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Case Report

DESMOPLASTIC SMALL ROUND CELL TUMOR (DSRCT)-A CASE REPORT AND REVIEW OF LITERATURE

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Abstract:

Desmoplastic small round cell tumor (DSRCT) is a rare pathologic entity that most frequently affects the peritoneal cavity and presents in pediatric and adolescent boys. It often presents at an advanced stage and has a generally poor prognosis. Sometimes it may involve liver at diagnosis. We present an unusual case of DSRCT who was present with mass in lower abdomen, pain, hepatomegaly and mild ascites. This tumor is characterized by nests of small undifferentiated cells that show immunohistochemical evidence of epithelial, mesenchymal and neural differentiation. In our patient histologicaly tumor had the characteristic features of DSRCT and were composed of small round cells with hyperchromatic nuclei and scanty cytoplasm. With various difficulties in diagnosis we ultimately reached at diagnosis by open biopsy and immuno-histochemistry. Now patient is on multidrug chemotherapy (modified p6 protocol). Diagnosis and management of a rare tumor needs high level of suspicion and in time intervention.

Key word: Childhood, Desmoplastic small round cell tumor, multidrug chemotherapy

Introduction:

Desmoplastic small round cell tumor (DSRCT) is highly aggressive malignant neoplasm that occurs in adolescents and young adult with a male preference(11:1).[1,2] 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 .[1,3] This tumor frequently occurs as multiple masses in abdomen involving the regional lymph nodes and the lining of the abdomen and pelvis ,[2] other primary sites have been reported, including the paratesticular region, [4-7] the pleural serosa, the posterior cranial fossa, soft tissues and bone, the ovary and the parotid gland.[1,8-9] DSCRT is also called "mesothelioblastoma.[1] It can co-express epithelial, neuronal, and mesenchymal markers. Clinical manifestations are often related to widespread abdominal disease. The most common sites of spread are the liver, lungs and bones. Distant metastases can be present at the time of diagnosis. Desmoplastic small round cell tumor (DSRCT) is an highly aggressive malignant neoplasm that WT1 resulting in an aberrant transcription factor involved in the pathogenesis of DSRCT.[1] Despite intensive therapy, a trial involving chemotherapy followed by allogeneic peripheral blood stem cell transplantation is currently in progress at the National Cancer Institute (USA). [4] Five year survival continues to be less than 15%. The average survival is <2 to 3 years. [2] New therapeutic approaches include molecularly targeted therapies and immunotherapy; the role of these modalities is yet to be defined.[1]

Case report:

A 8 yrs old boy of a non-consanguineous parents was admitted in Dhaka Medical College Hospital on 6/1/12 with masses in the abdomen and severe abdominal pain for 10 days. He had no h/o fever, night sweat, weight loss, blood transfusion, haematuria, hypertention, diarrhea, constipation or contact with

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TB patient. On examination, his vital signs were within normal limit. On abdominal examination, abdomen was distended, umbilicus centrally placed and everted, flanks were full. A mass was present in the hypogastrium region measuring 7x7cm², hard in consistency, irregular surface, tender & fixed with the underlying structures and free from overlying skin. Liver was palpable 5 cm from right costal margin along the mid clavicular line. It was firm, tender, smooth surface, sharp margin and upper border of the liver dullness was not obliterated. Ascites was present. Patient was not anaemic, icteric, lymph node not palpable & skin survey revealed normal.

Laboratory investigations showed hemogram was normal. ESR was 50 mm in 1st hour. Peripheral blood film was normal except thrombocytosis. Urine R/M/E was normal .USG of abdomen on 2nd February 2012 showed Space Occupying Lesion (SOL) in the liver, intra abdominal lymphadenopathy with ascites. CT guided Fine Needle aspiration Cytology (FNAC) from liver was in favor of heapatoblastoma. Alfha feto protein (0.54ng/ml), uric acid (4.60 mg/dl) were normal and LDH (1175 U/L) was raised. Another USG was done on 8th February2012 revealed a large hepatoblastoma with central necrosis and multiple focal lesions in both lobes, large solid irregular abdominal mass with ascites. FNAC from suprapubic mass was suggestive of histocytosis. and differentil diagnosis were Neuroblastoma and other small round cell tumor. Urinary VMA was 3.35 mg/24hrs (normal). CT scan showed feature suggestive of lymphoma with extensive hepatic metastasis with ascites. Subsequently, we consulted with pediatric surgery department and planned for total excision and biopsy. Then he underwent open abdominal surgery and found a large mass in relation to the gut & surrounding structure. Omentum was also involved.

Complete resection was not possible due to its widespread tissue involvement. The resected specimen had lobulated masses measuring 6 ×4 × 2 cm. The cut surface was gray white.. Solid and semi-cystic components were present. Histopathology report showed a malignant tumor made of sharply outlined nests of small tumor cells surrounded by a desmoplastic stroma. The tumor cells have hyperchromatic nuclei, scanty cytoplasm and indistinct cell border. Necrosis was evident at center of the cell nests and mitoses are frequent. Suggestive of desmoplastic small round cell tumor(Fig- 2)

Immunohistochemical was positive for AE1/ AE3(pancytokeratin), Vimentin, Desmin and EMA and negative for S100 protien, CD99, Chrogranin-A and GFAP. Both the clinical features and laboratory parameters consistent with desmoplastic small round cell tumor. We treated the patient by surgery that is incomplete resection of mass (as it is adhere with the bowel) followed by multiagent chemotherapy (p-6 protocol). Now patient's condition is improved and prepare for next cycle chemotherapy

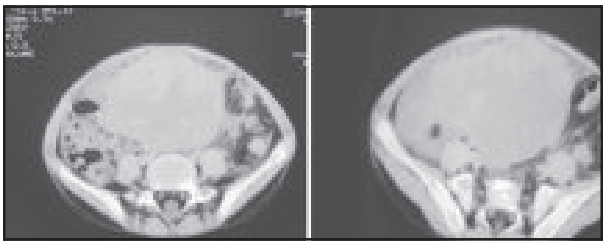

Fig.-1: *CT scans showing the tumor mass*

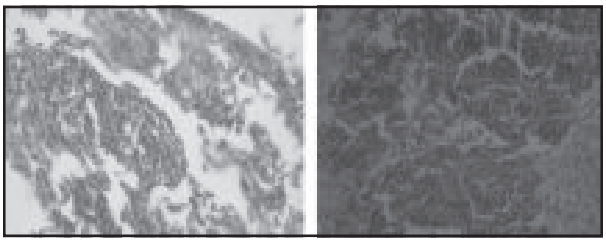

Fig.-2: *Histological features of DSRCT - shows nests of small undifferentiated round cells with surrounding desmoplastic stroma*

Discussion:

DSRCT belongs to the family of "small round blue cell tumors" commonly found in the pediatric population. These include Neuroblastoma, Non-Hodgkin's lymphoma, Rhabdomyosarcoma, Ewing's sarcoma, and Primitive neuroectodermal tumor (PNET).[2] Desmoplastic small round cell tumor is an aggressive cancer.[2,10,11,12] It is less common and less reported in pediatric age group. Desmoplastic small round cell tumor is classified as a soft tissue sarcoma. It is an aggressive tumor that primarily occurs as masses in the abdomen [4,10,] The tumor is considered a childhood cancer that predominantly strikes boys and young adults.[3,10] The disease rarely occurs in females, but when it does the tumors can be mistaken for ovarian cancer.[3] The most common presentation of DSRCT is abdominal pain and distention.[12-14] Our patient presented with abdominal pain, distention, a large palpable pelvic mass, ascites and hepatomegaly.

Murray JC also reported a case with such type of presentation in a 11 year old boy.[15] In our case patient was only 8 years old boy which was rarely reported before , male child presented with abdominal mass which is consistence with several other study.[1-2,4,15-16] A J Heikkila and APH Prebtani has shown a patient of older age presented with ascites which is not a common feature of DSRCT.[17] . In our patient there was ascites which was detected by physical examination and by abdominal USG. In Chinese study of 18 patients 4 patients presented with ascites, 17/18 and 10/18 patients presented with mass and abdominal pain respectively.[18]

In about one third of cases, morphological findings not considered to be typical of DSRCT. The characteristic histological feature may not be fully represented in material obtained by Fine Needle Aspiration Cytology (FNAC), due to the absence of desmoplastic stroma and non specific cytologic features. In our case initial histological finding from FNAC were not diagnostic of DSRCT. CT guided FNAC of liver showed hepatoblastoma. FNAC of suprapubic mass was suggestive of histocytosis. So FNAC can make trap for diagnostic dilemma. Open biopsy from intra-abdominal mass revealed desmoplastic small round cell tumor. DSCRT might have been classified as an atypical variety of small round cell tumors or as an unusual form of malignant mesothelioma, adenocarcinoma, carcinoid tumor, or germ cell tumor. The most striking histologic feature of DSCRT is the desmoplastic stroma that envelops the tumor cells. This stroma is generally densely collagenous or fibromyxoid and encases well-defined nests of primitive undifferentiated cells [2]

DSRCTs are treated first with chemotherapy, then with surgery to remove the tumor, if possible. Radiation therapy is sometimes given, depending on the tumor. In addition, some people with DSRCT are candidates for a bone marrow transplant. [2,19-20]. The application of large doses in chemotherapy (P6 proposal) has been approved to be effective against DSRCT at the NewYork Sloan-Kettering Memorial Cancer Center.[21] Ninety percent of the patients responded to the chemotherapy treatment that included cyclophosphamide, adriamycin, vincristine, ifosfamide, and etoposide. Compared with other chemotherapies, the P6 proposal provides a much better curative effect.[21] But the survival rate of DSRCT in a 3-year period is only 29%, though aggressive treatments were applied.[21] Mingo et al suggested that treatment should begin with chemotherapy, followed by surgery to resects all or most of the tumor.[22] Kushner BH et al treated 10 newly diagnosed DSRCT patients with p-6 protocol, only 2 patients were on CR but latter relapse occur.[21] Our patient treated with surgery followed by chemotherapy(p6 protocol). Despite aggressive treatment, including surgical debulking, radiation therapy, and multi agent chemotherapy, the prognosis in DSRCT is poor. The mean survival is 20 months after diagnosis.[2] Finally a multidisciplinary approach including early referral to a specialized centre is recommended in such complex cases.

Conclusion

Bulky peritoneal soft-tissue masses without an apparent organ-based primary site are characteristic of intra-abdominal desmoplastic small round cell tumor. Although it is occur in adolescent and young adult, it can be occur in lower age group also. So any child with abdominal distention and abdominal mass DSRCT should be kept in mind. Unfortunately, most patients come to clinical attention when the disease is an advanced stage and has disseminated, especially to the liver and to the retroperitoneal lymph nodes. High level of suspicion and appropriate investigations may confirm early diagnosis.

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