Case Report

# Spindle cell rhabdomyosarcoma in the adult: A rare case report

Ashlesha Jakkampudi[1], Sajitha Kaliyath[1], Padmaraj Hegde[2], Michelle Mathias[1], Vikram Shetty[2]

[1]Department of Pathology, K. S. Hegde Medical Academy, Nitte University, [2]Department of Oral and Maxillofacial Surgery, A B Shetty Memorial Institute of Dental Sciences, Mangalore, Karnataka, India

**Abstract**

Spindle cell/sclerosing rhabdomyosarcoma (RMS) is an uncommon type of RMS and has been classified as a separate entity by the WHO in 2013. It affects both children and adults with a greater incidence in males. These tumors can pose a diagnostic challenge and can be difficult to differentiate from other spindle cell malignant tumors in the head and neck. Here, we report a case of spindle cell/sclerosing RMS in a young woman presenting with a swelling on the left side of the face of 3 months duration. A careful correlation with the radiographic images, histopathological findings and immunohistochemistry helped to arrive at a diagnosis.

**Keywords:** Immunohistochemistry, rhabdomyosarcoma, spindle cell

**Address for correspondence:** Dr. Sajitha Kaliyath, Department of Pathology, K S Hegde Medical Academy, Nitte University, Deralakatte, Mangalore, Karnataka, India.
E-mail: drsk29@hotmail.com

**Submitted:** 25-Jul-2021, **Accepted:** 21-Dec-2021, **Published:** 28-Feb-2022

## INTRODUCTION

Rhabdomyosarcoma (RMS) is a rare soft-tissue malignant neoplasm that arises from the undifferentiated mesenchyme and histologically resembles the normal fetal skeletal muscle before innervation. The current incidence of RMS encompassing all ages in the head-and-neck region is 0.041 cases per 100,000 people.[1]

Three histological variants of RMS have been conventionally described: embryonal, alveolar and pleomorphic.[2] RMS comprise the most common soft-tissue sarcomas in children, and of these, embryonal sarcoma is the most common type.[3] The variants included under embryonal RMS include sarcoma botryoides, anaplastic type and spindle cell type. Spindle cell RMS are rare tumors forming only 5%–10% of all RMS cases and are now considered a distinct entity, separate but related to embryonal RMS. There is a male preponderance with a male-to-female ratio of 6:1.[3] It is associated with a favorable outcome in comparison with other subtypes. We report a case of this rare tumor presenting in a 27-year-old female as a progressively increasing swelling over the parotid and temporal regions and posed a challenge at the time of diagnosis.

## CASE REPORT

A 27-year-old female patient presented with the complaints of swelling over the left side of face involving parotid and temporal region of 3 months duration. The swelling had rapidly increased in size over the previous 4 weeks. There were no associated systemic symptoms and no facial

Access this article online

Quick Response Code:

Website: www.jomfp.in

DOI: 10.4103/jomfp.jomfp_251_21



**How to cite this article:** Jakkampudi A, Kaliyath S, Hegde P, Mathias M, Shetty V. Spindle cell rhabdomyosarcoma in the adult: A rare case report. J Oral Maxillofac Pathol 2022;26:S103-6.

muscle weakness. There were no significant illnesses in the past. Extraoral inspection showed gross facial asymmetry, dumbbell-shaped swelling measuring 6 cm × 3 cm in dimensions, extending from lateral canthus of left eye medially to preauricular region, superiorly from temporal region to lower border of mandible inferiorly. Extraocular movements were full and conjugate. On extraoral palpation, the swelling was firm in consistency and no local rise of temperature was noted. Tenderness was present on palpation over the swelling. On intraoral inspection, the swelling was noted in the molar teeth region. Ulceration was noted in the left posterior buccal mucosa. Intraoral palpation was not possible due to restricted mouth opening.

Magnetic resonance imaging of the neck revealed a 5.8 cm × 6.3 × 8.4 cm heterogeneously enhancing soft-tissue mass arising from masticator space with the erosion of ramus of mandible, adjacent base of the skull and minimal extension to the mandibular nerve through foramen ovale. Posteriorly, the lesion was also abutting anterior border of superficial and deep lobes of the left parotid gland with the loss of fat planes with superficial lobe. The possibility of a malignant tumor probably malignant peripheral nerve sheath tumor (MPNST) was considered.

Ultrasound-guided fine-needle aspiration cytology for cytological diagnosis of this lesion was inconclusive with scantily cellular material comprising few ductal epithelial cells, plenty of fibrous tissue fragments, mixed inflammatory cells.

A core biopsy was performed and showed a neoplasm comprising of nests and cords of tumor cells with scant-to-moderate eosinophilic cytoplasm and oval nuclei with minimal nuclear atypia in a fibrous and myxoid stroma. A diagnosis of a low-grade spindle cell neoplasm was considered and two differential diagnoses were made – first, of myoepithelial rich salivary gland neoplasm and the second, spindle cell neoplasm of possible neural origin. Immunoprofile showed the possibility of a low-grade tumor of myoepithelial origin with CD-34, cytokeratin and calponin positivity and ki-67 showing 15%–20% in the highest proliferating area. Stains for smooth muscle actin (SMA), STAT-6, P63, S100 and desmin were negative. In view of the small size of the biopsy and therefore the possibility of nonrepresentativeness, it was decided to proceed with excision of the tumor.

Gross specimen received in the histopathology laboratory was irregular pale white to tan yellow soft-tissue mass with attached muscle at the superior surface [Figure 1]. Cut surface

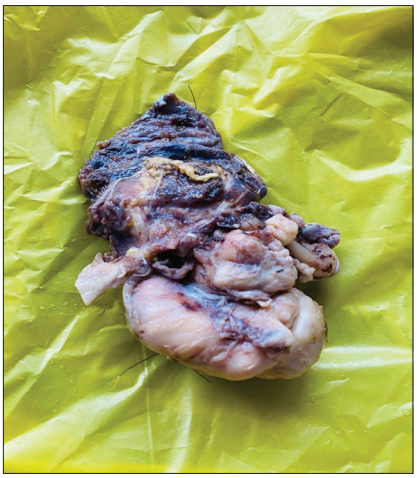

**Figure 1:** Gross appearance of spindle cell rhabdomyosarcoma showing irregular pale white to tan yellow soft-tissue mass with attached muscle at superior surface

showed a partially circumscribed tan yellow to gray-white tumor measuring 7.5 cm × 5 cm × 3.5 cm. Microscopic examination showed tumor cells with infiltrative margins arranged in fascicles and storiform patterns [Figures 2 and 3]. The cells were spindle-shaped with moderate eosinophilic cytoplasm and elongated hyperchromatic oval-to-spindle-shaped nuclei with increased mitosis, including atypical mitosis. The tumor cells were seen to infiltrate the temporalis muscle superiorly. No definite necrosis was identified. The attached bony tissue showed infiltration by tumor cells, suggesting a malignant spindle cell tumor – right temporal region infiltrating the zygoma and ramus of the mandible. Immunohistochemistry was carried out for further interpretation. The tumor cells were positive for SMA [Figure 4], desmin [Figure 5], MYOD1 and myogenin (focal) and negative for CK, CD34 and S100. The proliferation marker Ki67 showed 40–50% positivity [Figure 6]. Correlating the history, clinical, radiographic and histopathological findings, a final diagnosis of spindle cell RMS was made.

## DISCUSSION

An uncommon type of RMS, the spindle cell variant was initially grouped under embryonal RMS with a predilection for paratesticular and head-and-neck sites and associated with a comparatively favorable behavior in children. A subset of these tumors shows prominent hyaline sclerosis and pseudovascular growth pattern, suggesting morphologic overlap with sclerosing RMS. Both these tumors show recurrent MYOD1 gene mutations and hence have been classified as a single entity in the latest WHO classification.[4]

The spindle cell variant of embryonal RMS was first recognized as a rare entity in 1992 by the German-Italian

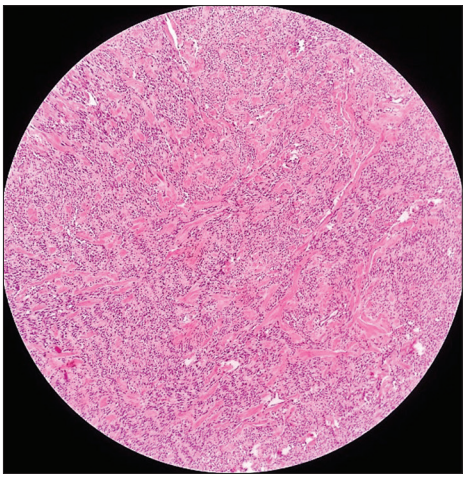

**Figure 2:** Low-power view – (H&E, ×100) – Tumor composed elongated spindle cells arranged in a fasciculated pattern

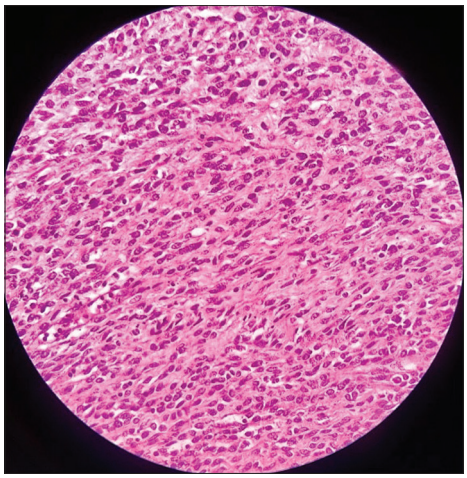

**Figure 3:** High-power view – (H&E, ×400) - Spindle-shaped cells with moderate eosinophilic cytoplasm and elongated hyperchromatic nuclei with increased mitosis

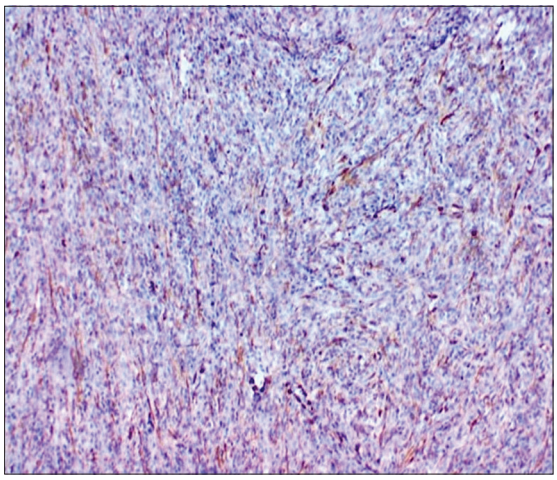

**Figure 4:** Positive immunostaining for smooth muscle actin

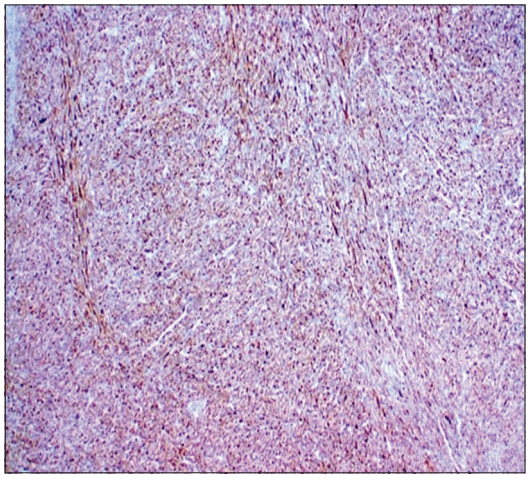

**Figure 5:** Positive immunostaining for desmin

Cooperative Soft Tissue Sarcoma Study.[5] It shows a male predilection with a tendency to occur in the paratesticular and head-and-neck regions and a low malignant potential. The sclerosing variant was first described in 2000 by Mentzel and Katenkamp.[1,5] The data regarding the clinical profile, histopathological findings and prognostic features of these tumors are scant as they have only been recently described as a separate entity. The tumor is seen in both children and adults and shows a wide age range from 0.3 to 79 years.[1] The tumor most commonly presents as a painless firm swelling.[6] The size varies from 1.5 to 35 cm.[7] The histological findings include the presence of small, round-to-spindle-shaped tumor cells with moderate nuclear pleomorphism. Scattered large rhabdomyoblasts with an eccentric nucleus and striated, eosinophilic cytoplasm may be seen.[8]

Depending on the amount of collagen interspersed between the tumor cells, the tumors have been earlier classified as collagen rich and collagen poor.[9] The tumor in the present case was predominantly collagen poor with cellular areas, and rhabdomyoblasts were not seen. These cellular spindle cell tumors may closely resemble leiomyosarcomas, MPNSTs with heterologous rhabdomyoblastic elements (malignant triton tumor) and fibrosarcomas. Similarly, in head-and-neck tumors, desmoplastic melanoma and spindle cell carcinoma should be considered first in the differentials in adults. The other differential diagnoses in a spindle cell tumor of the head and neck include inflammatory myofibroblastic tumor and synovial sarcoma.

Immunohistochemistry plays an important role in diagnosis. The morphology and immunoprofile of leiomyosarcomas very closely resemble RMS. Recognition of rhabdomyoblasts can point to the diagnosis of RMS, and additional immunomarkers such as myogenin and MyoD1, which are

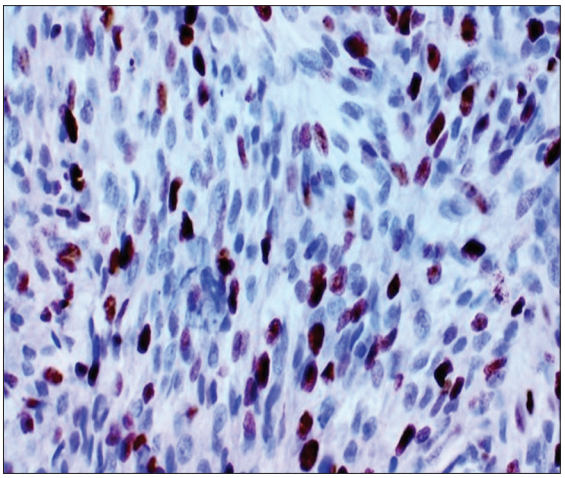

**Figure 6:** Immunoreactivity for Ki-67 – 40%–50% positivity

specific and sensitive for skeletal muscle differentiation.[10] MPNST and malignant triton tumor are seen in patients with neurofibromatosis and closely associated with nerve fiber. Tumor cells showing a perivascular accentuation and focal myxoid changes in MPNST are useful morphological clues in the distinction of both entities.[11] S100 positivity can also aid in the diagnosis.[6] RMS can show herringbone pattern and resemble fibrosarcomas morphologically; however, fibrosarcomas lack the markers of skeletal muscle differentiation can be helpful.

In the present case, owing to the proximity to the salivary gland and the small size of the core needle biopsy which showed a monomorphic population of oval-to-spindle cells, the possibility of myoepithelial origin was considered. The initial immunostains which were limited by the small size of the biopsy also suggested the same. However, the diagnosis was confirmed based on the morphology and immunohistochemical (IHC) stains on the resected specimens.

These tumors have been associated with various genetic alterations, most importantly, NCOA2 gene rearrangements in a subset of congenital cases and MYOD1 gene mutations with or without coexisting PIK3CA mutations in tumors occurring in older children or adults.[4]

In the adult population, these tumors have a more aggressive course. In the head-and-neck region, the complex anatomy and local aggressiveness of the tumors can make it difficult to get adequate free resected margins. The prognosis of the tumors depends on the size, resectability and stage of the tumor.[6]

## CONCLUSION

This case demonstrates the importance of considering the spindle cell variant of RMS in the differential diagnosis of spindle cell neoplasms of the head and neck. Diagnosis of spindle cell RMS indicates upfront chemotherapy. Hence, a careful correlation of the radiological, morphological and IHC findings helps in arriving at a diagnosis.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initial s will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

### Financial support and sponsorship

Nil.

### Conflicts of interest

There are no conflicts of interest.

## REFERENCES

1. Smith MH, Atherton D, Reith JD, Islam NM, Bhattacharyya I, Cohen DM. Rhabdomyosarcoma, spindle cell/sclerosing variant: A clinical and histopathological examination of this rare variant with three new cases from the oral cavity. Head Neck Pathol 2017;11:494-500.
2. Kaur P, Kaur A, Suri AK, Malik H. Spindle cell variant of embryonal rhabdomyosarcoma: A rare entity with diagnostic challenges. J Clin Diagn Res 2016;10:D17-8.
3. Parham DM, Barr FG. Embryonal rhabdomyosarcoma. In: Fletcher CD, Bridge JA, Hogendoorn PC, Mertens F, editors. WHO Classification of Tumours. Pathology and Genetics of Tumours of Soft Tissue and Bone. Lyon: IARC Press; 2013. p. 127-9.
4. Alaggio R, Zhang L, Sung YS, Huang SC, Chen CL, Bisogno G, *et al*. A molecular study of pediatric spindle and sclerosing rhabdomyosarcoma: Identification of novel and recurrent VGLL2-related fusions in infantile cases. Am J Surg Pathol 2016;40:224-35.
5. Cavazzana AO, Schmidt D, Ninfo V, Harms D, Tollot M, Carli M, *et al*. Spindle cell rhabdomyosarcoma. A prognostically favorable variant of rhabdomyosarcoma. Am J Surg Pathol 1992;16:229-35.
6. Carroll SJ, Nodit L. Spindle cell rhabdomyosarcoma: A brief diagnostic review and differential diagnosis. Arch Pathol Lab Med 2013;137:1155-8.
7. Nascimento AF, Fletcher CD. Spindle cell rhabdomyosarcoma in adults. Am J Surg Pathol 2005;29:1106-13.
8. Schildhaus HU, Lokka S, Fenner W, Küster J, Kühnle I, Heinmöller E. Spindle cell embryonal rhabdomyosarcoma of the prostate in an adult patient – Case report and review of clinicopathological features. Diagn Pathol 2016;11:56.
9. Newton WA Jr., Gehan EA, Webber BL, Marsden HB, van Unnik AJ, Hamoudi AB, *et al*. Classification of rhabdomyosarcomas and related sarcomas. Pathologic aspects and proposal for a new classification – An Intergroup Rhabdomyosarcoma Study. Cancer 1995;76:1073-85.
10. Iqbal HA, Anjum R, Naseem N. Rare variant of adult rhabdomyosarcoma presenting as a palatal swelling. Pak J Med Sci 2021;37:922-5.
11. Mentzel T, Kuhnen C. Spindle cell rhabdomyosarcoma in adults: Clinicopathological and immunohistochemical analysis of seven new cases. Virchows Arch 2006;449:554-60.