OPEN ACCESS

Saudi Journal of Pathology and Microbiology
Abbreviated Key Title: Saudi J Pathol Microbiol
ISSN 2518-3362 (Print) |ISSN 2518-3370 (Online)
Scholars Middle East Publishers, Dubai, United Arab Emirates
Journal homepage: https://saudijournals.com

Case Report

Spindle Cell / Sclerosing Rhabdomyosarcoma in Childhood: About A Case and Review of the Literature

Chadi Fadwa[1*], Rouas Lamiaa[2], Lamalmi Najat[3]

[1]Senior Resident, Department of Pediatric Pathology, Ibn Sina University Hospital, Rabat, Morocco
[2]Senior Professor, Department of Pediatric Pathology, Ibn Sina University Hospital, Rabat, Morocco
[3]Senior Professor and Head, Department of Pediatric Pathology, Ibn Sina University Hospital, Rabat, Morocco

DOI: 10.36348/sjpm.2021.v06i04.001 | **Received:** 28.02.2021 | **Accepted:** 01.04.2021 | **Published:** 06.04.2021

***Corresponding author:** Chadi Fadwa

Abstract

Rhabdomyosarcoma is a malignant mesenchymal tumor with striated muscle differentiation. Most commonly developing in children or adolescents. Previously three subtypes have been described: embryonic, alveolar and pleomorphic. Spindle cell / sclerosing rhabdomyosarcoma is a rare new subtype described in the latest edition of the WHO soft tissue, it develops in children and adults and it is subdivided into 3 genomic groups with different prognosis according to the latest advances in cytogenetics. We report a case of sclerosing rhabdomyosarcoma in a 15-year-old boy with painful swelling of the forearm that has progressed for 3 months. Microscopic examination shows entirely tumor fragments, consisting of an eosinophilic hyaline matrix delimiting tumor lobules and nests, sometimes with a pseudo-vascular and alveolar appearance. Tumor cells have small, irregular and hyperchromatic nuclei with coarse chromatin and eosinophilic cytoplasm. Rare rhabdomyoblast cells are noted. Immunohistochemistry shows expression of Desmine; Myogenin; CD99; Myo D1 and loss of SMA expression.

Key words: Rhabdomyosacoma, sclerosing, childhood, histopathology.

INTRODUCTION

Rhabdomyosarcoma (RMS) is the most common malignant soft tissue tumor seen in children. WHO recently classified RMS into 4 groups including embryonal, alveolar, pleomorphic and Spindle cell/sclerosing rhabdomyosarcoma subtypes. Spindle cell / sclerosing rhabdomyosarcoma accounts for 5-10% of rhabdomyosarcomas and it is subdivided into 3 genomic groups with different prognosis. It affects infants, children, and adults. Although it affects both sexes overall. Clinically, it presents as a rapidly growing and painful soft tissue mass associated with signs of local compression. The treatment consists of a complete resection of the tumor associated with systemic chemotherapy with or without radiotherapy.

CASE REPORT

We report a case of a 15-year-old boy with painful swelling of the forearm that has progressed for 3 months. Microscopic examination shows entirely tumor fragments, consisting of an eosinophilic hyaline matrix delimiting tumor lobules and nests, sometimes with a pseudo-vascular and alveolar appearance. Tumor cells have small, irregular and hyperchromatic nuclei with coarse chromatin and eosinophilic cytoplasm. Rare rhabdomyoblast cells are noted. Immunohistochemistry shows expression of Desmine; Myogenin; CD99; Myo D1 and loss of SMA expression.

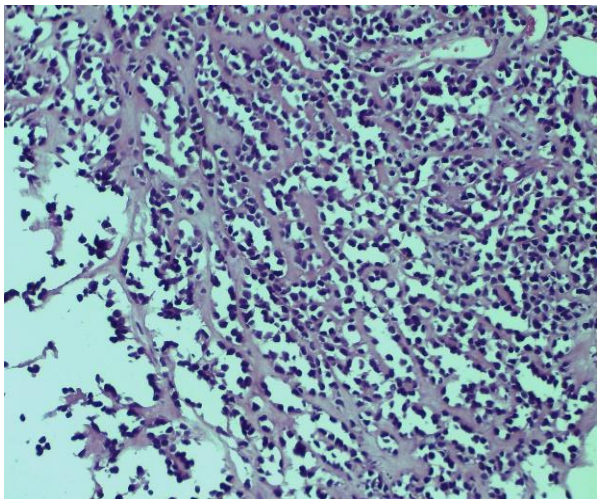

Fig-1: Eosinophilic hyaline matrix delimiting tumor lobules and nests (HEx4)

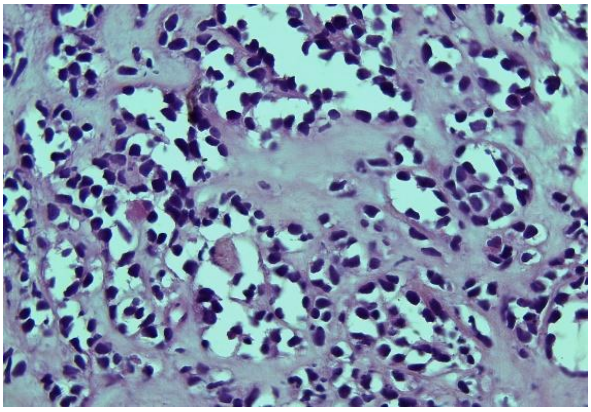

Fig-2: Eosinophilic hyaline matrix (HEx40)

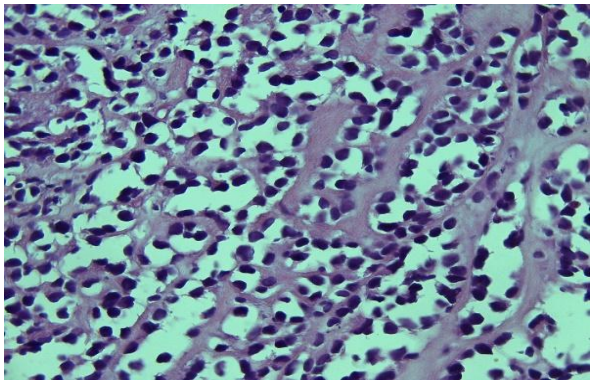

Fig-3: Pseudo-vascular and alveolar appearance (HEx40)

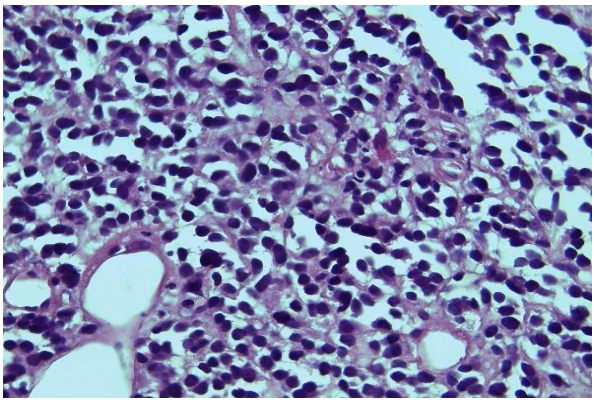

Fig-4: Small, irregular and hyperchromatic nuclei (HEx40)

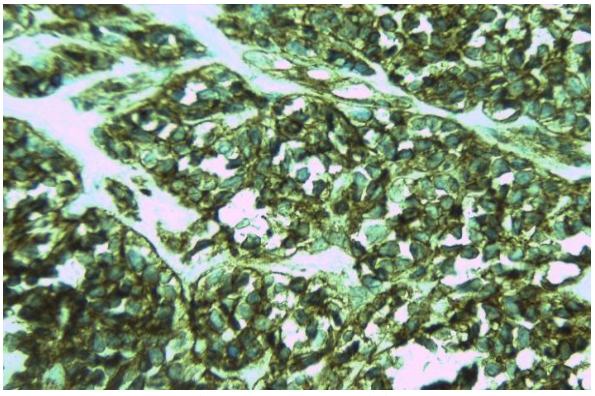

Fig-5: CD99 positive

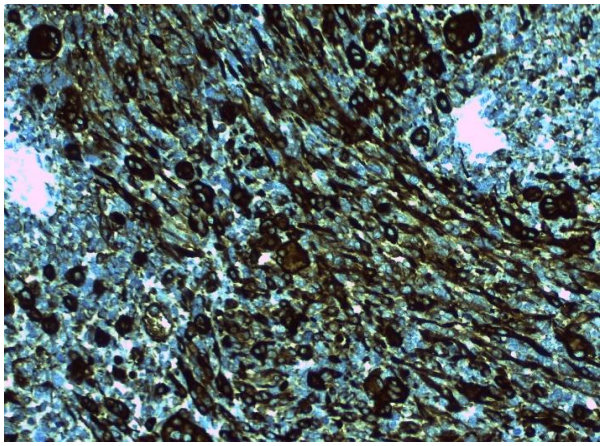

Fig-6: Desmine positive

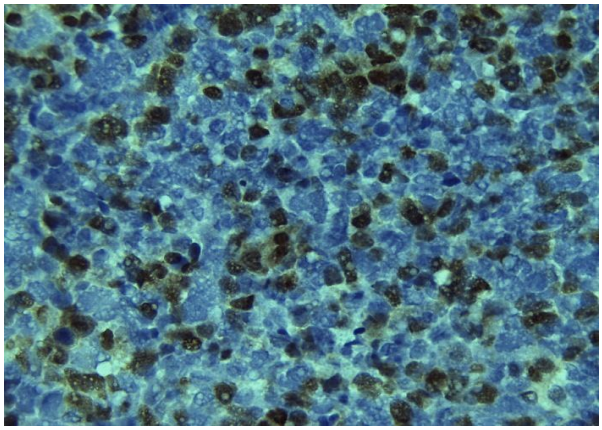

Fig-7: Myogenin positive

DISCUSSION

Rhabdomyosarcoma (RMS) is a rare and aggressive malignancy that may originate from primitive mesenchymal cells that arise anywhere in the body, including sites where striate muscle is not found [1]. It can be seen in both children and adults [2]. It has been traditionally classified into 3 main subtypes: embryonal, alveolar and pleomorphic. However a new, rare subtype called Spindle cell/sclerosing Rhabdomyosarcoma has been defined recently and classified as the fourth variant by the World Health Organization in 2013 [2]. Embryonal and Spindle cell/sclerosing subtypes arising especially in children and adolescents and especially have excellent prognosis. In contrast, alveolar and pleomorphic subtypes have a worse clinical outcome [2]. RMSs are common in children, representing 5% of all childhood cancers [1] with a peak incidence in those aged less than 4 years [3]. Different classification systems for RMSs have been described and according to the International Classification for Childhood Sarcomas, pediatric RMSs have been classified into embryonal (ERMS), alveolar (ARMS), botryoid, and spindle cell subtypes. The pleomorphic subtype is predominantly seen in adults [4]. Subtyping of RMS is important as they differ in clinical, histological, molecular and prognostic features from the other subtypes. This is

important especially in children and adolescents, where prognostically relevant subgroups are defined and treated by different protocols [2]. Mentzel and Katenkamp first reported a distinctive variant of RMS called Sclerosing, Pseudovascular Rhabdomyosarcoma; characterized by prominent hyaline sclerosis and a pseudovascular growth pattern in 2000. In 2002, Folpe et al. presented 4 new cases in adults with similar features and they entitled this variant as Sclerosing rhabdomyosarcoma [2]. The occurrence of a sclerosing pattern in pediatric rhabdomyosarcomas has been briefly reported [3]. The literature search identified 163 reported cases of ssRMS. Although ssRMSs have been reported in all age groups. There was a mild predilection for boys/men (60%) [1]. In children the most common site of involvement is the paratesticular region, however deep soft tissues in head and neck are the foremost localizations in adults [2]. Frequently, ssRMSs show mildly high intensity on T2W and iso-intensity on T1W images compared to the muscles. On post-contrast images, ssRMSs have shown heterogeneous enhancement [1]. The size of the lesion varied from 3.2 cm to 10.5 cm [4]. Microscopically, the prominent hyaline matrix separate the undifferentiated round or oval tumor cells into cords, nests, or small alveolar patterns, frequently reminiscent of sclerosing epithelioid fibrosarcoma, chondrosarcoma or even angiosarcoma [5]. Rare to scattered rhabdomyoblasts can be seen usually throughout the tumor. Nuclear atypia, hyperchromasia and mitotic figures are common [2]. Intratumoral hemorrhages and necrotic changes are occasionally seen in ssRMSs, as in the other subtypes [1]. By immunohistochemistry, tumor cells usually show strong and diffuse positivity for desmin and MyoD1, and a variable extent of nuclear reactivity for myogenin from focal to diffuse pattern [5]. The pancytokeratin, neural, and neuroendocrine markers are negative [4]. Although the optimal treatment for spindle cell/sclerosing RMS has been not reached, the mainstay therapeutic method should also, similar to most soft tissue tumors, be surgery and adjuvant chemotherapy or radiotherapy can be added [5]. The treatment failure rate for pediatric SRMS is high (43.75%) [4]. The VAC (Vincristine, Actinomycin D and Cyclophosphamide) regimen is one of the recommended therapeutic options. More than half of these tumors recur or progress locally or distantly without bone marrow invasion [2].

CONCLUSION

Rhabdomyosarcoma (RMS) is the most common malignant soft tissue tumor seen in children. Sclerosing rhabdomyosarcoma is a new subtype included in the World Health Organization (WHO) 2013 classification as Spindle cell/sclerosing Rhabdomyosarcoma (SSRMS). This rare entity is characterized by particular clinicopathological and prognostic aspects that will have to be recognized by the pathologist and clinician in order to find an optimal therapeutic strategy to improve the prognosis.

REFERENCES

1. Momosaka, D., Togao, O., Hiwatashi, A., Yamashita, K., Yoshimoto, K., Mori, M., ... & Honda, H. (2017). Spindle cell/sclerosing rhabdomyosarcoma with intracranial invasion without destroying the bone of the skull base: a case report and literature review. Acta radiologica open, 6(8), 2058460117727316.
2. Çelik, Ö. İ., Dere, Y., Çelik, S. Y., & Tuna, B. (2016). Sclerosing Rhabdomyosarcoma of the Lip in a Child; A Case Report. Muğla Sıtkı Koçman Üniversitesi Tıp Dergisi, 3(2), 15-17.
3. Vadgama, B., Sebire, N. J., Malone, M., & Ramsay, A. D. (2004). Sclerosing rhabdomyosarcoma in childhood: case report and review of the literature. Pediatric and Developmental Pathology, 7(4), 391-396.
4. Kumar, A., Singh, M., Sharma, M. C., Bakshi, S., & Sharma, B. S. (2014). Pediatric sclerosing rhabdomyosarcomas: a review. International Scholarly Research Notices, 2014.
5. Zhao, Z., Yin, Y., Zhang, J., Qi, J., Zhang, D., Ma, Y., ... & Zhou, J. (2015). Spindle cell/sclerosing rhabdomyosarcoma: case series from a single institution emphasizing morphology, immunohistochemistry and follow-up. International journal of clinical and experimental pathology, 8(11), 13814.