Case Report

A report of a case of desmoplastic small round cell tumor of peritoneum

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ABSTRACT

Desmoplastic small round cell tumor (DSRCT) is a rare subtype of "small round blue cell tumors" typically arising from the peritoneum. An 18-year-old male patient presented with palpable nontender abdominal mass associated with dragging sensation. Imaging revealed a large heterogenous soft tissue mass in the pelvis and lower abdomen along with irregular peritoneal thickening. Histopathology and immunohistochemistry suggested DSRCT. Neoadjuvant chemotherapy followed by laparotomy with debulking surgery of the pelvic mass was done after 5 months of starting chemotherapy. Follow-up contrast enhanced computed tomography abdomen showed a reduction of the size of residual disease. However, follow-up magnetic resonance imaging done after 3 months of surgery showed disease recurrence. The tumor should be suspected in young males when imaging is showing a pelvic mass along with multiple peritoneal soft tissue deposits.

Key words: Desmoplastic small round cell tumor, imaging, peritoneum, round blue cell tumors

INTRODUCTION

Desmoplastic small round cell tumor (DSRCT) belongs to the family of "small round blue cell tumors" and is a rare neoplasm of peritoneum with <200 cases reported worldwide.[1,2] The first case was reported in the literature in the year 1989 by Gerald and Rosai.[3] DSRCT was proposed to be a distinct entity in 1991 by Gerald *et al*. with the characterization of specific pathological features of the tumor.[4] It mostly occurs in children and adolescents with male preponderance.[1] The cell of origin is unknown, however, it is postulated to originate from the serosa owing to its common occurrence on mesothelial surfaces. The typical location of DSRCT is peritoneum and presents as multifocal lesions.[4] Since the patient often presents with abdominal pain or palpable lump, a computed tomography (CT) scan of the abdomen is most often used for initial diagnosis.[5] Ultrasound, CT, or magnetic resonance imaging (MRI) reveals multiple tumor nodules "studding" the peritoneal cavity and causing indentation/scalloping of the liver surface. Histologically, DSRCT is characterized by the desmoplastic stroma that envelops the hyperchromatic tumor cells.[6] We present a case of DSRCT of the peritoneum in an 18-year-old male presenting with suprapubic lump and urinary complaints. Imaging evaluation by ultrasonography, CT, and MRI was done. The patient was subsequently given chemotherapy with "P6 regimen" which resulted in partial resolution of the lesions. This was followed by debulking surgery, and adjuvant chemotherapy.

CASE REPORT

An 18-year-old male patient presented with suprapubic mass and increased the frequency of micturition. It was associated with dragging sensation and was not associated

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with pain. On physical examination, a nontender mass was palpable in the suprapubic region with ill-defined margins. On sonographic evaluation, a large heterogenous soft tissue mass was seen in the pelvis and lower abdomen. MRI revealed large soft tissue mass in the pelvis in the rectovesical pouch and was appearing hypointense on T1 weighted images; intermediate in signal intensity on T2 weighted images with central necrotic T2 hyperintense areas, and showing heterogenous contrast enhancement [Figure 1]. Diffusion restriction was seen on diffusion-weighted images. It was indenting and displacing the urinary bladder anteriorly and causing mild right sided hydroureteronephrosis due to compression of the ureter. The rectosigmoid colon was compressed by the mass. Irregular peritoneal thickening and nodules were noted along the hepatic surface, omentum, hepatorenal, and lienorenal recess. Ultrasound-guided fine needle aspiration cytology was indicative of DSRCT [Figure 2]. Immunohistochemistry showed reactivity for epithelial (cytokeratin antigen) and muscular marker (desmin antigen) [Figure 3a and b]. 5 cycles of neoadjuvant chemotherapy were given with P6 protocol comprising seven cycles of chemotherapy (Courses 1, 2, 3, and 6 of HD-CAV - high-dose cyclophosphamide on days 1 and 2, doxorubicin and vincristine on days 1, 2, and 3. Courses 4, 5, and 7 of ifosfamide and etoposide for 5 days), which resulted in partial remission of the disease. No major complications were seen during the course of chemotherapy. This was followed by laparotomy with debulking surgery of the mass in the rectovesical pouch. Adjuvant chemotherpy was given with ifosfamide, dacarbazine, and doxorubicin. Follow-up contrast enhanced CT of the abdomen after debulking surgery and adjuvant chemotherapy showed a reduction in the size of the pelvic mass lesion [Figure 4]. There was significant regression in the size of subcapsular hepatic, omental, and peritoneal lesions. The patients were followed up with MRI after 3 months, which depicted disease recurrence at the rectovesical pouch [Figure 5] and increased size and number of the peritoneal nodules. Locoregional dissemination was seen as multiple nodular lesions in the paracolic gutters and mesentery in addition to lesions occurring in the hepatic surface, omental, Morrison's pouch, and lienorenal recesses. The lesions were hyperintense on T2 and isointense to hypointense on T1 weighted images with evidence of diffusion restriction and contrast enhancement. Thus, the tumor response to therapy was poor. The patient did not come for follow-up,

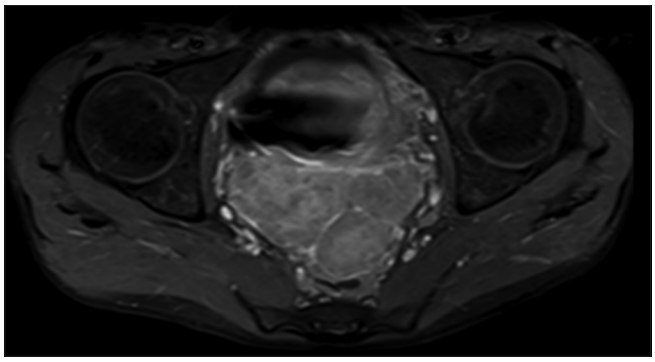


Figure 1: Contrast-enhanced magnetic resonance imaging shows heterogenous enhancement of the mass in the rectovesical pouch indenting the urinary bladder anteriorly and compressing the rectum posteriorly

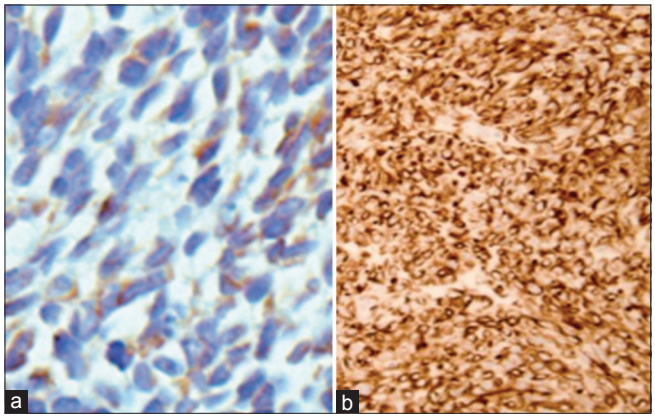


Figure 3: Positive immunostaining of desmoplastic small round cell tumor for keratin (a) and Desmin (b)

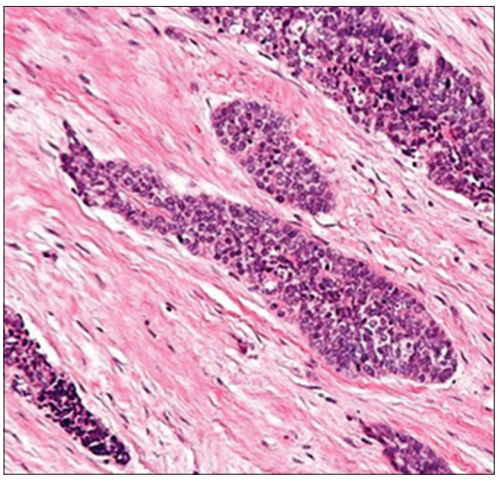


Figure 2: Histopathological analysis of the tumor shows well-defined nests of small round blue tumor cells separated by abundant desmoplastic stroma

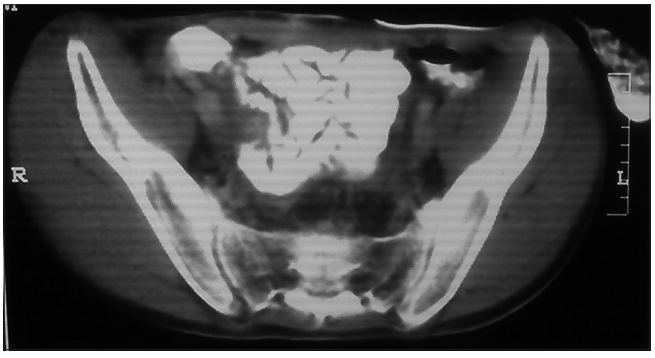
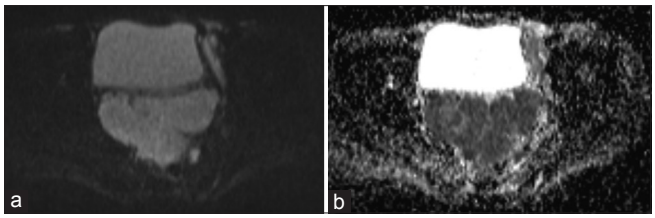


Figure 4: Contrast-enhanced computed tomography postdebulking surgery shows significant resolution of the mass within the rectovesical pouch with the oral contrast opacified ileal loops seen at the previous site of the lesion



Figures 5: Follow-up magnetic resonance imaging done 3 months after the surgery shows recurrence of the mass showing diffusion restriction on diffusion weighted images (a) and apparent diffusion coefficient maps (b)

however, is surviving for 18 months after the disease diagnosis.

DISCUSSION

DSRCT belongs to the family of small round cell tumors such as Ewing's sarcoma (EWS), neuroblastoma, malignant lymphoma, primitive neuroectodermal tumor, rhabdomyosarcoma, and small cell mesothelioma.[1] It mostly occurs in second and third decades of life typically in 18–25 years age group with a male to female ratio of 3:1.[4] The cell of origin is unknown, however, it is postulated to originate from the serosa owing to its common occurrence on mesothelial surfaces.[7]

The most common location of DSRCT is peritoneum, however, other rare sites of occurrence such as central nervous system, base of skull, sinus cavity, pleural region, lung, paratesticular region, ovary, and stomach have been described in literature.[8,9] Uncommonly, metastatic pulmonary nodules, thoracic lymph nodes, pericardial effusion, and bone secondaries have also been described. Usually, the presentation is of a diffuse and multifocal intraabdominal disease akin to carcinomatosis. The multiplicity of lesions is typical, presenting as large masses, and/or extensive seeding in the visceral and parietal peritoneum with the locoregional dissemination of the disease.[9]

The typical clinical features include abdominal distension, discomfort, pain, and weight loss accompanied by gastrointestinal or genitourinary complaints such as constipation, intestinal obstruction, umbilical hernia, and dysuria.[1] Some patients may present with acute abdominal pain incapacitating the ability to walk upright. Physical examination usually depicts an ill-defined palpable hard abdominal mass, which may be tender. In this case, the patient presented with ill-defined palpable nontender suprapubic lump associated with increased frequency of micturition and dragging sensation.

Imaging evaluation with ultrasound, CT, and MRI often reveals diffuse and nodular, usually multiple masses in the pelvis or abdominal cavity located in the greater omentum, mesentery, and abdominopelvic peritoneal surfaces. On ultrasound, these masses show heterogenous echo pattern with hypoechoic or anechoic areas indicative of necrosis or cystic degeneration with increased vascularity seen on color doppler.[10]

CT scan is the modality of choice depicting multiple nodular heterogenously hypodense masses "studding" the peritoneal cavity often causing indentation or "scalloping" of the liver surface.[11] These may be associated with focal calcification, hemorrhage or necrosis.[1,12] Secondaries to the liver and abdominal lymph nodes are usually seen. Accompanying evidence of ascites, urinary tract, and/or bowel obstruction may be present. A dominant peritoneal soft tissue lesion measuring at least 5 cm without any visceral organ of origin is highly suspicious for DSRCT. A large mass >10 cm may be visualized in rectovesical or rectouterine pouch.[13,14] The location of mass in the retrovesical or rectouterine pouch has been attributed to the dynamics of the natural flow of peritoneal fluid and the dependent site of the pouch of Douglas.[1] Association has been noted between the presence of retrovesical mass, ascites, abdominal lymphadenopathy, and hepatic metastasis.[15] Not surprisingly, masses may be seen in paracolic and paravertebral spaces. In this case, there was a mass lesion in the rectovesical pouch with associated multiple nodules causing scalloping of the hepatic surface.

MRI is useful in the evaluation of the extent of disease. However, the findings are nonspecific. Lesions appear isointense or hypointense to muscle on T1 and heterogeneously hyperintense on T2 weighted images.[16] Contrast enhanced images show heterogeneous enhancement. Occasional hypointense signal on T2 and minimal enhancement have been reported, indicating densely packed cells, and desmoplastic component. Intratumoral high signal on T1 may be present due to hemorrhage. There have been very few reports elaborating the MRI findings with none reporting the diffusion weighted imaging. In our case, there was intratumoral diffusion restriction on diffusion-weighted imaging/apparent diffusion coefficient images.

Histopathology shows clusters of blue primitive undifferentiated tumor cells arranged in a round or elongated fashion within the abundance of desmoplastic/fibromyxoid stroma.[9] The tumor cells are small to medium in size and show round/oval hyperchromatic nucleus and scanty cytoplasm. Immunohistochemistry shows reactivity for epithelial (keratin, epithelial membrane antigen), mesenchymal (vimentin), neural (neuron specific enolase and CD56), and myogenic (desmin) markers.[17] There was a similar appearance in our case. DSRCT cells also depict chromosomal translocation, t(11;22)(p13;q12) which fuses

the N-terminus of the EWS gene to the C-terminus of the Wilms tumor gene.[18,19]

Management of DSRCT is aggressive multimodality approach comprising neoadjuvant chemotherapy, debulking of the tumor (90%), and radiotherapy.[9] Combination of three modalities shows an overall response rate of 39% and 3 years survival rate of 50%. Despite reports of prolongation of survival, complete cure of disease is rarely possible. Alkylating agents such as cyclophosphamide and ifosfamide are important components of chemotherapy. The neoadjuvant chemotherapy cornerstone regimen comprises cyclophosphamide, doxorubicin, and vincristine alternating with ifosfamide and etoposide. Multidrug chemotherapy leads to approximately 40% tumor response.[20] Kushner *et al.* reported disease remission with P6 protocol comprising seven cycles of chemotherapy (Courses 1, 2, 3, and 6 of HD-CAV - high-dose cyclophosphamide 2 on days 1 and 2, doxorubicin and vincristine on days 1, 2, and 3. Courses 4, 5, and 7 of ifosfamide and etoposide for 5 days).[21] Role of anthracycline-based chemotherapy is suggested in some metastatic cases. However, disease recurrence is very common, often within 6 months.[22] In our case, P6 regimen was used which resulted in partial disease remission followed by recurrence.

Role of radiotherapy in DSRCT is controversial, especially that of whole abdominal-pelvic radiotherapy owing to the toxicity and lower degree of response.[23]

At Present, there is no real consensus regarding the management strategy. The disease has a fulminant course with almost two-thirds of patients succumbing to the disease within 3 years of diagnosis. The median survival is approximately 17 months with the overall 5 years survival rate reported to be 15%.[24]

CONCLUSION

DSRCT is an aggressive tumor occurring in young adult males. Imaging depicts multiple heterogeneously enhancing bulky masses studding the peritoneal surfaces, mesentery, and omentum. Pathologically, they resemble other "small round blue cell tumors" comprising sheets of round cells with hyperchromatic nuclei and scanty cytoplasm. This disease often goes misdiagnosed and is usually difficult to manage. It is imperative to suspect the disease in young male when imaging suggests multiple heterogeneous peritoneal soft tissue masses since early detection and management helps in restricting the tumor and thereby improving the survival. At Present, there are no screening or preventive measures for this disease. Due to the rare occurrence of the tumor, clinical trials are not always possible which are of utmost importance for developing management protocols.

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Conflicts of interest
There are no conflicts of interest.

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