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Case Report

Primary uterine rhabdomyosarcoma: a unique presentation

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ABSTRACT

Uterine rhabdomyosarcoma in adults is extremely rare. We present the case of a 61-year-old P1L1 postmenopausal female who came with abdominal distension and pedal edema since, 20 days. The typical presentation of post-menopausal bleeding was absent in this patient. The clinical presentation, massively raised CA 125 levels and CT findings pointed towards an ovarian malignancy. USG and MRI leaned towards carcinoma endometrium. The final diagnosis was clinched on immunohistochemical studies. The tumor had an aggressive course with a fatal outcome within days.

Keywords: Uterine rhabdomyosarcoma, Immunohistocytochemistry, MRI

INTRODUCTION

Rhabdomyosarcoma is the most frequently diagnosed soft tissue tumor found in children, comprising up to nearly 50% of all paediatric soft tissue tumors.[1] Adult primary Rhabdomyosarcoma of gynaecologic origin is an uncommon phenomenon being extremely rare and challenging to diagnose.[2] Rhabdomyosarcoma can originate anywhere in the body; the head and neck region being most frequent site of involvement in children and deep soft tissue of limbs in adults. Cervix is the most common gynaecological site of adult rhabdomyosarcoma.[3] Histopathologically uterine Rhabdomyosarcoma is divided into three major categories classified on behaviour and prognosis, namely pleomorphic, alveolar and embryonal types with embryonal further subclassified into botryoid and spindle shape variants.[2] The prognosis of uterine rhabdomyosarcoma specifically is extremely unfortunate, with the vast majority of patients presenting with extensive disease at time of clinical diagnosis as was seen with our patient.[4] Owing to the scarcity of literature available, the optimal therapy of adult patients with rhabdomyosarcoma of gynaecologic origin is not defined.

CASE REPORT

A 61-year-old P1L1 postmenopausal woman presented with the chief complaints of abdominal distension, and pedal oedema since, 20 days. There was abdominal pain since, 1 day localized to the lower abdomen which was insidious in onset, dull aching and non-radiating. This was associated with shortness of breath, loss of appetite, lethargy and easy fatigability. Patient was a known case of hypertension since, two years on medications. No significant past family and surgical history. She had one uncomplicated pregnancy 30 years ago and delivered vaginally at full term. She had her menarche at 14 years of age and had attained menopause 11 years ago. Her past menstrual cycles were regular. There was no history of any oral contraceptive pills or hormonal replacement therapy taken. On examination general condition was fair with bipedal edema. There was reduced air entry on both sides on auscultation with saturation being 96% on room air. On per abdomen examination the abdomen was found to be distended with gross ascites. On per speculum examination the cervix appeared to be bulky but healthy. No bleeding. On per vaginal examination the uterus size could not be assessed. USG Abdomen and pelvis showed multiple

hypoechoic lesions in the liver with free fluid in the abdomen. The uterus was bulky in size 10.6×6.4×7.2 cm with a hypoechoic mass in the endometrial cavity extending up to the endocervical canal measuring 9.6×5.3 cm. no necrotic areas. No vascularity on colour doppler? Ca endometrium There were peritoneal deposits seen in the pelvis. Doppler studies of limbs were normal. ECG and 2d echo were normal. Laboratory investigations showed HB-11 wbc-11,000. Liver, kidney function tests and INR was within normal limits. CA-125 was massively raised 2140 (normal <35). In view of massive pleural effusion and gross ascites thoracocentesis and ascitic fluid tapping were done and fluid was sent for analysis which came out to be positive for adenocarcinoma. CECT abdomen plus pelvis plus thorax showed enlarged uterus with homogeneously enhancing mass in endocervical canal with locoregional lymphadenopathy. Bilateral bulky ovaries. Massive asictis. Peritoneal thickening, hepatic and pulmonary metastasis. Massive right sided pleural effusion.no brain metastasis. MRI pelvis showed and enlarged uterus with a 9.6×4.5 cm mass distending the uterine cavity and endocervical canal. Bilateral ovaries were bulky with multiple peritoneal deposits and ascites.

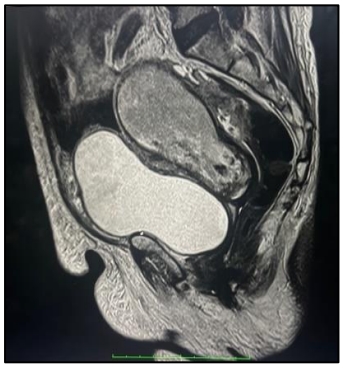

Figure 1: Sagittal section of MRI showing a growth in the endocervical canal and uterine cavity.

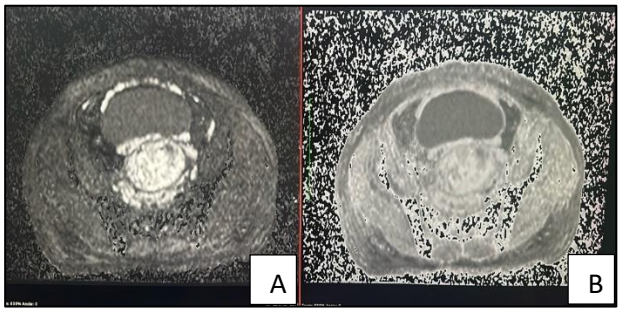

Figure 2 (A and B): AXIAL section showing hyperintensity on DWI and low values on ADC sequences corresponding to diffusion restriction in the highly cellular mass.

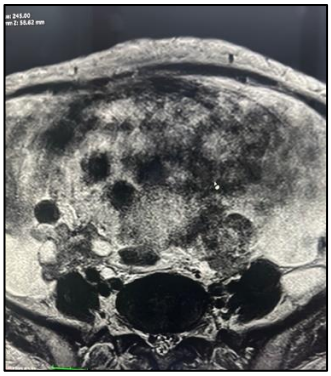

Figure 3: Axial section of MRI showing bilateral bulky ovaries.

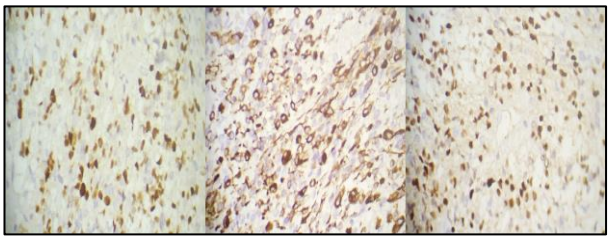

Figure 4: IHC study positive for DESMIN, Myogenin and Myo1d.

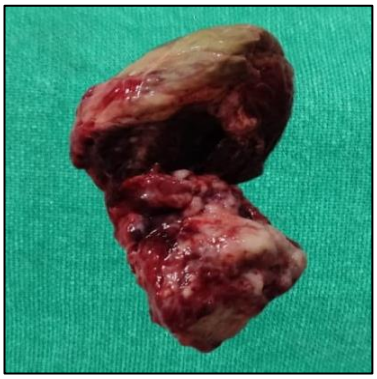

Figure 5: Fleshy mass passed per vaginally by patient at home.

An endometrial biopsy was taken which showed poorly differentiated neoplasm with sections showing necrosis and tumor cells which have nuclei that are round to oval, hyperchromatic with variably positive nucleoli and clear to eosinophilic cytoplasm. d/d poorly differentiated carcinoma and high-grade sarcoma.

Immunohistochemistry was done showed tumor cells positive for DESMIN, MYOGENIN and MYOD1. Heterogenous positive for CD10. P53 wild type positivity. Tumor cells negative for SMA, Caldesmon, Cylind1, CD99, Synaptophysin, CD45, ER, PR, CK, WT1, CK and CD117. Favours pleomorphic Rhabdomyosarcoma. The patient gave history of passage of a fleshy mass per vaginally at home. The case was referred to a medical oncologist who advised palliative radiotherapy. However,

before treatment could be initiated the patient succumbed due to advanced stage of the tumor.

DISCUSSION

Rhabdomyosarcoma is a malignant neoplasm that originates from undifferentiated myogenic progenitor cells. It is predominantly a paediatric disease, and its occurrence in adults is exceedingly rare. Adult rhabdomyosarcoma is exceedingly uncommon, accounting for less than 4% of all soft tissue sarcomas specifically, and 1% of all malignancies generally.[5,6]

Cervix happens to be the most common site of occurrence in adult gynaecological malignancies as was seen by occurrence of growth in endocervical canal in our case. Clinically, vaginal bleeding is the most frequent presenting symptom in patients with uterine rhabdomyosarcoma which was absent in our case which gave the clinical picture of an ovarian malignancy due to widespread metastasis.[3] Histopathologically it is extremely difficult to differentiate and rule out rhabdomyosarcoma from leiomyosarcoma, high-grade endometrial stromal sarcoma, adenosarcoma and carcinosarcoma.[6] The positive IHC for Myogenin and MyoD1 and negative IHC for Caldesmon and estrogen receptor can be helpful in confirming the diagnosis.[3] Pleomorphic (60%-70%) is the most frequent histopathological type is and correlates with poor prognosis as was seen in our case.

Alveolar being the least common (less than 5%), characterized genetically by FOXO1 chromosomal rearrangements and associated with unfavourable prognosis. Lastly, embryonal accounts for 30%-40% of all uterine Rhabdomyosarcomas, characterized molecularly by (KRAS)/p53 mutations and correlates with good prognosis.[7] The treatment strategies for uterine rhabdomyosarcoma are not well defined and include an aggressive multimodality therapy comprising combination chemotherapy, radiotherapy and surgery, whenever possible. Ferrari and colleagues explored the treatment outcomes in 171 adult patients with RMS originating from various sites. The reported chemotherapy response and five-year OS rates were 85% and 40%, respectively.[8] Little and colleagues scrutinized the multimodal (surgery, radiotherapy and chemotherapy) treatment outcomes in 82 adult patients with RMS. The 10-year OS and DFS rates were 40% and 41%, respectively.[9]

CONCLUSION

Uterine rhadomyosarcoma in adults is extremely rare having a dismal prognosis. The diagnosis is often challenging and delayed. Histopathological examination in addition to immunohistochemistry and cytogenetic studies can aid in the definitive diagnosis. Aggressive multimodality therapy yields better clinical outcomes.

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