

Solid variant of alveolar rhabdomyosarcoma with patent ductus arteriosus

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ABSTRACT

Alveolar rhabdomyosarcoma is a high-grade malignant soft tissue tumor of mesenchymal origin. It is a rare tumor with an annual incidence of one per million. It is generally seen between 10 and 25 years of age. We present a rare case of solid variant of alveolar rhabdomyosarcoma in a 6-month-old child with patent ductus arteriosus. There was no evidence of lung or lymph node metastasis. She was treated by wide local excision.

Key words: Alveolar rhabdomyosarcoma, Ewing's sarcoma, patent ductus arteriosus, solid variant of alveolar rhabdomyosarcoma

INTRODUCTION

Alveolar rhabdomyosarcoma (ARMS) is a subtype of the rhabdomyosarcoma family of soft tissue tumors. ARMS tend to occur within skeletal muscle. The embryonic derivation of these tumors is presumed to be mesoderm. It is the second most common rhabdomyosarcoma, accounting for approximately 31% of all rhabdomyosarcoma.^[1] It represents approximately 1% of all malignancies among children and adolescents.^[2] It is important to recognize this variant as it has a worse prognosis than embryonal variety.^[3]

CASE REPORT

A 6-month-old female child, known case of patent ductus arteriosus (PDA) presented in pediatric outpatient department with the complaint of a painless and gradually progressive swelling over back since 2 months. There was no history of trauma. Two-dimensional (2D) echo revealed a moderate size (6mm) PDA with left to right shunt and normal pulmonary artery pressure. On examination, a firm,

nontender, 5 cm × 5 cm sized swelling was noted at the medial aspect of left scapular region. Contrast enhanced computed tomography (CECT) thorax was suggestive of well-defined mass lesion in subcutaneous plane posterior to erector spinae muscle on left side at T₅-T₉ vertebral level. Magnetic resonance imaging (MRI) dorsal spine revealed a well-defined soft tissue lesion of approximately 5.3 × 4.8 × 2.3 cm in subcutaneous tissue superficial to erector spinae muscle on left side, consistent with the diagnosis of neoplastic mass [Figure 1a and b]. Fine needle aspiration cytology (FNAC) of the swelling showed cellular smear with presence of small, primitive round to polygonal cells arranged dyscohesively, singly or in cords. The cell had scanty to moderate cytoplasm and hyperchromatic eccentric nuclei, with prominent nucleoli [Figure 2a]. Cytoplasmic tails were seen in some cells. Cytoplasm showed diffuse periodic acid Schiff (PAS) positivity. Mitotic figures were noted. Possibility of a malignant round cell tumor, with the differential diagnosis of Ewing's sarcoma/primitive neuroectodermal tumor (PNET) and embryonal rhabdomyosarcoma (ERMS) were considered. Excision biopsy was advised.

The excised specimen showed a firm to soft greyish growth with brownish areas having irregular but well-defined margins embedded in surrounding fibrofatty tissue and muscle. The growth measured 5.0 cm × 3.5 cm × 1.5 cm. Microscopic examination revealed tumor composed of small round cells predominantly in solid sheets with few fibrovascular septae in between. The cells showed moderate pleomorphism, round to oval hyperchromatic eccentric

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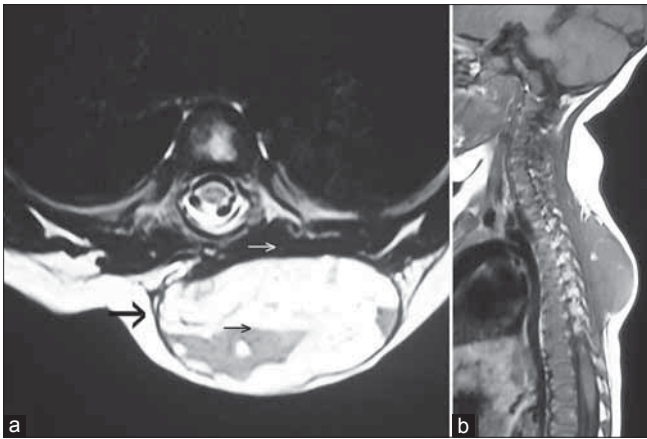


Figure 1: (a) Magnetic resonance imaging dorsal spine (axial view) at the level of T7 vertebra showing well-defined lesion in subcutaneous tissue plane (thick black arrow) superficial to erector spinae muscle (white arrow). The lesion is heterogeneously hyperintense with multiple fluid levels within it (thin black arrow). (b) Sagittal view showing the lesion at T5-T9 vertebral level

nuclei, and scanty deeply eosinophilic cytoplasm [Figure 2b and c]. Mitotic activity was brisk. There were a few alveoli-like spaces. Areas of hemorrhage and necrosis were also seen.

Immunohistochemistry (IHC) markers revealed strong nuclear staining with myogenin [Figure 2d]. CD 99, leukocyte-common antigen (LCA), synaptophysin, and S-100 were negative. The findings were consistent with solid variant of alveolar rhabdomyosarcoma (SARMS).

Patient had a recurrence three months later. Wide local excision was done followed by skin grafting. Recurrent tumor also had similar histology. Patient was advised radiotherapy. She is on regular follow-up and is disease free during four months observation.

DISCUSSION

A small round cell sarcoma in a child calls for the differential diagnosis among ERMS, neuroblastoma, PNET/Ewings sarcoma, lymphoma, granulocytic sarcoma, melanotic neuroectodermal tumors, sclerosing small round cell tumors and other tumors having predominantly small round cells. Factors to be considered are complete clinical data like patient age, location of the growth, duration, size and results of other investigations, especially radiological findings. Multiple sections need to be studied for specific features like alveolar structures, rhabdomyoblastic differentiation, rosettes, intracytoplasmic glycogen/pigment etc., In this case with a large tumor over the scapular region with FNAC showing small primitive round cells with eccentric nuclei, absent lymphoglandular bodies, or rosettes and PAS positive cytoplasm suggested Ewings sarcoma or ERMS. Bone involvement was excluded by CT and MRI. Negative immunoreactivity for CD 99 also excluded Ewing's sarcoma/PNET.

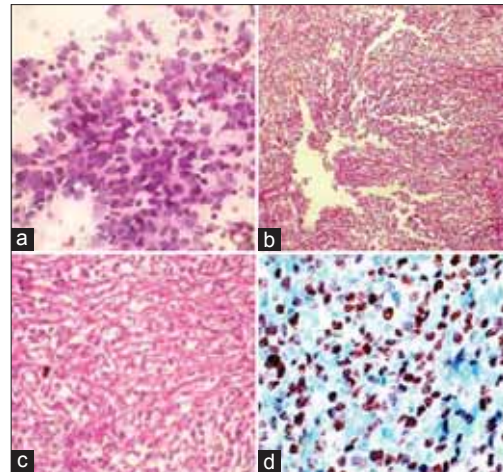


Figure 2: (a) Fine needle aspiration cytology of the swelling showing large, round to polygonal cells with scanty to moderate cytoplasm and hyperchromatic nuclei, with nucleoli (hematoxylin and eosin (H and E), $\times 400$). (b) Photomicrograph showing small round cell proliferation in solid pattern with vague alveolar pattern. (H and E, $\times 100$). (c) High power view showing round cell population (H and E, $\times 400$). (d) Immunohistochemistry showing strong nuclear staining with myogenin

The solid pattern of growth interrupted by few fibrovascular septae, vague alveolar pattern and the rhabdomyoblastic morphology evident in some cells favored a diagnosis of SARMS. There was no loose myxoid area or spindling of cells usually seen with other variants of ERMS. The immunostaining with myogenin confirmed our suspicion.^[4]

SARMS with solid sheets of small round cells are particularly prone to be confused with Ewings or lymphoma in this age group. The conventional classification of rhabdomyosarcoma into embryonal, botryoid, alveolar and pleomorphic types as proposed by Horn and Enterline in 1958. It was later modified to include SARMS by Tsokos.^[5,6] This variant tends to occur in younger children as compared to classical variant which tends to occur in older children and adolescents (10-25 years) with predilection for deep soft tissues of extremities. Our case is peculiar in that it occurred in a 6-month-old child with PDA. Literature search did not show any case with such association.

Most ARMS cases are associated with a specific translocation, $t(2;13)(q35;q14)$ ^[7] or its variant $t(1;13)(q36;q14)$ ^[8] resulting in PAX3-FKHR or PAX7-FKHR fusion. SARMS seem to be more likely than non-solid types to be fusion negative.^[9] We could not perform the cytogenetic test in our case.

Lymph node, lung and bone marrow are the common sites of metastasis. A quarter of patients have metastatic disease at the time of diagnosis.^[2] Our patient did not show any metastasis probably due to early detection. Risk factors that influence outcome of ARMS include primary site, size of primary tumor, extent of local spread and the presence of nodal and distant metastasis.^[2] Total excision of the tumor is

part of the recommended therapy for rhabdomyosarcomas in the trunk and extremities.^[10]

CONCLUSION

We report a rare association of SARMS in a 6-month-old child with PDA. A multidisciplinary approach comprising of histology, immunohistochemistry, chromosome analysis, and molecular cytogenetics play an important role in the diagnosis of this tumor.

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