Case Report

Retroperitoneal primitive neuroectodermal tumor in an adult: A rare case report and review of the literature

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ABSTRACT

Primitive neuroectodermal tumor (PNET) and Ewing's sarcoma (EWS) are small round cell tumors occurring mainly in children and adolescents. Their occurrence in adults is rare. The abdominal cavity and retroperitoneal PNET/EWS are also relatively rare, grow rapidly in size, compressing surrounding organs/large vessels, and make surgical resection difficult. We report one such rare occurrence of a retroperitoneal PNET in 41-year-old male who presented with abdominal pain and constipation. Contrast enhanced computed tomography abdomen showed large lobulated necrotic hypodense enhancing lesion extending from epigastrium to hypogastrium and involving entire abdomen. Excision of retroperitoneal mass with omentectomy was done. Microscopic examination revealed a malignant small round cell tumor with homer wright rosettes and the tumor cells were positive for CD99.

Key words: Ewing's sarcoma, immunohistochemistry, primitive neuroectodermal tumor, retroperitoneal, sarcoma

INTRODUCTION

Primitive neuroectodermal tumor (PNET) and Ewing's sarcoma (EWS) are small round cell tumors occurring mainly in children and adolescents. Their occurrence in adults is rare.[1-3] It can occur in multiple tissues and organs including kidney, adrenal, bladder, liver, small intestine, colon and rectum, with a preferred location within the chest area, limbs and around the spine. But their occurrence in abdominal, pelvic or retroperitoneal locations are rare.[3,4] Since these tumors morphologically resemble other small round cell tumors, diagnosis should confirmed by immunohistochemistry for CD99, the product of the MIC2 gene, that is expressed in more than 95% of EWS/PNETs Here we present a case of the retroperitoneal PENT in a 41-year-old man and discuss the clinical features, histopathological/immunohistological characteristics, diagnosis, treatment and prognosis.

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CASE REPORT

A 41-year-old male patient presented with complaints of abdominal fullness since 2 months and constipation since 20 days. On examination abdomen was protuberant and umbilicus was inverted. There was fullness in bilateral flanks. Contrast enhanced computed tomography (CECT) abdomen showed large lobulated, necrotic, hypodense enhancing lesion extending from epigastrium to hypogastrium and involving entire abdomen. The lesion was abutting the anterior abdominal aorta, displacing bowel loops laterally [Figure 1]. Excision of retroperitoneal mass was done along with omentectomy, resection and anastomosis of transverse colon. Gross examination of the specimen sent to us revealed a soft tissue mass measuring 16 cm × 16 cm × 11 cm, cut surface of which revealed a variegated appearence with

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solid, cystic, hemorrhagiec and necrotic areas. Sixteen tumor nodules were identified in the mesentry. On microscopic examination, there were lobules of tumor cells seperated by fibrous septa. The tumor cells comprised of small cells with round vesicular nuclei, inconspicuous nucleoli, scanty eosinophilic cytoplasm with frequent homer wright rosettes and pseudorosettes [Figure 2]. The tumor cells were positive for CD99 [Figure 3].

DISCUSSION

PNET and EWS are malignant tumors composed of small round cells, frequently occur in the soft tissue and bone. They have a common neuroectodermal origin, with similar morphological conformation, specific chromosomal translocation which is related to the EWS gene on chromosome 22, belonging to the PNET/EWS family tumors. PNET and EWS were once believed to be different types of tumors because of their different differentiations: PNET belongs to neuroectodermal differentiation whereas EWS tends to be undifferentiated. With the advances in pathology, bone EWS, the peripheral neuroepithelioma/PNET, Askin's tumor, as well as other types of tumors with their classification still in debate are now all considered as PNET/EWS family tumors.[5,6]

PNET and EWS occur mainly in children and adolescents, rarely in adults.[1-3] It can occur in multiple tissues and organs including kidney, adrenal, bladder, liver, small intestine, colon and rectum, with a preferred location within the chest area, in the limbs and around the spine. However their occurrence in abdominal, pelvic or retroperitoneal locations are rare.[3,4] To the best of our knowledge, not more than 33 cases of retroperitoneal PNET have been reported in the literature.[7]

The CECT appearance of PNET is nonspecific, but it is the widely used diagnostic modality for identifying this tumor. CECT often shows heterogenous lobulated mass displacing the adjacent organ. Histopathologically, the characteristic findings are well-defined clusters of undifferentiated small round cells, surrounded by desmoplastic stroma. The tumor cells are characterized by small hyperchromatic nuclei with scanty cytoplasm and presence of Homer Wright rosettes. PNET typically expresses high amount of MIC2 (CD99) antigen and exhibits highly characteristic chromosomal translocation t (11:22) (p13:q12), which leads to fusion of WTI and EWS genes. In our case, the diagnosis was established by typical histopathological features along with positivity for CD99 antigen.

The differential diagnosis of this group of neoplasms is characterized by small, round, relatively undifferentiated cells and include EWS, rhabdomyosarcoma, synovial sarcoma, nonHodgkinia lymphoma, neuroblastoma, hepatoblastoma and nephroblastoma. Other differential

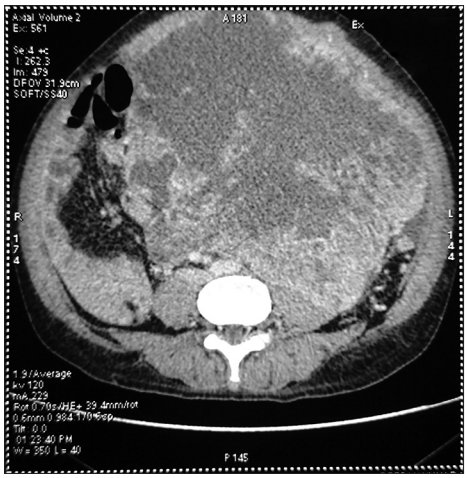

Figure 1: Contrast enhanced computed tomography abdomen showed large lobulated necrotic hypodense enhancing lesion involving entire abdomen

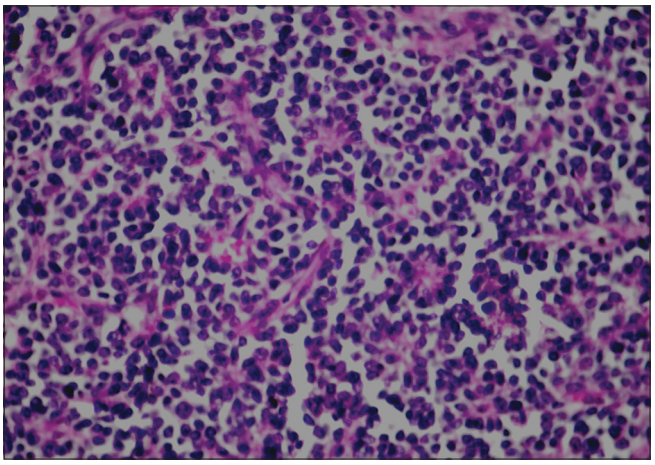

Figure 2: Photomicrograph showing sheets of small round cells with homer wright rosettes (H and E, ×40)

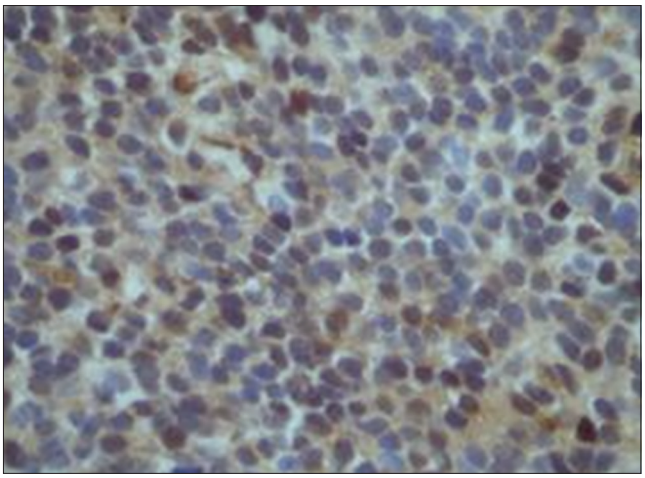

Figure 3: Tumor cells revealing CD99 positivity (IHC, ×40)

diagnoses of small round cell tumors include small cell osteogenic sarcoma, undifferentiated hepatoblastoma, granulocytic sarcoma, and intra-abdominal desmoplastic small round cell tumor. EWS/PNET is characterized by a balanced chromosomal translocation which generates a fusion transcript of the EWS gene and the Friend leukemia virus integration 1. Thus the diagnosis should be confirmed on immunohistochemistry for CD99 which is the product of the MIC2 gene, and is expressed in more than 95% of EWS/PNETs with a diffuse membranous staining pattern.

Although the prognosis is poor, survival can be improved with multi-modal approach. Lal *et al*.[8] recommended surgical resection of >90% of the tumor burden. They used adjuvant chemotherapy with P6 protocol along with radiation to abdominopelvic region and reported significant survival.[8] There are isolated case reports highlighting the role of neoadjuvant chemotherapy in management of these lesions.[9] Sable *et al*.[7] subjected the patient to induction chemotherapy and the tumor responded to it by reduction in its size and vascularity which was documented on postchemotherapy CECT. Postchemotherapy en block resection of the mass was done and the patient received concurrent chemoradiation. The patient was kept under regular follow-up and remained asymptomatic over 2 years. Thus, a down staging approach with induction chemotherapy of large borderline operable retroperitoneal PNETs would increase the resectability rates, translating into improved survival.[7]

Baldini *et al*.[2] showed that 5 years survival for adults with EWS/PNET was 49% in those having localized disease treated with combined modality.[7] His results also showed favorable predictors of survival being primary origin from the bone, size <8 cm, and favorable objective response to chemotherapy.[2] However in our case, patient suffered from respiratory distress postoperatively and was diagnosed with iatrogenic left sided pneumothorax. Patient subsequently succumbed to cardiac arrest and expired.

CONCLUSION

Though PNET is a rare tumor in the retroperitoneum of adults, it should be considered in the differential diagnosis of small round cell tumors occurring in this site and the diagnosis should confirmed by immunohistochemical positivity for CD99. Retroperitoneal PNETs have poor prognosis and survival can be improved with multimodal treatment.

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Conflicts of interest

There are no conflicts of interest.

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