

Case Report

Rhabdomyosarcoma Presenting as Metastatic Abdominal or Chest Lumps: Report of 2 Cases

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Abstract: Rhabdomyosarcoma is a malignant mesenchymal tumor with skeletal muscle differentiation. It is the most common soft tissue sarcoma in children and adolescents. Its incidence is 4.5 cases per 1 million people aged 0-20 year. Embryonal, alveolar, pleomorphic and sclerosing /spindle cell types are the major variants of rhabdomyosarcoma. Embryonal rhabdomyosarcoma is the most common type. Rhabdomyosarcoma is highly aggressive and usually patient presented with pressure symptoms or metastasis. We are reporting 2 cases of rhabdomyosarcoma with one patient presented with abdominal wall nodules and other patient with bilateral breast lumps.

Keywords: Rhabdomyosarcoma, embryonal, metastatic, alveolar.

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INTRODUCTION

Rhabdomyosarcoma is the most common soft tissue sarcoma in children and adolescents. It is rare in adults accounting to <1% of all rhabdomyosarcomas. Embryonal rhabdomyosarcoma is the most common subtype followed by alveolar variant. Pleomorphic rhabdomyosarcomas occurs in adults, spindle cell/sclerosing type affects all ages, embryonal and alveolar types usually affects children and adolescents. Rhabdomyosarcomas most commonly arising in the head and neck region, genitourinary tract and retroperitoneum. These tumors usually present with an aggressive clinical course with an over all poor prognosis.

Case 1

21 year old male presented with abdominal pain for 2 weeks duration, On examination 2 painless firm nodules of 3 x 2 cm. size identified, one at infraumbilical region and one at back of trunk.

No palpable lymphadenopathy or splenomegaly. Moderate ascites was present. Systemic examination, routine blood examination, peripheral

smear, biochemical parameters and bone marrow study were normal.

USG abdomen showed a large mass in epigastric region and a well defined lesion in subcutaneous plane of infraumbilical region. Moderate hepatomegaly with secondaries seen in right lobe of liver. Moderate ascites present

Colour doppler- USG scrotum showed a 0.6 x 0.5 cm well defined hypoechoic lesion within the left testicular parenchyma.

CECT thorax and abdomen – showed well defined retroperitoneal soft tissue density in upper abdomen, 12.4x10.4x9cm, Left lower cervical and mediastinal lymphnodes. Multiple lesions seen in both lobes of liver Ascites present.

No lung infiltrates or metastasis seen. Features were suggestive of malignant retroperitoneal lymphoma

FINE NEEDLE ASPIRATION CYTOLOGY from the abdominal wall swelling was taken features were suggestive of round cell neoplasm.

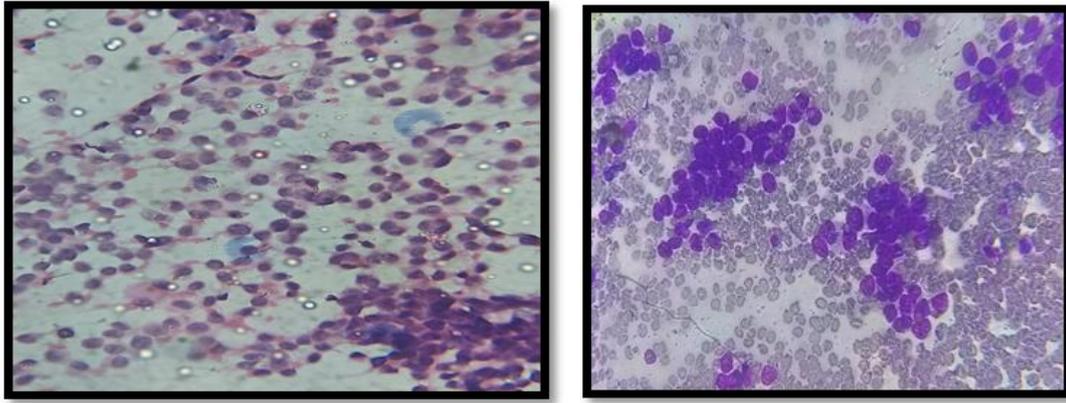


Figure 1: Fine needle aspiration cytology of abdominal wall nodule showing round cells

MACROSCOPY - Single fibrofatty tissue, cut section whitish with dark brown areas

MICROSCOPY- Fibrocollagenous tissue and adipose tissue with an infiltrating neoplasm composed of cells arranged in alveolar pattern and in sheets. Individual

cells are round/oval with scanty to moderate cytoplasm, round hyperchromatic nucleus. 3-4 mitosis/10hpf noted. No necrosis seen.

Features- suggestive of round cell sarcoma.

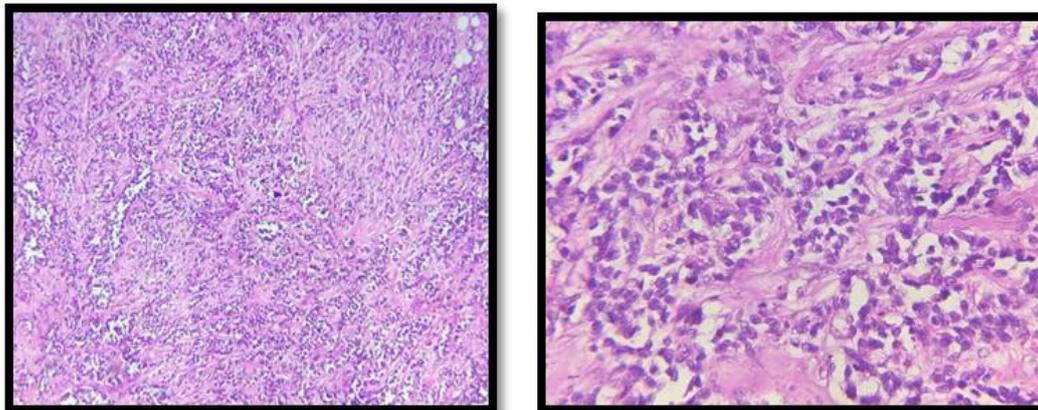


Figure 2: Histopathology images showing cells arranged in alveolar pattern

IMMUNOHISTOCHEMISTRY- Neoplastic cells are
CK, LCA: Negative

Vimentin: Cytoplasmic positivity
Myogenin: Strong nuclear positivity
NKX2.2: Negative

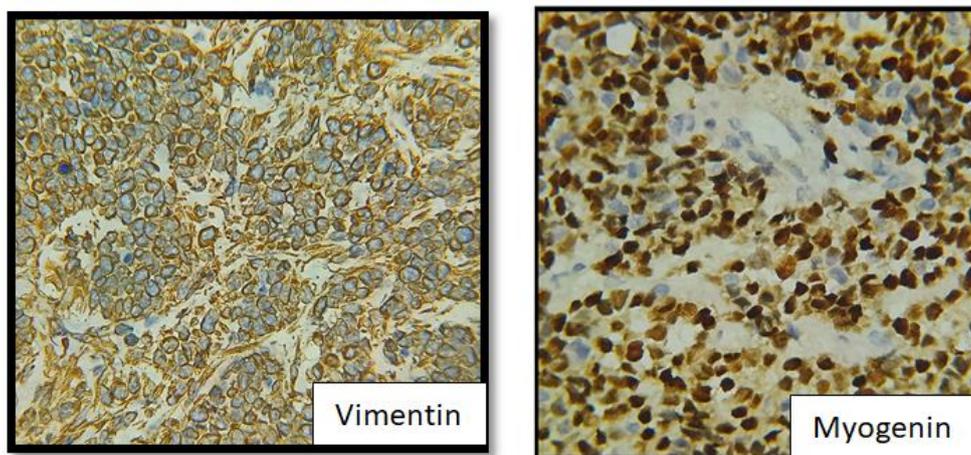


Figure 3: Neoplastic cells showing cytoplasmic positivity for vimentin and strong nuclear positivity for myogenin

**DIAGNOSIS: ALVEOLAR RHABDOMYOSARCOMA
Case 2**

17 year old girl presented with a painless lump in (R) breast, which gradually increasing in size for 2 month duration.

No family history of carcinoma breast. On examination multiple lumps of varying sizes identified in both breasts. No generalized or axillary lymphadenopathy.

Systemic examinations, routine blood examinations and biochemical investigations were normal.

USG breast- showed multiple lesions in both breast, largest is in right breast, 3x2 cm

CEMRI breasts- multiple well defined lobulated soft tissue lesions of varying size in both breasts, larger one at upper outer quadrant of right breast measuring 4.2x4x3.9 cm

CT scan neck, chest and pelvis - showed sclerotic vertebral lesions in lower dorsal and lumbar spine.

¹⁸F FDG Whole body high definition PET/CT scan

FDG avid multiple soft tissue density lesions in bilateral breast parenchyma, FDG avid lytic areas in sacrum, acetabulum and D11 vertebral body. FDG avid soft tissue thickening along plantar aspect of left foot.

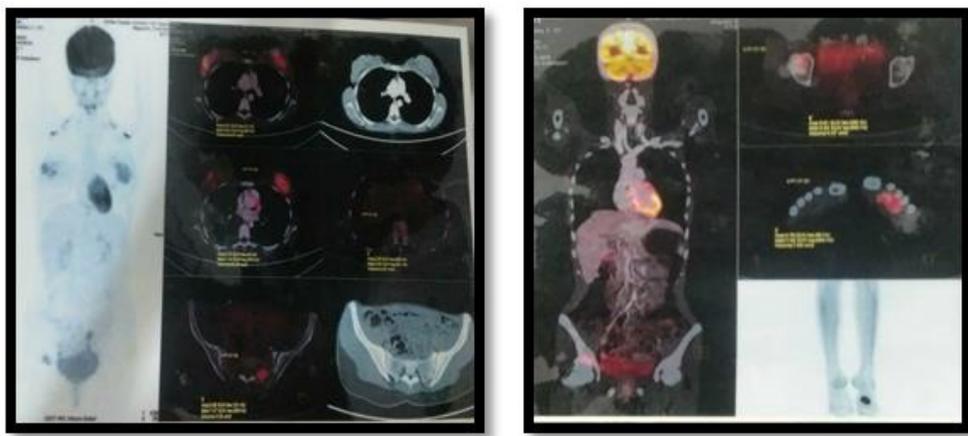


Figure 4: PET Scan showing FDG avid lesions

MICROSCOPY

Excision biopsy of the lump in right breast was done in another centre and diagnosed as malignant adenomyoepithelioma. Slide review done in our centre

and features were suggestive of a small round cell neoplasm.

Trucut biopsy of bilateral breast lesions taken, histopathological features were consistent with small round neoplasm.

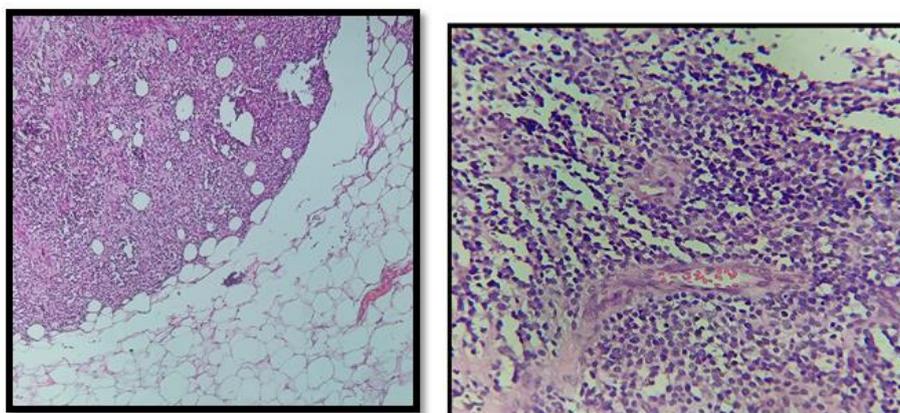


Figure 5: Histopathology showing sheets and nests of round cells

IMMUNOHISTOCHEMISTRY- Neoplastic cells are

CK, LCA : Negative
EMA, Synaptophysin : Negative

Vimentin, Myogenin : Positive
CD99 : Patchy positivity
Ki67 : 80%

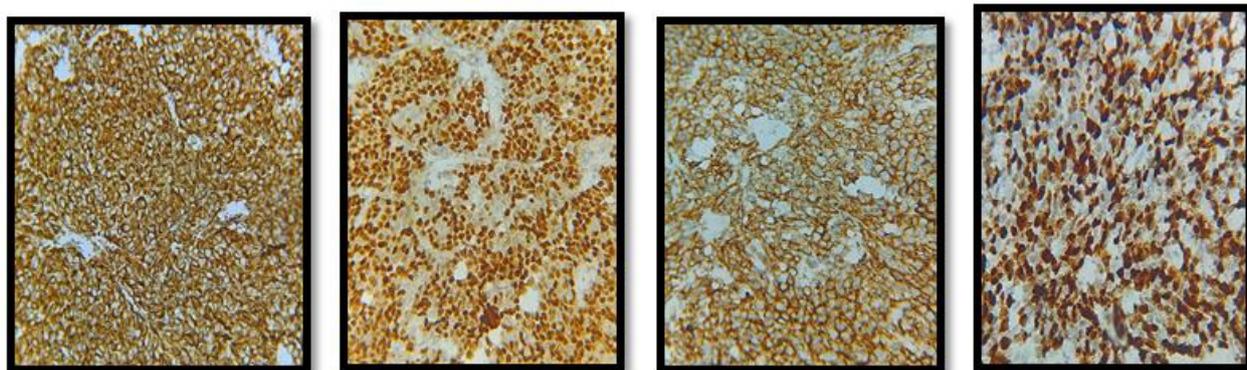


Figure 6: Immunohistochemistry studies –neoplastic cells showing strong positivity for vimentin (cytoplasmic), myogenin (nuclear), and patchy positivity for CD99. Ki67 proliferation index=80%

DIAGNOSIS: EMBRYONAL RHABDOMYOSARCOMA MANAGEMENT

Both patients were treated with Vincristine, Dactinomycin and cyclophosphamide. Both of them showed excellent response to chemotherapy.

Multimodality treatment including surgery, chemotherapy and radiotherapy can provide an improved outcome.

FOLLOW UP

- Repeat PET CT/ other radiological imaging showed excellent response in patient with alveolar rhabdomyosarcoma.
- Patient with breast lesions showed multiple stable residual sclerotic and lytic areas.
- Both patients are on regular follow up.

DISCUSSION

Rhabdomyosarcoma is the most common soft tissue sarcomas in children and adolescents.

Embryonal rhabdomyosarcoma is the most common type with one third of cases occurring in children aged less than 5 years of age.

It is a malignant soft tissue tumor with morphological and immunophenotypical features of embryonal skeletal muscles. Head and neck region and genitourinary system are the most frequent sites. Biliary tract, retroperitoneum or abdomen are less frequent sites.

Its incidence is 4.5 cases per 1 million people aged 0-20 years. Slightly more common in males. Clinical symptoms generally related to mass effects.

Embryonal rhabdomyosarcoma is associated with several syndromes such as Costello syndrome (H RAS gene mutation), Neurofibromatosis type 1, Noonan syndrome, Beck with wiedemann syndrome, DICER1 syndrome and Li Fraumeni syndrome (T P53 mutation)

Sporadic cases of embryonal rhabdomyosarcoma are aneuploid with whole chromosome gains including polysomy 8, extra copies of chromosomes 2,11,12,13 and or 20. Histopathology shows primitive mesenchymal cells in various stages of myogenesis. Stellate cells represent most primitive form of this spectrum. Scattered differentiating rhabdomyoblasts also can be seen. Neoplastic cell are positive for desmin, myogenin and or MYOD1. Lack of FOXO1 gene rearrangement is characteristic. Patients aged 1-9 years have better outcome than infant or adolescent.

Alveolar rhabdomyosarcoma (ARMS) is a malignant neoplasm composed of a monomorphic population of primitive round cells showing skeletal muscle differentiation. It is the second most common rhabdomyosarcoma constituting about 25%. ARMS most commonly arises in the deep soft tissue of the extremities. Head and neck, paraspinal region and perineal region are the other common sites. Peak incidence is seen among individuals aged 10-25 years.

Histopathology shows highly cellular neoplasm composed of primitive round cells with scant cytoplasm and hyperchromatic nuclei arranged in alveolar pattern or in sheets. Rhabdomyoblastic differentiation of ARMS is shown immunohistochemically with positive reactions to desmin, myogenin and MYOD1. Nuclear expression of myogenin is strong and diffuse.

Approximately 85% of histologically diagnosed alveolar rhabdomyosarcomas contain characteristic fusion genes. PAX3-FOXO1 (70-90%) and PAX7-FOXO1 (10-30%) are the major fusion genes. The prognosis for patients with fusion- positive ARMS is worse than for those with fusion negative rhabdomyosarcoma and embryonal rhabdomyosarcoma.

Pleomorphic rhabdomyosarcoma is a high grade pleomorphic sarcoma, usually of adults composed of bizarre brightly eosinophilic polygonal, round and spindle cells that display skeletal muscle differentiation. These tumors are most common in sixth to seventh

decades of life and more common in males. Usually arise in the deep soft tissue of lower extremity.

Etiology is unknown, genome-wide surveys have identified recurrent losses of DNA, gains, and amplifications. This copy number patterns differs from that found in either alveolar or embryonal rhabdomyosarcoma.

Pleomorphic rhabdomyosarcomas are large well marginated tumors with a pseudo capsule and a whitish or fleshy cut surface. Tumors are composed of sheets of large atypical and frequently multinucleated, polygonal spindled or rhabdoid cells with eosinophilic cytoplasm. Metastasis to lung are common. Neoplastic cells show strong positivity with desmin and myogenin. These are highly aggressive sarcomas with bad prognosis.

Spindle cell / Sclerosing rhabdomyosarcoma is a type of rhabdomyosarcoma that has fascicular spindle and or sclerosing morphology. Accounts for 3-10% of rhabdomyosarcomas. The head and neck region is the most common site of involvement followed by extremities. VGLL2, CITED2, EWSR1 mutations can be detected by FISH studies. MYOD1 mutation can be identified by PCR or by Sanger sequencing. Congenital /infertile rhabdomyosarcomas with gene fusion show a favourable clinical course. MYOD1- mutant spindle cell / sclerosing rhabdomyosarcoma follow an aggressive course despite multimodality therapy and have a poor prognosis.

CONCLUSION

Rhabdomyosarcoma is the most common malignant soft tissue tumor in children and adolescents that shows skeletal muscle differentiation. It is rare in adults accounting to <1% of rhabdosarcomas. These tumors usually present with an aggressive clinical course with an over all poor prognosis. An accurate diagnosis with the help of histopathology and immunohistochemistry are essential to initiate a multimodality treatment. Neoadjuvant chemotherapy, surgery and adjuvant chemoradiation therapy can increase the patient survival.

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