http://jmscr.igmpublication.org/home/
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v10i7.06

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Desmoplastic Small Round Cell Tumor of Small Intestine: A Case Report and Review of Literature

Authors
Pawan Kumar, Diptajit Paul*, Abhishek Soni, Vivek Kaushal
Department of Radiation Oncology
Pt B D Sharma PGIMS, Rohtak, Haryana India 12401
*Corresponding Author
Dr Diptajit Paul
Pt BD Sharma PGIMS, Rohtak, Haryana India 124001

Abstract
Desmoplastic small round cell tumor (DSRCT) is an uncommon entity characterized by sarcomatous abdomino-pelvic mass arising from serosal surface of peritoneum. Till date, only a few cases of DSRCT has been reported in literature. Here we presented a case of small intestinal DSRCT in a young male presented with peri-umbilical pain of 3-months duration. Patient underwent exploratory laparotomy with resection of large ileal mass. Histopathology of surgical specimen revealed desmoplastic small round cell tumor. Patient was planned for adjuvant chemotherapy with VAC regimen. But after receiving 2-courses, patient condition deteriorated and patient was succumbed to death. This case report demonstrated the aggressiveness of DSRCT.
Keywords: *Adjuvant chemotherapy, desmoplastic small round cell tumor, exploratory laparotomy, VAC regimen.*

Introduction

Desmoplastic small round cell tumor (DSRCT) is an infrequent but fatal tumor. Till date, less than 1000 cases have been documented in literature. Here we presented DSRCT of small intestine in a young male treated by surgery and adjuvant chemotherapy.

Case Report

A 29-year young male, without any other comorbidities or chronic illness, presented with peri-umbilical pain for 3-months duration, which was insidious in onset, dull aching in nature, mild to moderate in intensity and not radiating to any other site. It was associated with few episodes of vomiting which was non-bilious, non-projectile, but not associated with any hematemesis. He also had off and on history of high grade fever for last 1-month, not associated with any rigor or chill, and relieved by taking antipyretics. He had no associated history of altered bowel habits, blood in stools, weight loss, anorexia, headache or any chest discomfort. Patient was vegetarian, non-smoker, non-alcoholic. There was no history of malignancy in family members. General physical examination was normal. Per abdomen examination revealed rigid, tender abdomen with a vaguely palpable mass around umbilicus.

Complete hemogram and routine blood biochemistry parameters of the patient were within normal limits. Contrast enhanced computed tomography (CECT) of abdomen revealed irregular ileal wall thickening with a large heterogenous lesion in right iliac fossa, measuring 11.0 × 8.0 cm and associated luminal narrowing at places along with multiple retro-peritoneal lymph nodes largest of size measuring 2.7 × 1.5 cm [figure 1]. Patient underwent exploratory laparotomy with ileostomy and mass of size 10 × 10 cm was resected along with approximately 50 cm of distal ileum. Histopathology of surgical specimen revealed small round blue cell tumor with trabecular and nesting pattern, which also involved lympho-vascular structures. The neoplastic cells illustrated wide spread positivity for vimentin immunocytochemical stain and stain for cytokeratin, cluster differentiation 117 (CD 117), leukocyte common antigen (LCA), cluster differentiation 3 (CD 3), cluster differentiation 20 (CD 20), cluster differentiation 56 (CD 56) were negative. The histopathological appearance and immunohistochemical profile of the biopsy tissue confirmed it to be desmoplastic small round cell tumor (DSRCT).

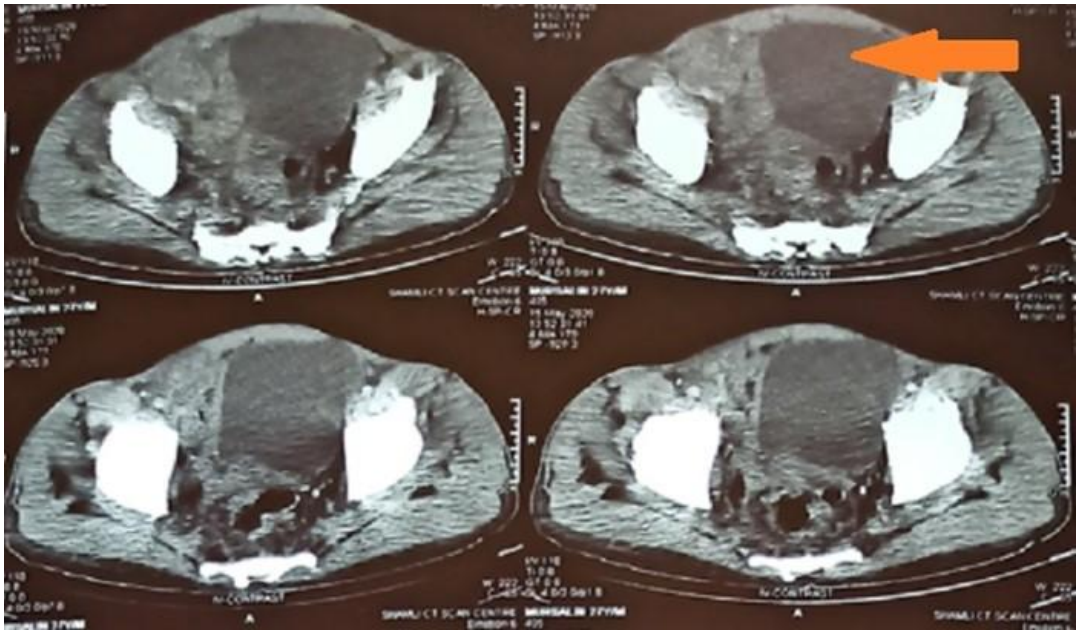

Figure 1: CECT scan of abdomen showing large mass measuring 11.0 × 8.0 cm in right iliac fossa (orange arrow) with luminal narrowing

To prevent recurrence, patient was planned to given adjuvant chemotherapy with 3-weekly VAC (vincristine, adriamycin, cyclophosphamide) regimen. But, unfortunately, after receiving 3-courses patient conditions deteriorated and patient succumbed to death.

Discussion

Desmoplastic small round cell tumor (DSRCT), although not new, still an infrequent occurring entity. It is very aggressive malignancy occurring mostly in young adult patients, 3rd decade of life being most common age-group.[1] Cases of DSRCT predominates in males, although cases in females also reported in literature.[2] It is thought to arise from serosal surface of peritoneum, characterized by abdominopelvic sarcomatosis exhibiting with multi-phenotypic potential. However, rare sites of occurrence like ovary, testis, central nervous system, pancreas even orbit are also documented.[1-4] A single case report of transverse colon DSRCT, managed only by surgery without any radiation or chemotherapy, and having good post-operative survival was also reported.[5] Histopathologically, DSRCT is a small blue round cell tumor with epithelioid and spindle cells surrounded by growth of connective tissue,

which is described as desmoplasia. First described by Gerald and Rosai in 1989 as a separate disease, it is molecularly characterized by a pathognomonic translocation between Ewing sarcoma RNA binding protein 1 gene (EWSR1) and Wilms tumor suppressor gene (WT1).[6]

Most DSRCT patients present with advanced disease and abdomen is the most commonly affected organ. High chances of spread to other organs make this disease more vulnerable. Common presenting symptoms are abdominal pain and/or distention, abdominal mass or lump, sometimes very huge, nausea, constipation, and/or weight loss. Nearly half of patients have distant metastases at the time of initial presentation; common sites of distant metastasis being liver, lungs, and lymph nodes.[7]

Multimodality approach involving surgery, chemotherapy, and radiotherapy is the mainstay of management. However, in most cases these aggressive strategies are not possible to employ due to poor general condition of patient and obviously advanced stage of disease.

Surgery is always the first preferred therapeutic option, as gross tumour resection is associated with prolonged survival (3-year survival of 58% compared to 0% in the non-resected patients).[8] Removal of at least 90% of the tumour burden as debulking procedure also confer favourable outcome.

Chemotherapy is part and parcel of management of DSRCT, irrespective of surgical intervention. This tumor is highly chemosensitive, chemotherapeutic agents help to control the systemic burden of advanced disease and prevent chances of micro-metastasis in early stage disease. Anthracycline based combination chemotherapy are used in majority of case series; others drugs used are vincristine, cyclophosphamide, dactinomycin, ifosfamide & etoposide in various combination. Others drugs used as 2nd line regimens are gemcitabine, docetaxel, topotecan, temozolomide and dacarbazine.[9] Role of hyperthermic intraperitoneal chemotherapy (HIPEC) with cisplatin is also evaluated with variable outcome.[10] Concept and choice of chemotherapy agents are employed from regimens used in sarcoma especially in Ewing sarcoma. Nevertheless, most DSRCT patients eventually die from the tumor burden, chemotherapy prolonged their survival.

DSRCT is also radiosensitive tumor; whole abdominal radiotherapy in a dose of 30 Gy with boost to residual disease is well established practice in early days.[11,12] Treatment was given by conventional 2-dimensional technique. However, severe toxicity of whole abdominal RT, both early and late, and high rates of local recurrence prompt the need of intensity modulated based whole abdominal RT, with IMRT having obviously less toxicity profile.[13] However, most of the recent day's practice employed RT in metastatic setting and for palliation of symptoms rather than as curative approach. It is obvious that pain control and palliation was encouraging with radiotherapy.

Currently role of targeted agents in DSRCT is usually in progressive staged disease, despite first-line or second-line chemotherapy; although better systemic therapies for front-line treatment are urgently needed. Different agents used as targeted therapies are pazopanib, imatinib & sorafenib (multi-kinase inhibitor); ridafirolimus & sirolimus (M-TOR inhibitors); leflunomide & olarutumab (anti- PDGFR agent); ganitumab (IGF-1 receptor antibody) and anti-androgens with variable response rate.[14] A single case showed good response with over 4-months survival in metastatic settings treated by apatinib, a multi-kinase VEGF inhibitor.[15] It is to be hoped that, some potential targets such as the MeRT/EMT switch and DNA repair are worthy of further investigation.[14] Immunotherapy are also tried in management of DSRCT. A phase I trial of intraperitoneal radioimmunotherapy with I-8H9 is ongoing in patients with DSRCT, particularly of intra-abdominal origin and having ascites.[16] Novel approach warrant further study include Yttrium [90Y] microsphere treatment for liver metastasis in DSRCT.[17]

In a study done by Lal et al, combined modality treatment with chemotherapy, surgery and radiotherapy conferred a 3-year survival of 55% compared to 27% for those who did not receive all three treatments.[18] DSRCT possesses aggressive behaviour and poor prognosis, with a median survival rate of 14 to 34 months.[19] The 3-and 5-year survival rates in patients with DSRCT have been documented to be 44% and 15%, respectively.[1] The longest survivor after the diagnosis was reported to be 101 months, which is surprisingly very high.[20]

Conclusion

DSRCT is a rare form of abdominal tumor having advanced course at presentation. Despite multimodality treatment approach, most cases experienced dismal outcome. Newer targeted agents depending on its molecular pathway and other new modes of treatment may bring a new way in its management. More prospective studies are needed to conclude standard treatment guideline.

Acknowledgement: Nil
Source of Support: Nil

Consent: All authors declared that written informed consent was obtained from the patient himself for publication of this case report and accompanying images. However, identification of the patient is not disclosed anywhere in the article.

References:

1. Saleh, Daniyah et al. "Desmoplastic Small Round Cell Tumor of Pancreatic Origin in a Young Child: A Case Report and Review of Literature." The American journal of case reports vol. 21 e922762. 13 Jul. 2020, doi:10.12659/AJCR.922762
2. Hatanaka, Kanako C et al. "Desmoplastic small round cell tumor of the parotid gland-report of a rare case and a review of the literature." Diagnostic pathology vol. 14,1 43. 18 May. 2019, doi:10.1186/s13000-019-0825-1
3. He, Xue-Rui et al. "Primary desmoplastic small round cell tumor in the left orbit: a case report and literature review." International ophthalmology vol. 39,2 (2019): 471-475. doi:10.1007/s10792-018-0829-y
4. Abu-Zaid, Ahmed, et al. "Desmoplastic small round cell tumor of stomach." Case Reports in Gastrointestinal Medicine 2013 (2013).
5. Huang, Jian Ping et al. "Desmoplastic small round cell tumor in transverse colon: report of a rare case." International surgery vol. 100,5 (2015): 809-13. doi:10.9738/INTSURG-D-14-00134.1
6. Gerald, W L, and J Rosai. "Case 2. Desmoplastic small cell tumor with divergent differentiation." Pediatric pathology vol. 9,2 (1989): 177-83. doi:10.3109/15513818909022347
7. Salako, O et al. "DESMOPLASTIC SMALL ROUND BLUE CELL TUMOUR OF THE ABDOMEN - A CASE REPORT." Journal of the West African College of Surgeons vol. 5,4 (2015): 79-89.
8. Wong, Han Hsi et al. "Desmoplastic small round cell tumour: characteristics and prognostic factors of 41 patients and review of the literature." Clinical sarcoma research vol. 3,1 14. 26 Nov. 2013, doi:10.1186/2045-3329-3-14
9. Subbiah, Vivek et al. "Multimodality Treatment of Desmoplastic Small Round Cell Tumor: Chemotherapy and Complete Cytoreductive Surgery Improve Patient Survival." Clinical cancer research : an official journal of the American Association for Cancer Research vol. 24,19 (2018): 4865-4873. doi:10.1158/1078-0432.CCR-18-0202
10. Hayes-Jordan, Andrea, et al. "Continuous hyperthermic peritoneal perfusion for desmoplastic small round cell tumor."

Journal of pediatric surgery 42.8 (2007): e29-e32.

11. Kushner, B H et al. "Desmoplastic small round-cell tumor: prolonged progression-free survival with aggressive multimodality therapy." Journal of clinical oncology : official journal of the American Society of Clinical Oncology vol. 14,5 (1996): 1526-31. doi:10.1200/JCO.1996.14.5.1526
12. Goodman, Karyn A et al. "Whole abdominopelvic radiotherapy for desmoplastic small round-cell tumor." International journal of radiation oncology, biology, physics vol. 54,1 (2002): 170-6. doi:10.1016/s0360-3016(02)02871-7
13. Pinnix, Chelsea C et al. "Whole abdominopelvic intensity-modulated radiation therapy for desmoplastic small round cell tumor after surgery." International journal of radiation oncology, biology, physics vol. 83,1 (2012): 317-26. doi:10.1016/j.ijrobp.2011.06.1985
14. Bexelius, Tomas S et al. "Mini-Review on Targeted Treatment of Desmoplastic Small Round Cell Tumor." Frontiers in oncology vol. 10 518. 21 Apr. 2020, doi:10.3389/fonc.2020.00518
15. Shi, Chunyu et al. "Effective treatment of apatinib in desmoplastic small round cell tumor: a case report and literature review." BMC cancer vol. 18,1 338. 27 Mar. 2018, doi:10.1186/s12885-018-4135-x
16. Hayes-Jordan, Andrea et al. "Management of desmoplastic small round cell tumor." Seminars in pediatric surgery vol. 25,5 (2016): 299-304. doi:10.1053/j.sempedsurg.2016.09.005
17. Subbiah, Vivek, Ravi Murthy, and Pete M. Anderson. "[90Y] yttrium microspheres radioembolotherapy in desmoplastic small round cell tumor hepatic metastases." Journal of Clinical Oncology 29.11 (2011): e292.
18. Lal, Dave R et al. "Results of multimodal treatment for desmoplastic small round cell tumors." Journal of pediatric surgery vol. 40,1 (2005): 251-5. doi:10.1016/j.jpedsurg.2004.09.046
19. Biswal, Biswa Mohan et al. "Intra-abdominal desmoplastic small round cell tumor: Presentation of four cases and review of the literature." Indian journal of medical and paediatric oncology : official journal of Indian Society of Medical & Paediatric Oncology vol. 31,1 (2010): 24-7. doi:10.4103/0971-5851.68849
20. Gil, Alberto et al. "Clinical perspective on desmoplastic small round-cell tumor." Oncology vol. 67,3-4 (2004): 231-42. doi:10.1159/000081323.