

Intra-abdominal desmoplastic small round cell tumour

Ansari NP¹, Mamun MA², Islam MS³, Begum A⁴

Abstract

Intra-abdominal desmoplastic small round cell tumour (DSRCT) is a rare and highly aggressive malignant neoplasm with a strong predilection in young male adults. and frequently presents as a large abdominal mass. Here, we present a case of DSRCT in a 40 years male with mild ascitis.

➤ CBMJ 2013 July: Vol. 02 No. 02 P: 75-77

Key words: Desmoplastic small round cell tumour(DSRCT), young male adult, Intra-abdominal

Introduction

Intra-abdominal desmoplastic small round cell tumour (DRSCT) is of uncertain heterogenesis with a predilection for the peritoneal surfaces of the abdomen and pelvis and, rarely, the pleura^{1, 2, 3,4,5,6,7P}. Solitary exmples of arising from the scalp,brain,parotid gland,ethmoid sinus,hand and liver have been reported DRSCT. Predominantly occurs in male(male to female ratio of 4:1 and adolescents and young adults (mean age 22 years in the largest series)^{4P}.It can also occur in the elderly^{8PB}.^{PB} It usually presents as a single mass or multiple nodules within the abdominal cavity. Accompanying ascites is the rule; malignant cells can be easily identified in the fluid^{9P}. Patients of intra-abdominal DRSCT typically present with abdominal distension, pain and a palpable abdominal, pelvic or scrotal mass. We report a case of intra-abdominal desmoplastic small round cell tumour and discuss the clinical presentation, diagnostic histologic and immunophenotypic features and management of such a case.

Then leparatomy followed by excision biopsy was done.



Gross examination revealed an excised part of colon with an attached mass at the outer surfaces measuring 8x6x4 cm. Cut surface were solid gray-white in appearance.

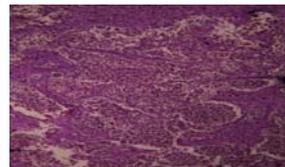


Figure 2: A. A low-magnification photograph of a DSRCT shows characteristic bands of collagen material separating nests of small round blue cells.

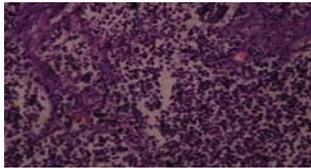
Case Report

A 40 years old male presented with complaints of pain in the left upper quadrant of abdomen with the history of weight loss and constipation for few months. He did not have any family history of cancer. On examination there was a non tender mobile palpable mass in the left upper quadrant of the abdomen. Ultrasound examination of the abdomen revealed a colonic mass with mild ascitis. Barium enema revealed extrinsic compression of the colon close to the splenic flexure.

1. *Dr. Nazma Parvin Ansari
Assistant Professor of Pathology
Community Based Medical College Bangladesh.
2. Dr. Mohammad Abdullah Al Mamun
OSD, DGHS, Dhaka.
3. Dr. Md. Saiful Islam
Assistant Professor of Orthopedics
Community Based Medical College Bangladesh.
4. Dr. Ambia Begum,
Assistant Professor of Pathology
Community Based Medical College Bangladesh.

*** Address of correspondence:**

E-mail : path_napalash@yahoo.com
Mobile : 0088-01727428441



B. A higher power view of the same tumor shows hyperchromatic nuclei, condensed chromatin, and mitotic figures.

Histopathology of the specimen showed a neoplastic lesion composed of clusters of small round to oval monotonous cells with very scanty cytoplasm. Nuclei were small and pleomorphic with inconspicuous nucleoli. Marked desmoplastic response was identified around the tumor cell clusters.

Discussion:

Desmoplastic small round cell tumor (DSRCT) was first described in 1989 by Gerald and Rosai who described a distinct type of small round blue cell tumor with a predilection for serosal surfaces such as the peritoneum and the tunica vaginalis that affected mostly Caucasian males in the second or third decade of life^{10P}.

DSRCT presents in most of the cases as an abdominal mass. Patients may complain abdominal pain, weight loss and constipation. The most common presentation is bulky abdominal disease present in a young adult, often males. DSRCT is regional and the major bulk of these tumors is intraabdominal. Liver metastases are common. Other distant sites include lymph nodes, lung and bones. It may present as a painful lump in the umbilicus known as a "Sister Mary Joseph nodule," which is the secondary to metastatic cancer^{11,12P}.

Microscopically sharply outlined tumour cells separated by abundant stroma (desmoplastic). The tumour cells are usually small, round and monotonous with hyperchromatic nuclei, high mitotic activity and very scanty cytoplasm. Stroma is made up of fibroblast and myofibroblasts result from the secretion of fibroblastic growth factor by the tumour cells. Stroma also contain

proliferating vessels resulting from secretion of angiogenic factor by the tumour cells. Morphological variation of DSRCT include tumour with very scanty stroma, presence of tubular and glandular formations, signet ring cells, rhabdoid cells, and clusters of pleomorphic large tumour cells with bizarre nuclei.^{5,13,14,15P}

The differential diagnosis includes Ewing sarcoma family tumors, rhabdomyosarcoma, neuroblastoma, lymphoma, synovial sarcoma, ectomesenchymoma and blastemic Wilms' tumor in children; and small cell carcinoma, carcinoid tumor, neuroendocrine carcinomas, Merkel cell carcinoma and small cell mesothelioma in adults.

The immunohistochemical profile of this tumour is distinctive. It displays simultaneous expression of epithelial (keratin, EMA) muscular (desmin), and neural (neuron specific enolase) markers.

DSRCT is associated with the unique chromosomal translocation (11;22) (p13;q12)P^{16P} which results in fusion of the N-terminal activation domain of EWS (Ewing sarcoma) gene on 22q12 with the C-terminal DNA-binding domain of WT1 (Wilms tumour gene1) on 11p13P^{17,18,19P}. This finding is of practical importance in the differential diagnosis with other small round cell tumour of childhood^{20,21P}.

A multi-disciplinary approach of high dose chemotherapy, aggressive surgical resection, radiation, and stem cell rescue improves survival for some patients. Reports have indicated that patients will initially respond to first line chemotherapy and treatment but that relapse is common.

Unfortunately, these modalities frequently do not provide a durable response, and the prognosis for patients with DSRCT remains poor. Despite aggressive therapy, 3-year overall survival has been estimated at 44% and the 5-year survival rate remains around 15%.

Conclusion:

The behaviour of DSRCT is extremely aggressive, perhaps more so than that of any other malignant small round cell tumour. The median survival is less than 3 years. However prolonged progression free survival has been achieved in some cases with aggressive multimodality therapy.

References:

1. Gerald WL, Miller HK, Batifora H et al. Intraabdominal desmoplastic small round cell tumour. *Am J Surg Pathol* 1991;15:499-513.
2. Cummings OW, Ulbright TM, Young RH, et al. Desmoplastic small round cell tumour of the para-Testicular region: a report of six cases. *Am J Surg Pathol* 1997;21:219-225.
3. Young RH, Eichhorn JH, Dickersin GR, et al. Ovarian involvement by the intra-abdominal desmoplastic small round cell tumour with divergent differentiation: a report of three cases. *Hum Pathol* 1992;23:454-464.
4. Gerald W, Ladanyi M, de Alava E, et al. Clinical pathologic, and molecular spectrum of tumour associated with t(11;22)(p13;q12): desmoplastic small round cell tumour and its variants. *J Clin Oncol* 1998;16:3028-3036.
5. Ordonez NG. Desmoplastic small round cell tumour. I. A histopathologic study of 39 cases with emphasis on unusual histologic patterns. *Am J Surg Pathol* 1998;22:1303-1313.
6. Ordonez NG. Desmoplastic small round cell tumour. II. An ultrastructural and immunohistochemical study with emphasis on new immunohistochemical markers. *Am J Surg Pathol* 1992;22:1314-1327.
7. Lae ME, Roche PC, Jin L, et al. Desmoplastic small round cell tumour. A clinicopathologic, immunohistochemical and molecular study of 32 tumours. *Am J Surg Pathol* 2002;26:823-835.
8. Wolf AN, Ladanyi M, Paull G, Blaugrund JE, Westra WH. The expanding clinical spectrum of desmoplastic small round cell tumour; a report of two cases with molecular confirmation. *Hum Pathol* 1999;30:430-435.
9. Crapanzano JP, Cardillo M, Lin O, Zakowski MF. Cytology of desmoplastic small round cell tumour. *Cancer* 2002;96:21-32.
10. Lae ME, Roche PC, Jin L, Lloyd RV, Nascimento AG. Desmoplastic small round cell tumor: a clinicopathologic, immunohistochemical, and molecular study of 32 tumors. *Am J Surg Pathol*. 2002 Jul;26(7):823-35.
11. Doros L, Kaste SC, Rodriguez-Galindo C. Sister Mary Joseph's nodule as presenting sign of a desmoplastic small round cell tumor. *Pediatr Blood Cancer*. 2008 eb;50(2):388-90.
12. Albano EA, Kanter J. Images in clinical medicine. Sister Mary Joseph's nodule. *N Engl J Med*. 2005 May 5;352(18):1913.
13. Dorsy BV, Benjamin LE, Fauscher F, Klenske B, Venook AP, Warren RS, Weidner N. Intraabdominal desmoplastic small round-cell tumour: expansion of the pathologic profile. *Mod Pathol* 1996;9:703-709.
14. Pasquinelli G, Montanaro I, Martinelli GN. Desmoplastic small round-cell tumour: a case report on the large cell variant with immunohistochemical, ultrastructural and molecular genetic analysis. *Ultrastruct Pathol* 2000;24:333-337.
15. Ordonez NG, Sahin AA. CA 125 production in desmoplastic small round-cell tumour: report of a case with elevated serum levels and prominent signet ring morphology. *Hum Pathol* 1998;29:294-299.
16. Rodriguez E, Sreekantaiah C, Gerald W, Reuter VE, Motzer RJ, Chaganti RS. A recurring translocation, t(11;22)(p13;q11.2), characterizes intra-abdominal desmoplastic small round-cell tumours. *Cancer Genet Cytogenet* 1993;69:17-21.
17. Gerald WL, Rosai J, Ladanyi M. Characterization of the genomic breakpoint and chimeric transcripts in the EWS-WT1 gene fusion of desmoplastic small round-cell tumour. *Proc Natl Acad Sci USA* 1995;92:1028-1032.
18. Lae ME, Roche PC, Jin L, Lloyd RV, Nascimento AG. Desmoplastic small round cell tumour: A clinicopathologic, immunohistochemical and molecular study of 32 tumours. *Am J Surg Pathol* 2002;26:823-835.
19. Sandberg AA, Bridge JA. Updates on the cytogenetics and molecular genetics of bone soft tissue tumours. Desmoplastic small round-cell tumours. *Cancer Genet Cytogenet* 2002;138:1-10.
20. Argatoff LH, O'Connell JX, Mathers JA, Gilks CB, Sorensen PH. Detection for the EWS/WT1 gene fusion by reverse transcriptase polymerase chain reaction in the diagnosis of intra-abdominal desmoplastic small round-cell tumour. *Am J Surg Pathol* 1996;20:406-412.
21. de Alava E, Ladanyi M, Rosai J, Gerald WL. Detection of chimeric transcripts in desmoplastic small round-cell tumour and related developmental tumours by RT-PCR. A specific diagnostic assay. *Am J Pathol* 1995;147:1584-1591.