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Case Report

Adult spindle cell/ sclerosing rhabdomyosarcoma of the buccal maxillary gingiva: Unique entity, a rare case report

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ABSTRACT

Rhabdomyosarcoma is a malignant neoplasm of mesenchymal cells, which shows varying degrees of striated muscle cell differentiation. It predominantly occurs in children while rarely found in adults. Involvement of the oral cavity accounts for only 10-12% of all head and neck cases. Herewith, we report a rare case of oral spindle cell / sclerosing rhabdomyosarcoma in a 47-year-old male presented with a small mass involving the gingiva of right upper incisor. The mass was excised with a preoperative diagnosis of gingival epulis and subjected to histopathological and immunohistochemical examination which confirmed it to be spindle cell / sclerosing rhabdomyosarcoma. Data regarding its clinical course, genetic abnormalities and prognosis as a combined subtype is scant.

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1. Introduction

Rhabdomyosarcoma (RMS) is the most common childhood soft tissue sarcoma in children under 15 years of age and accounts for an estimated 4.5% of all childhood cancers. It is rare in person older than 45 year and accounts for an estimated 2- 5 % of all adult sarcomas.¹ According to WHO (2013), It is subtyped into an embryonal RMS, an alveolar RMS; a pleomorphic RMS and more recently, spindle cell and sclerosing RMS (S-ScRMS).² The spindle cell variant was originally proposed in 1992 by Cavazzana, and was initially considered as a variant of an embryonal RMS^{2,3} while sclerosing variant was first described in 2000 by Mentzel & Katenkamp and was identified as another distinctive variant of a RMS.^{2,4} Several studies had described a relationship between spindle cell and sclerosing RMS that lately have emerged together as a distinct subtype of a RMS.^{5–8} Spindle cell/ sclerosing rhabdomyosarcoma is

an uncommon subtype, accounting for 5-10% of all cases of RMS. It affects both children and adults, with a male to female ratio of up to 6:1.² Due to the rarity of the cases in the literature and recent reclassification of S-ScRMS, every case report is valuable to the literature. Hence, the present case report holds a value.

2. Case Report

A 47-year-old male presented with a complaint of swelling over gingiva of right upper incisor since one month. The patient had history of oral injury 6 month back. Clinical examination revealed 1x1cm, soft, abscess like, non-tender, non-fluctuating mass on maxillary gingiva. X-ray demonstrated no bony changes. As the clinical findings suggested a tumorous lesion, a provisional diagnosis of gingival epulis was made. An incisional biopsy under local anesthesia was performed and sent for histopathological examination.

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On microscopy, it revealed well defined tumor in superepithelium which was raising and stretching the epithelium. Tumor was composed of interlacing bundles of uniform oval to short spindle cells separated by thin septae. The cells showed mild to moderate pleomorphic, fusiform nuclei, dispersed chromatin, definite visible nucleoli and ill-defined eosinophilic cytoplasm. Many mitotic figures were seen. No necrosis. Opinion was given as spindle cell tumor (Figure 1a).

Immunohistochemistry revealed cells were positive for vimentin and strongly positive for desmin (Figure 1b). Hence suggested mesenchymal origin of tumor. Further confirmation was made by application of markers like MyoD1, CK, Ki-67. Results were positive in nuclei for MyoD1 in majority of tumor cells (Figure 1c), weak positive in aberrant pattern for CytoKeratin (Figure 1d) and 30% for Ki-67. The tumor cells were negative for SMA, Myogenin, EMA, S-100, CD34, P-63, HMB-45 & Melan-A.

Histopathology and immunohistochemistry findings were consistent with diagnosis of spindle cell/ sclerosing rhabdomyosarcoma (FNCLCC Grade 1).

After Surgery, the patient is well & there is no evidence of recurrence till now.

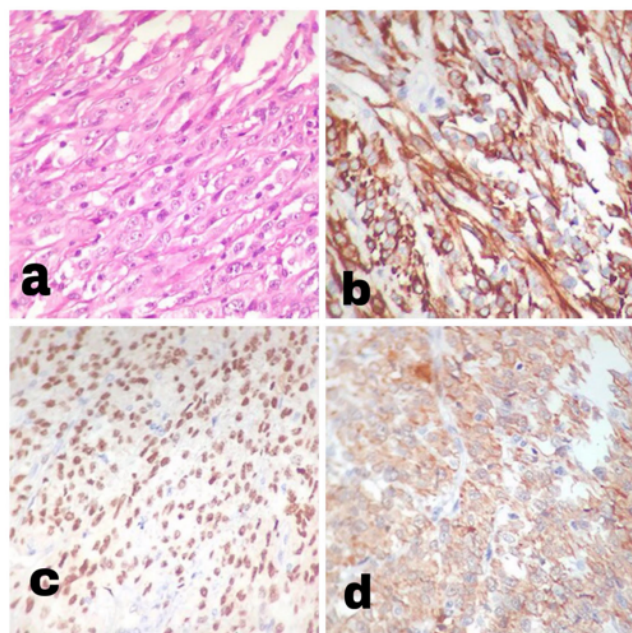


Fig. 1: (a): Interlacing bundles of uniform oval to short spindle cells separated by thin septae. (On H & E, 40x); (b): Immunohistochemistry revealed strong positivity for Desmin (40x); (c): Immunohistochemistry revealed nuclear positivity for MyoD1 in majority of tumor cells. (40x); (d): Immunohistochemistry revealed cells positive in aberrant pattern for CytoKeratin. (40x)

3. Discussion

Rhabdomyosarcoma (RMS) is malignant neoplasm of mesenchymal cells. It is most common soft tissue sarcoma in children. On the contrary, it is very rare to see RMS in adults.¹⁵ Spindle cell/sclerosing rhabdomyosarcoma is a rare skeletal-muscle tumor with distinctive clinicopathologic characteristics and is designated in the WHO classification (2013) as a separate fourth broad category under RMS.² Spindle cell RMS (SRMS) was first reported by Cavazzana et al³, Lesion with spindle cell morphology are commonly identified in the paratesticular region of pediatric patients,^{2,16} while in adults >50% of cases affect the deep soft tissues in the head and neck. Lesion with sclerosing morphology in both age groups are most common in the limbs.²

Histologically these tumours are characterized by predominant population of spindle neoplastic cells. Nuclear atypia, hyperchromasia and mitotic figures are common. Occasionally RMS with spindle cell morphology may show stromal hyalinization with tumor cells imparting a pseudovascular appearance, and characterizing the sclerosing variant of RMS.^{2,3}

We described a case of oral spindle/sclerosing rhabdomyosarcoma in a 47-year-old male. Comparative studies of similar cases^{9–14} in literature has been summarized, please refer to Table 1.

SRMS provides a diagnostic challenge due to its similarity to other spindle cell neoplasms. Therefore, immunohistochemistry plays a pivotal role in the diagnosis of SRMS/ScRMS. The immunohistochemistry markers to rule out other differential diagnosis of spindle cell tumours is summarized (refer to Table 2).

According to WHO (2013) spindle cell RMS shows diffuse expression of desmin and positivity for SMA and myogenin. Sclerosing RMS shows limited expression of desmin and myogenin, but strongly positive for MyoD1.² The diffuse and strong positivity for MyoD1 supports our diagnosis, as Myo-D1 has been shown to be a more sensitive marker for sclerosing variant of RMS.^{10,13}

Patients with S-ScRMS demonstrate heterogenic genetic alterations that may have particular importance on prognosis.¹⁰ Recent studies focusing on SRMS-ScRMS shows a variable prognosis based on their age at diagnosis & genetic abnormalities. SRMS in infants exhibit recurrent NCOA2 and VGLL2 related fusions and are associated with a favorable outcome & long-term survival^{10,17} In contrast, SRMS-ScRMS with MYOD1 mutations follow an aggressive clinical behavior and poor prognosis, irrespective of the patient's age.^{10,17,18}

Most RMS are treated with conventional surgery, chemotherapy and radiotherapy.¹⁰ Despite efforts at various treatment techniques, prognosis of S-ScRMS in adults is significantly worse, with a rate of recurrence and metastasis of approximately 40-50%.²

Table 1: Comparative studies of cases of rhabdomyosarcoma in literature

Authors	Number of cases	Year of Study	Age / Sex of the Patient	Location of swelling	Provisional diagnosis before Biopsy	Diagnosis after Biopsy	IHC Marker	Definite Diagnosis
Mina et al. ⁹	1	2018	32y/ F	Maxillary gingiva	Nonspecific inflammatory lesion	Spindle cell sarcoma	Positive for Desmin Myogenin MyoD1 Ki 67 >10% i) Positive for desmin, MSA vimentin & Myogenin ii) Not available iii) positivity to desmin, smooth muscle actin, myogenin, and Myo-D1	Pleomorphic RMS due to less spindle-shaped cells with more remarkable nuclear pleomorphism and bizarre tumor cells. i) Spindle cell variant of RMS ii) Recurrent RMS iii) Spindle cell/ Sclerosing RMS (S-ScRMS)
Smith MH et al. ¹⁰	3	2017	i)24y/M ii)39y/M iii)28y/M	i)mass of the right hard palate/posterior maxillary alveolar gingiva ii) left buccal mucosa. iii) swelling in left mandibular area.	i)soft tissue tumor ii) K/C/O spindle cell variant of RMS iii)soft tissue tumor	i)Spindle cell sarcoma ii)Recurrent RMS iii)Spindle cell variant with sclerosis		
Chi et al. ¹¹	1	2007	33y/F	anterior maxillary gingiva.	Soft tissue tumor	Spindle cell tumor	positive for desmin, myogenin, (MyoD1).	Embryonal RMS
Joy T et al. ¹²	1	2018	52 y/M	mandibular gingiva	Soft tissue tumor	spindle cell variant of squamous cell carcinoma	strongly & diffusely positive for desmin, diffusely & focally positive for MyoD1 and myogenin Vimentin (+) Increased Ki-67 (>60%), diffusely positive for desmin , myogenin, focally positive for CD99 and WT-1	Spindle cell RMS
Robinson JC et al. ¹³	1	2012	40y/ M	Mandibular gingiva and buccal mucosa	Nonspecific inflammatory lesion	high-grade pleomorphic undifferentiated sarcoma		Sclerosing RMS
Hartmann, S et al. ¹⁴	1	2014	41y/ M	Below tongue	soft tissue tumor	myofibrosarcoma	Positive for Desmin, Myogenin, Myo D1, Negative for Caldesmon	Spindle cell RMS

Abbreviations: RMS- Rhabdomyosarcoma, SMA- smooth muscle actin, MyoD1 – Myogenic differentiation antigen 1

Table 2: Differential Diagnosis of Spindle Cell Tumors with Immunohistochemistry Markers

Tumor	Desmin	Myogenin	SMA	keratin	S100 protein	CD34	Other Markers
Synovial Sarcoma	-	-	-	+++	+/-	-	CD 56, Calretinin
Leiomyosarcoma	+/-	-	+++	-	-	-	Caldesmon (+)
PEComa	+/-	-	+++	-	-	-	HMB-45, Melan- A
Spindle Cell Melanoma	-	-	-	-	+	-	SOX 10 (+)
Malignant peripheral Nerve Sheath Tumor	-	-	-	-	+++	+/-	Desmin expression seen in rhabdomyoblastic elements (malignant triton tumor)
Neurofibroma	-	-	-	-	+++	+++	EMA/Claudin (+)
Nodular Fasciitis	-	-	+++	-	-	-	Rare focal desmin (+)
Spindle cell RMS	Diffuse	+	+/-	-	-	-	Myo D1 (+), Strong Myo D1 positivity in Sclerosing RMS
Spindle cell lipoma	-	-	-	-	-	+++	Loss of nuclear Rb protein expression
Perineurioma	-	-	-	-	-	+/-	EMA (+), Claudin-1 (+)

Abbreviation s: SMA, smooth muscle actin; + positive, +++ strongly positive, - Negative, +/- occasionally positive

4. Conclusion

Oral rhabdomyosarcoma can develop insidiously and due to variable clinical presentations and histopathological appearances, early lesions may be mistaken for inflammatory, benign neoplastic or infectious processes. Furthermore, due to the rarity of the cases in adults & recent reclassification of S-ScRMS, misdiagnosis is common. Hence, immunohistochemistry plays an important role in identification of this lesion.

5. Conflict of Interest

The authors declare that there are no conflicts of interest in this paper.

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None.

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