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IP Journal of Diagnostic Pathology and Oncology

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Case Report

Non paratesticular spindle cell variant of RMS in a three year old

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ARTICLE INFO

Article history:
Received 16-07-2020
Accepted 25-07-2020
Available online 03-09-2020

Keywords:
Paediatric
Rhabdomyosarcoma
Spindle cell

ABSTRACT

Rhabdomyosarcoma (RMS) is a malignant mesenchymal (myogenic) tumor , and diagnosed on cytology as one of the Blue round cell tumors of paediatric age group.
Here we are reporting a case of right side abdominal mass in a three-year-old child whose FNA smears, along with immunocytochemistry was diagnosed as RMS. The intriguing finding on smears was the presence of a large number of malignant elongated cells which stained with both desmin and myogenin, as crisply as the round cell component.
The diagnosis of Spindle cell variant of subtype, embryonal was further confirmed on histopathology sections and extended IHC panel.
This variant is itself rare and nonparatesticular regions are further infrequent in paediatric population.

1. Introduction

Rhabdomyosarcoma (RMS) is a malignant mesenchymal tumor of myogenic origin. It is the most common childhood and adolescent sarcoma showing features of skeletal muscle differentiation. Three major variants with characteristic histologic appearances are embryonal, alveolar, and pleomorphic. Embryonal Rhabdomyosarcoma accounts for approximately 60% of all rhabdomyosarcomas. It mostly affects children below 10 years of age (mean is 7 years), adolescents and young adults and is uncommon in patients older than 40. [1]

Alveolar RMS is associated with a specific chromosomal translocation, t(2;13)(q35;q14), or its variant, t(1;13)(p36;q14), resulting in the formation of a fusion gene, PAX3-FKHR or PAX7-FKHR , while recurrent genetic alteration has not been identified in either embryonal and pleomorphic RMS. [2]

However recurrent NCOA2 gene rearrangements in a small subset of spindle cell RMS occurring in infants has been noted. [3]

2. Case

A three-year-old male child presented with rapidly enlarging abdominal mass and pain with occasional vomiting, for the past two months.

USG guided FNA was performed and smears stained with Hematoxylin & Eosin (H&E) and Giemsa.

Smears revealed moderate cellularity comprising of few round pleomorphic cells accompanied by a large proportion of spindled atypical cells. Myogenin and desmin (immunocytochemical markers for tumors of muscle origin) were positive in both components of malignant cells (Figures 1 and 2).

Diagnosis of 'Round cell tumor suggestive of Rhabdomyasarcoma' was issued.

His blood parameters and bone marrow examination showed normal hematopoiesis. Core biopsy done priorly at another institute suggested Wilms tumor.

CECT abdomen and thorax revealed a relatively well defined cystic lesion with peripheral as well as internal irregular enhancing areas in abdominopelvic region, predominantly on right side suggesting neoplastic etiology. (Figure 4)

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https://doi.org/10.18231/j.jdpo.2020.068

Biopsy (Figure 3) from the mass was submitted and H&E stained sections showed a malignant mesenchymal neoplasm comprising of spindle shaped cells arranged in nodules and fascicles (>50% of neoplasm).

Individual cells were pleomorphic, having irregular elongated nuclei, scant cytoplasm, intermixed with the hyalinising bands of collagen fibres. A small population of plump to polygonal rhabdomyoblasts with prominent nucleoli and eosinophilic cytoplasm, focal necrosis, myxoid degeneration, apoptosis were also found. Mitotic count was 3-6/10 hpf.

Immunohistochemistry (IHC) resulted as

Myogenin and Desmin: Positive;

CD34, S100, beta catenin: Negative;

KI67: 30%

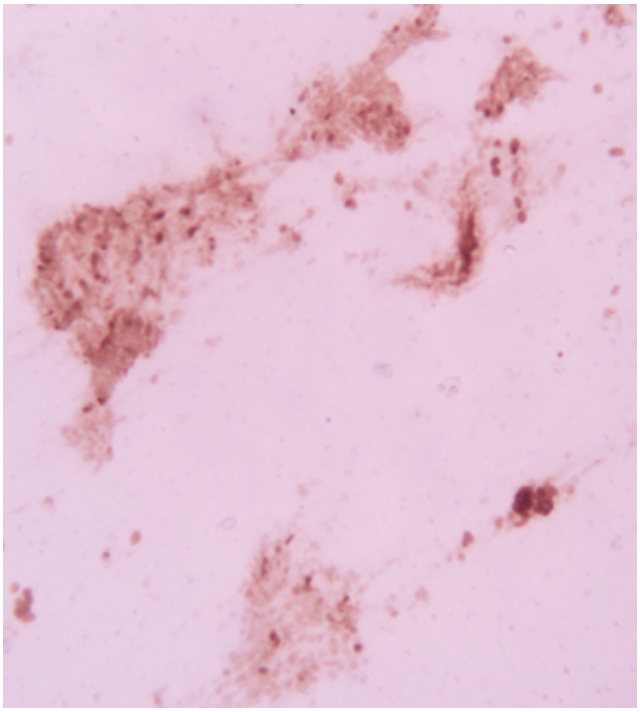

Fig. 1: FNA: Rhabdomyoblasts Desmin Positive (400X)

2.1. Diagnosis

Rhabdomyosarcoma -spindle cell variant of embryonal was signed out

3. Discussion

Spindle cell variant of embryonal RMS was firstdescribed in children in 1992.[4] Common locations of occurrence are paratesticular and head -neck regions. Other rarely affected locations are viscera and retroperitoneum.[5]This variant constitutes 6% of embryonal rhabdomyosarcoma with male to female ratio of 6:1[6]

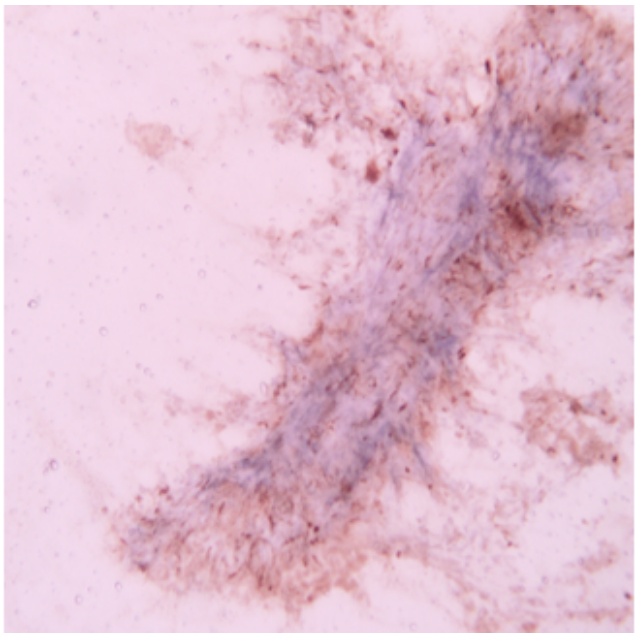

Fig. 2: FNA: Spindled cells Myogenin Positive (400X)

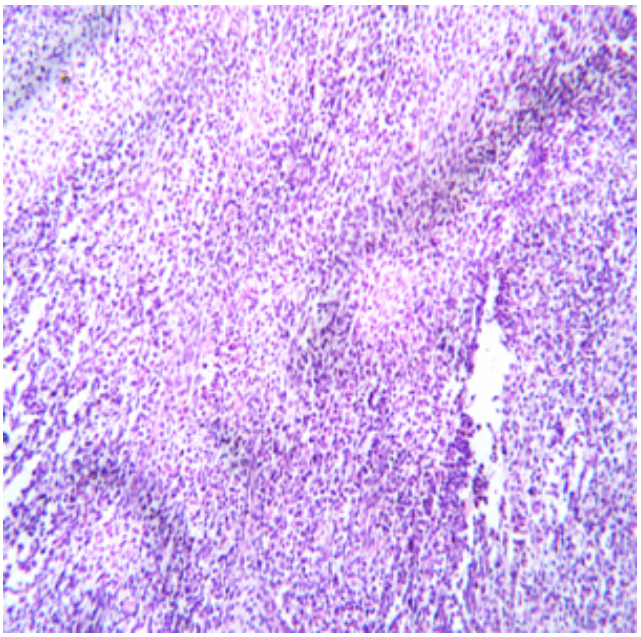

Fig. 3: H&E Rhabdomyoblasts wrapped by spindled cells (H&E, 400X)

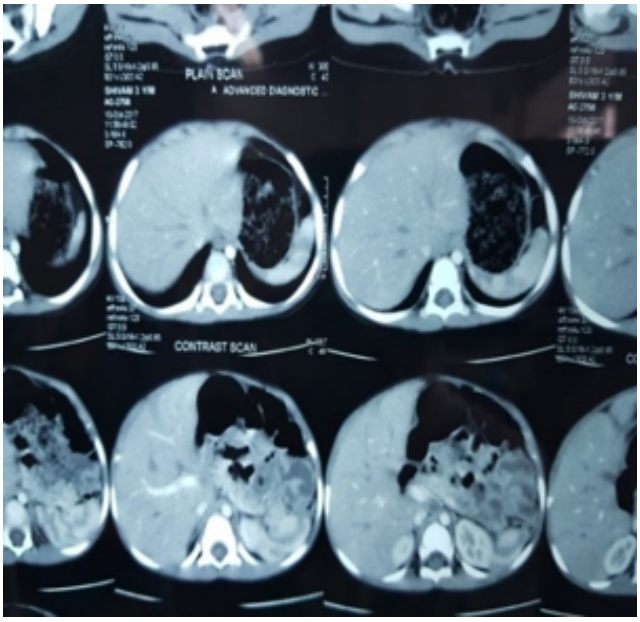

Fig. 4: CECT Abdomen showing neoplastic mass on right side

Cytogenetic studies reveal aneuploidy with mostly whole chromosome gains as well as non-recurrent structural changes.[7,8] PAX3- and PAX7-FOXO1 fusions are virtually always absent.[9]

Recently, frequent and recurrent MyoD1 homozygous p.L122R mutations have been identified as pathognomonic events in adult-type spindle cell RMS[10–12] and have unfavorable prognosis because of broader morphological spectrum, including sclerosing and pseudovascular types.[12]

MYOD1 mutant tumors also showed coexistent PIK3CA mutations.[11,13,14] Rare tumors with sclerosing morphology were also found to harbour mutations in FGFR4[15]

Mutation-negative and fusion-negative spindle cell/sclerosing rhabdomyosarcomas have a propensity to arise in genitourinary or intraabdominal locations and follow a favorable clinical course.

In paediatric population it is diagnosed at an early stage. Lymph node metastasis is seen in approximately 16% patients.[5]

Major prognostic factors in these tumors are resectability, tumor size (which also correlates with resectability), histologic subtype, and tumor stage.[16]

A specific grouping system for Spindle Cell-RMS has not been identified hence this variant falls into the general grouping system.

Group I completely resected tumors without lymph node involvement

Group II is composed of cases with complete resection and lymph node involvement or microscopic residual disease.

Group III includes patients with gross residual disease.

Group IV contains tumors with distant metastases.

This grouping system is used to plan additional radiation therapy for the tumor.[17]

In children with any histological type of rhabdomyosarcoma, the multimodality approach has led to a cure rate of approximately 70%[18] with 95% survival at 5 years.[4]

Paratesticular lesions appear to have an even better prognosis than tumors of nonparatesticular sites.[19]

In adult patients there is worse prognosis with recurrence and high metastasis rate.

The differentials in this case were

Fibrosarcoma: herring bone pattern was missing, rhabdomyoblasts were present

Infantile fibromatosis: beta catenin was negative

Leiomyosarcma: Usually high grade with necrosis and mitosis and myoglobin would be negative

Rhabdomyoma: Benign tumor without necrosis or rhabdomyoblasts

Nonparatesticular regions are frequent in adults, here we are reporting such a case in a three-year-old child.

4. Source of Funding

None.

5. Conflict of Interest

None.

References

1. Ognjanovic S, Linaberyam, Charbonneau B, Ross JA. Trends in childhood rhabdomyosarcoma: incidence and survival in the United States. *Cancer*. 1975;115(18):4218–26.
2. Barr FG. Gene fusions involving PAX and FOX family members in alveolar rhabdomyosarcoma. *Oncogenesis*. 2001;20(40):5736–46.
3. Mosquera JM, Sboner A, Zhang L, Kitabayashi N, Chen CL. Recurrent NCOA2 gene rearrangements in congenital/infantile spindle cell rhabdomyosarcoma. *Genes Chromosomes Cancer*. 2013;52:538–50.
4. Cavazzana AO, Schmidt D, Ninfo V, Harms D, Tollot M. Spindle cell rhabdomyosarcoma. A prognosticallyfavorable variant of rhabdomyosarcoma. *Am J SurgPathol*. 1992;16:229–35.
5. Leuschner I. Spindle cell variants of embryonal rhabdomyosarcoma in the paratesticular region. A report of the intergroup rhabdomyosarcoma study. *AM J SurgPatho*. 1993;17:221–230.
6. Chiles MC. Sclerozing rhabdomyosarcomas in children and adolscents: a clinicopathologic review of 13 cases from the intregroup rhabdomyosarcoma study group and children's oncology group. *Pediatr pathol*. 2004;7:538–94.
7. Cores R. Adult sclerosing rhabdomyosarcoma: cytogenetic link with embryonal rhabdomyosarcoma. *Virchows Arch*. 2005;446:64–7.
8. Zambrano E. pediatric sclerosing rhabdomyosarcoma. *Int J Surg Pathol*. 2006;14:193–9.
9. Vadagama B. sclerosing rhabdomyosarcoma in childhood: case report and review of literature. *Pediatr Dev Pathol*. 2004;7:391–6.
10. Kohsaka S, Shukla N, Ameur N, Ito T, Ng CK. A recurrent neomorphic mutation in MYOD1 defines a clinically aggressive subset of embryonal rhabdomyosarcoma associated with PI3K-AKT pathway mutations. *Nat Genet*. 2014;46:595–600.
11. Agaram NP, Chen CL, Zhang L, LaQuaglia MP, Wexler L, Antonescu CR. RecurrentMYOD1mutations in pediatric and adult sclerosing

and spindle cell rhabdomyosarcomas: Evidence for a common pathogenesis. *Genes, Chromosomes Cancer*. 2014;53(9):779–87.
12. Szuhai K, de Jong D, Leung WY, Fletcher CD, Hogendoorn PC. Transactivating mutation of the MYOD1 gene is a frequent event in adult spindle cell rhabdomyosarcoma. *J Pathol*. 2014;232(3):300–7.
13. Owosho AA, Huang SC, Chen S. A clinicopathologic study of head and neck rhabdomyosarcomas showing FOXO1 fusion-positive alveolar and MYOD1-mutant sclerosing are associated with unfavorable outcome. *Oral Oncol*. 2016;61:89–97.
14. Rekhi B, Upadhyay P, Ramteke MP, Dutt A. MYOD1 (L122R) mutations are associated with spindle cell and sclerosing rhabdomyosarcomas with aggressive clinical outcomes. *Mod Pathol*. 2016;29(12):1532–40.
15. Alaggio R, Zhang L, Sung YS. A molecular study of pediatric spindle and sclerosing rhabdomyosarcoma: identification of novel and recurrent VGLL2-related fusions in infantile cases. *Am J SurgPathol*. 2016;40(2):224–235.
16. Fletcher CD. Distinctive soft tissue tumors of the head and neck. *Mod Pathol*. 2002;15(3):324–330.
17. Raney RB, Anderson JR, Barr FG, Donaldson SS, Pappo AS, Qualman SJ, et al. Rhabdomyosarcoma and Undifferentiated Sarcoma in the First Two Decades of Life: A Selective Review of Intergroup Rhabdomyosarcoma Study Group Experience and Rationale for Intergroup Rhabdomyosarcoma Study V. *J Pediatr Hematol/Oncol*. 2001;23(4):215–20.
18. Punyko JA, Mertens AC, Gurney JG, Yasui Y, Donaldson SS, Rodeberg DA, et al. Long-term medical effects of childhood and adolescent rhabdomyosarcoma: A report from the childhood cancer survivor study. *Pediatric Blood Cancer*. 2005;44(7):643–53.
19. Wa N, Gehan EA, Webber BL. Classification of rhabdomyosarcomas and related sarcomas: pathologic aspects and proposal for a new classification-an Intergroup Rhabdomyosarcoma Study. *Cancer*. 1995;15(6):1073–85.

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Cite this article: Jaiswal R, Yadav S, Dubey DB. Non paratesticular spindle cell variant of RMS in a three year old. *IP J Diagn Pathol Oncol* 2020;5(3):347-350.