxel	100 mng/m^2
HD-IFO	High-dose ifosfamide	NR
VIP	Etoposide	NR
	Ifosfamide	NR
	Cisplatin	NR

NR: Not reported, VDC/IE: Vincristine/doxorubicin/cyclophosphamide alternating with etoposide/ifosfamide, GD: Gemcitabine/docetaxel, PAVEP: Platinum (Cisplatin)/adriamycin/vepeside (etoposide)/etoposide/phosphamide (cyclophosphamide), VDI: Vincristine/doxorubicin/ifosfamide, VAIA: Vincristine/Adriamycin/ifosfamide/actinomycin D, VIDE: Vincristine/ifosfamide/doxorubicin/etoposide, VIP: Etoposide/ifosfamide/cisplatin, I/T: Irinotecan/Temozolamide, T/C: Topotecan/Cyclophosphamide, HD-IFO: High-dose ifosfamide

control of the disease, recurrences are still common, with 88% of cases treated with either of the two recurring intraperitoneally.[1,2,28]

Mello *et al*., concluded that radiotherapy seems to improve survival after CRS, with better peritoneal PFS, PFS, and OS.[29] However, their study had several limitations, such as being retrospective in nature, lack of statistical power (due to the small number of patients), and the need for randomized prospective studies to confirm their results. These studies suggest that WART, particularly when delivered using IMRT techniques, may improve local control and survival outcomes in DSRCT patients, albeit with a significant risk of toxicity.[34] This risk likely contributed to the limited use of WART in the reported cases of DSRCT among female patients. Only 4 out of the 38 cases reviewed used radiotherapy as part of their treatment protocol.

Immunotherapy/targeted therapy

Multiple targeted therapies have also been investigated or are currently being investigated as possible treatment options, when the first-and second-line treatments have already failed. Clinical trials pertaining to these targeted therapies are summarized in Table 3.[28,29,32] Translocation of EWSR1-WT1 gene fusion upregulates the expression of platelet-derived growth factor receptor-α, vascular endothelial growth factor (VEGF) and other proteins related to tumor and vascular cell proliferation (due to the loss of the normal function of the zinc finger region of the WT1 gene which acts as a repressor of transcription).[28-30] These upregulated factors can be potential targets for therapy.[32]

Tyrosine kinase inhibitors have shown inadequate activity with DSRCT. No benefit was obtained with the use of Imatinib[2,28-32] while Sorafenib and Sunitinib demonstrated limited efficacy.[2,28-32] Apatinib and Anlotinib resulted in improvement of symptoms with PFS of 4 months but only in one patient.[2,28-32] Pazopanib has shown the most promise with clinical benefit in 62%–78% of cases, reporting a median PFS of 4.5–9.2 months and an estimated median OS of 12.5–15.7 months.[2,28-32]

Anti-VEGF therapies are also considered. The use of Bevacizumab resulted in an OS of 34 months among patients who underwent partial resection.[2,28,29,31,32] It is also currently being investigated with irinotecan, temozolomide as a combination with P6 chemotherapy for newly diagnosed DSRCT.[31,32] Ramicirumab, another anti-VEGF therapy, is being analyzed as well with chemotherapy.[2,28-30]

Agents that target the PI3K/AKT/MTOR are also investigated. Rapamycin causes apoptosis in JN-DSRCT-1 cells derived from a patient with DSRCT.[2,28,29] Temsirolimus use resulted in PFS up to 10 months. When used in combination with cixutumumab, stable disease was achieved in 2/3 of patient.[2,28-30]

IgG1R antibody inhibitors are another option. Cixutumumab, a monoclonal IgG1antibody against human insulin-like growth factor-1 receptor, as previously mentioned, has achieved stable disease

Table 3: Clinical trials using novelty therapy for desmoplastic small round cell tumor patients

Phase	Conditions	Interventions	Clinical trials.Gov identifier
Target therapy			
I	Newly diagnosed DSRCT	Irinotecan + temozolomide + bevacizumab + high-dose alkylator-based chemotherapy	NCT01189643
I/II	Refractory DSRCT	Imatinib mesylate	NCT00417807
II	Relapsed/refractory DSRCT, ES	AMG 479 (anti-IGF-1R)	NCT00563680
II	Relapsed/refractory DSRCT, ES	Imatinib mesylate	NCT0062205
II	Nonresectable/metastatic sarcoma or endometrial carcinoma with somatic deficient MMR, including DSRCT	Nivolumab plus Ipilimumab	NCT02982486
I/II	Relapsed/refractory neuroblastoma, osteosarcoma, and other GD2 + solid tumor cancers	Humanized anti-GD2 x anti-CD3 bispecific antibody (hu3F8-BsAb)	NCT03860207
Target therapy + chemotherapy			
I	Unresectable/metastatic soft tissue sarcoma, including DSRCT	Cixutumumab and doxorubicin	NCT0072014
I/II	Relapsed/refractory DSRCT	Ramucirumab + cyclophosphamide + vinorelbine	NCT04145349
II	DSRCT, ES, PNET	VDC/IE + temsirolimus/sorafenib/ temozolomide/bevacizumab	NCT01946259
I/II	Relapsed or refractory desmoplastic small round cell tumor	Prexasertib, irinotecan, temozolomide	NCT04095221
I/II	NTRK-fusion-positive advanced or metastatic solid tumors	PBI-200 (TRK inhibitor)	NCT04901806
Immunotherapy			
I	B7H3 expressing relapsed/refractory solid tumors, including DSRCT	Enoblituzumab	NCT02982941
I	Relapsed or refractory non-CNS solid tumors, including DSRCT	B7H3-specific CAR T cells versus B7H3 and CD19-specific CAR T cells	NCT04483778
I	Relapsed or refractory non-CNS solid tumors	EGFR-specific CAR T cells versus EGFR and CD19-specific CAR T cells	NCT03618381
II	Unresectable locally advanced or metastatic disease, which is resistant or refractory to standard therapy, or for which standard therapy does not exist	Pembrolizumab	NCT03012620
I	Select sarcoma patients, including myxoid liposarcoma and other sarcomas that share similar chromosomal translocations to Ewing sarcoma	Seclidemstart (LSD1 inhibitor)	NCT03600649
I	Refractory B7-H3-expressing melanoma, SCCHN, NSCLC, Urothelial Cancer, and other B7-H3-expressing cancers	Enoblituzumab + Pembrolizumab	NCT02475213
I	Refractory B7-H3-expressing melanoma, SCCHN, NSCLC, and other B7-H3-expressing cancers	Enoblituzumab + Ipilimumab	NCT02381314
Radio immunotherapy			
I	Solid tumors involving the peritoneum, including DSRCT	RIT with 131I-8H9	NCT01099644
II	Solid tumors involving the peritoneum, including DSRCT	131- I-omburtamab + radiation	NCT04022213
I	Relapsed or refractory malignant solid tumors, including DSRCT	CLR 131 IV administration	NCT03478462
Other therapy			
II	Refractory/recurrent solid tumors, including DSRCT	Allogeneic hematopoietic stem cell transplantation after chemotherapy	NCT04530487

VDC/IE: Vincristine/doxorubicin/cyclophosphamide alternating with etoposide/ifosfamide, NSCLC: Nonsmall cell lung cancer, SCCHN: Squamous cell carcinoma of the head and neck, DSRCT: Desmoplastic small round cell tumor, IV: Intravenous, RIT: Radioimmunotherapy, PFS: Progression free survival, IP: Intraperitoneal, MDT: Maximum tolerated dose, DLT: Dose limiting toxicity, NTRK: Neurotropic tyrosine receptor kinase, ES: Ewing Sarcoma, MMR: Mismatch Repair, PNET: Primitive neuroectodermal tumor, EGFR: Epidermal growth factor receptor, CAR: Chimeric antigen receptor T cells, PBI: paltimatrectinib, CNS: central nervous system, AMG: ganitumab

when given in combination with temsirolimus.[2,28,29] Ganitumab, a human anti-type 1 insulin-like growth factor receptor antibody, has also induced stable disease or partial response for ≥ 24 weeks with clinical

benefit in 25% of cases and reported median PFS of 19 months.[2,28-32]

PARP inhibitor is another treatment option being studied because Mello *et al*. and Loktev and Shipley, discovered that PARP1 expression was seen in 100% of clinically derived DSRCT 2 tumor tissues.[2,28-30] *In vivo* studies for now showed that olaparib + temozolomide have synergistic effects, inducing cell cycle arrest in N-DSRCT-1 cell.[2,28-30]

Immune checkpoint, like PD-1 has been found to be expressed by DSRCT cells. Trials employing the use of the immune checkpoint inhibitor pembrolizumab are currently ongoing.[2,28-32]

Loktev *et al*. reported higher expression of androgen receptor in DSRCT, however, the use of receptor blocking agents remains controversial. *In vitro* studies using flutamide demonstrated efficacy by inhibiting the growth of tumor cells. Bicalutamide + leuprolide showed clinical tumor benefits for 3–4 months.[2,28-30]

Autologous stem cell transplant (ASCT) has been used with promising results. Complete response was seen after CRS, chemotherapy (VAC + VAP), and ASCT. Even among patients with residual tumors, ASCT improved disease-free survival and OS.[2,28-32] It also showed improved OS among those in remission.[2,28-32] However, further studies are still needed to validate these findings.

Another emerging alternative treatment for pediatric solid tumors to which DSRCT belongs to is immune therapy. However, this has been proven to be difficult for DSRCT because of their low tumor mutation burden, making them immunologically ignorant.

These novel therapies are overall safe and well tolerated, but most agents mainly produce disease stabilization rather than disease regression. Moreover, trials with a significant number of patient population are still lacking, such that the use of these agents is currently limited to clinical trials. Due to the paucity of data regarding its efficacy in DSRCT, none of the 38 cases reviewed, including the index case, made use of novel therapeutic agents.

In summary, multimodal treatment is still the preferred treatment regimen for DSRCT, improving OS (median survival, 37.7 months).[1,2,28,35] Factors predictive of 3-year OS include the absence of EPM, complete surgical resection, postoperative WAP-RT, and postoperative chemotherapy.[1,2,28]

Conclusion

DSRCT is a rare and aggressive tumor with a poor prognosis, often diagnosed at an advanced stage due to its nonspecific clinical features. Early diagnosis, genetic testing, and accurate staging are essential for management. Treatment involves a multimodal approach, including optimal CRS, chemotherapy (particularly the P6 regimen), and radiotherapy. Emerging therapies, including targeted treatments and stem cell transplantation, show promise but require further research. Despite treatment advances, survival remains limited, and the need for improved therapies remains urgent.

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Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Authorship contributions

Julienne Katrina B. Beltran MD - Involved in the conceptualization, methodology, data curation, writing of the original draft, review and editing.

Jean Anne B. Toral MD - Involved in review and editing of the draft.

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Conflicts of interest

There are no conflicts of interest.

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