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CASE ARTICLE

Intra Abdominal Desmoplastic Small Round Cell Tumour - A Case Report

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ABSTRACT

This case report is prepared to discuss the very rare highly malignant neoplasm that characteristically present as a single mass within the abdominal cavity. FNAC revealed intra abdominal small round blue cell tumour. Histological feature is the characteristic display nests of malignant small round cells within a florid desmoplastic stroma.

An unusual feature of this tumour is its polyphenotypic differentiation with simultaneous trilinear coexpression of epithelial (CK, EMA), neural (NSE, S100 protein) & myogenic (Desmin, Vimentin) markers.

Keywords*: DSRCT, Polyphenotypic, FNAC*

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INTRODUCTION

DSRCT (Desmoplastic Small Round Cell Tumour) is a rare, high-grade malignant tumour, chiefly affects adolescent and young adult specially males, between 15 to 35 yrs of age. It occurs predominantly in the abdomen i.e. any portion of peritoneal cavity, omentum, retro peritoneum and frequently confined to pelvis. [1]So, it presents as a large mass within the abdomen and may be accompanied with extensive tumour implants throughout the peritoneum. Although most tumours arise in abdomen/ pelvis, these lesions can occur in extra-abdominal locations like CNS (Central Nervous System), pleura and paratesticular region. It pursues an aggressive clinical course with an extremely poor prognosis. The lesion is characterized by a proliferation of small round cell deposited in an abundant desmoplastic stroma and multiphenotypic differentiation by immunohistochemistry.[2,3]

CASE REPORT

A 22 year old male patient came to the surgery OPD with a large mass in his abdomen for a period of 3 months. Physical examination of the patient showed 12 x 9 x 5 cm hard ovoid mass in the epigastric and hypo gastric region. No ascitis was found. A detailed systemic examination and laboratory investigations were done which were within normal limit. Ultrasonography revealed a huge intraabdominal soft tissue mass with diffused tiny calcified spots without obvious organ of origin. Ultrasound guided FNAC was carried out from different site of the mass.

A fine needle aspiration cytology (FNAC) of the lesion revealed malignant small round cell tumour.

On laparotomy a large mass was detected in the greater omentum, without any identifiable visceral site of origin. The mass (tumour) was excised and sent for histopathological examination in 10 % formalin solution.

Grossly the specimen measured (12 x 8 x 4)cm, solid boss elated mass, hard to cut. Cut surface was gray in colour with areas of cystic changes and necrosis [**Figure 1(a), 1(b)**]. Paraffin embedded sections was prepared from different parts of the tumour and were studied by H&E stain, PAS stain and IHC stain.

Microscopically, H&E stained section showed sharply demarcated nests and trabeculae of small round tumour cells separated by a well-defined, florid desmoplastic stroma [**Figure 2(a), 2(b)**]. Irregular shaped large island of tumour cells with cystic change and necrosis are surrounded by broad band of desmoplastic stroma. [**Figure 2(c)].** Stroma is composed of mixture of fibroblastic and myofibroblastic cells.

Immunohistochemical stains were done using a panel of antibodies against Cytokeratin, Desmin, Vimentin, NSE and Chromogramin. 1HC stain for Cytokeratin shows diffuse cytoplasmic reactivity [**Figure 3(a**)]. Desmin shows perinuclear dot like pattern of staining. Desmin formed globoid or puncatate paranuclear mass within the tumour cells [**Figure 3(b)].** Immunostain for Vimentin showed positive reaction both for tumour cells and surrounding stroma [**Figure 3(c)]** Chromogranin was negative.

Taking into consideration of clinical feature, FNAC findings, appearance of the tumour, characteristic histological and immunohistoprofile, the case was diagnosed as Intra Abdominal Desmoplastic Small Round Cell Tumour.

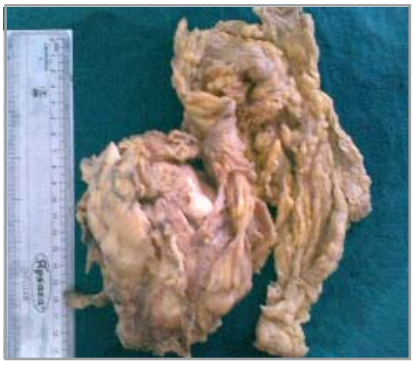

Figure 1 (a) Gross photograph of the Specimen with omentum

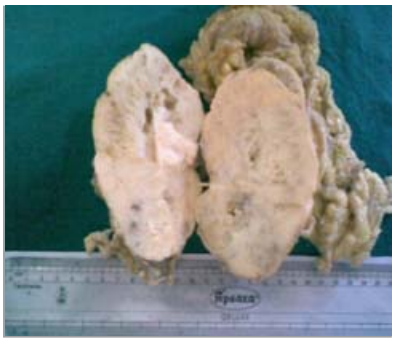

Figure 1 (b) Cut surface of the tumor showing gray-white, areas of necrosis and cystic change

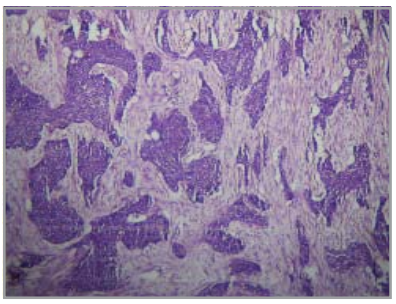

Figure 2(a) Sharply demarcated nests of tumor cells separated by a florid desmoplastic stroma (H & E X 200)

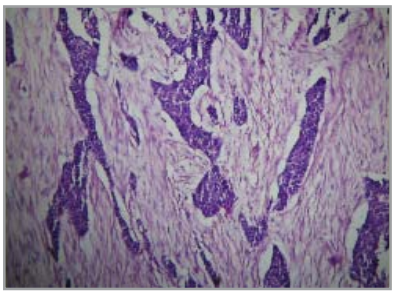

Figure 2(b) Trabeculae of tumor cells separated by a florid desmoplastic stroma (H & E X 200)

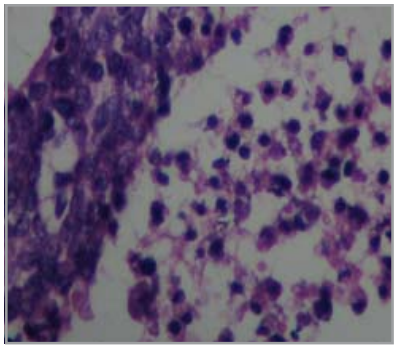

Figure 2(c) Higher magnification to show cystic space with central nacrosis in a large irregular nest, surrounded by broadband of desmoplastic stroma

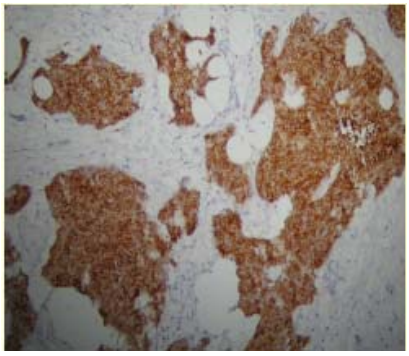

Figure 3(a) Immunohistochemical stain for Cytokeratin shows defuse cytoplasmic reactivity (I HC X 100)

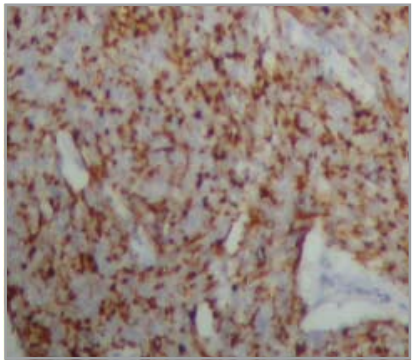

Figure 3(b) Immuno stain for Desmin shows perinuclear dot like pattern of staining (IHC X 100)

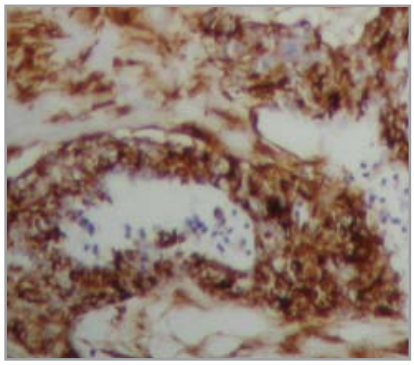

Figure 3(c) Immuno stain for Vimentin shows positive reaction both for tumor cells and surrounding stroma (IHC X 200)

DISCUSSION

Gerald W described the first case of DSRCT in 1989 and Rosai J. [4] The incidence of DSRCT is difficult to estimate because of its rarity. This aggressive malignancy is found predominantly in pediatric age group and young adult with a sex ratio (male to female) 3:1.

DSRCT is descriptively named malignancy of uncertain histogenesis. Although due to its site of presentation with predilection for serosal involvement, initially it was thought that it may be a primitive tumour of mesothelial origin. But it is now hypothesized to arise from progenitor cell with multiphenotypic differentiation. As the name implies, histologically it is composed of sharply outlined islands of small round tumour cells separated by a distinct dense desmoplastic stroma.[2,3]

An unusual feature of this tumour is its polyphenotypic differentiation with simultaneous trilinear coexpression of epithelial, neural and mesenchymal markers.[4,5,6] Tumour often expresses Desmin, Vimentin, EMA, CK, NSE and S100 protein. One of the promising markers is an antibody to Desmin. Perinuclear dot like pattern is a unique pattern of Desmin immunoreactivity peculiar to DSRCT.

Genetically the tumour is characterized by a unique reciprocal translocation (11:22) (p13/q12) involving the EWS gene on chromosome 22 & WT1 gene on chromosome 11 to produce EWS and WT1 fusion gene transcript. This transcript code for a chimeric protein acts as a transcriptional activator and fails to suppress tumour growth.[7,8]

Genetic studies provide valuable support to pathological diagnosis where rare morphological variant like stroma poor tumour, tumour with rhabdoid like cells, glandular or tubular forms or aberrant immunoreactivity is encountered.[9] Moreover to establish and to confirm diagnosis, genetic study is essential. This malignancy puts diagnostic and therapeutic problems, indeed diagnosis can be suspected by radiologic and histologic features but it is asserted by immunohistochemical and cytogenetic study[10,11]because of the large number of differential diagnosis and anatomo-pathologic polymorphism.

Intra-abdominal small round cell tumour like Neuroblastoma, PNET, extra skeletal Ewings sarcoma, Embryonal and Alveolar Rhabdomyosarcoma, NHL and some other less frequent tumour like small cells carcinoma, Undifferentiated carcinoma and malignant Mesothelioma were included in the differential diagnosis of DSRCT.

Neuroblastoma can be distinguished from DSRCT with elevated levels of urinary catecholamines and their product, presence of fibrillary neuropils and HW psendorossetes in histology and positive IHC reaction for NSE, Chromogranin, Synaptophysin and negative

reaction to EMA Vimentin. Microscopically, tip off to the diagnosis for PNET and EWS are (a)Lobular peritheliomatous growth pattern with areas of geographical necrosis and (b) PAS positive glycogen rich cytoplasm. CD99 is highly sensitive marker for PNET and EWS and negative for epithelial marker. Presence of consistent chromosomal translocation (11:22) (q24/q12) with production of chimeric fusion gene transcript EWS/FLI-1 is confirmatory for diagnosis.

Rhabdomyosarcoma (Emlryonal and Alveolar) can be differentiated from DSRCT by careful analysis of presence of variable cells like strap, racket, ribbon and tadpole with presence of bi or tri nucleation, prominent nucleoli and abundant eosinophillic cytoplasm. Immunohistochemically positive for muscle marker and negative for epithelial and neural marker. Although DSRCT shows myogenic differentiation with positive Desmin reaction MyoD1 and Myogenin are negative in DSRCT, whereas they are strongly positive in Rhabdomyosarcoma.

No curative treatment has been yet documented; current therapeutic options include multiagent chemotherapy, aggressive surgical debulking and radiotherapy. [12, 13]

CONCLUSION

Although DSRCT is a rare tumour it has distinctive clinical feature, appearance and a unique cytogenetic profile with young male predominance and aggressive biological behavior that distinguishes it from other round cell tumour.

Conflict of interest: None declared

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