Article published online: 2024-11-06

THIEME OPEN ACCESS

Case Report

Spindle Cell Variant of Rhabdomyosarcoma: An Aggressive Clinical Case of the Oral Cavity

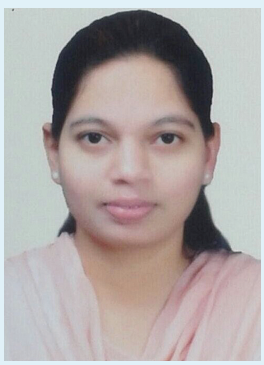
Mayadevi Poojari[1] Irappa Madabhavi[2,3,4]

[1] Department of Radiation Oncology, Oncolife Cancer Centre, Talegaon Dabhade, Pune, Maharashtra, India
[2] Department of Medical and Pediatric Oncology and Hematology, J. N. Medical College and Research Institute, Belgaum, Karnataka, India
[3] Department of Medical and Pediatric Oncology and Hematology, Kerudi Cancer Hospital, Bagalkot, Karnataka, India
[4] Nanjappa Hospital, Davangere, Karnataka, India

Address for correspondence Mayadevi Poojari, MBBS, DMRT, DNB, Department of Radiation Oncology, Mahatma Gandhi Cancer Hospital, Miraj 416410, Maharashtra, India (e-mail: mayapoojari29@gmail.com).

South Asian J Cancer

Abstract



Mayadevi Poojari

Rhabdomyosarcoma (RMS) is a malignant, soft tissue neoplasm consisting of cells derived from the primitive mesenchyme that exhibit a profound tendency to myogenesis. Historically, the spindle cell and sclerosing variants were subcategorized under embryonal type RMS. Spindle cell/sclerosing RMS (S-ScRMS) was recently recognized in 2013 by the World Health Organization as a stand-alone entity. Current data support that certain S-ScRMS cases have a more aggressive clinical course with a reduction of long-term survival, and those found in the head and neck region often exhibit extensive local recurrence. Here, we highlight an aggressive clinical course of S-ScRMS by presenting a case of a 7-year-old male child who presented with complaints of swelling in left cheek since 15 to 20 days. It was associated with pain. He was diagnosed as S-ScRMS and received chemotherapy but the disease was aggressive and progressed on chemotherapy. He received radiation but still the disease was inoperable, hence was kept on supportive care.

Keywords

- rhabdomyosarcoma
- oral cavity
- spindle cell
- soft tissue neoplasm
- tomotherapy

Introduction

Rhabdomyosarcoma (RMS) is a malignant, soft tissue neoplasm consisting of cells derived from the primitive mesenchyme that exhibit a profound tendency to myogenesis. It is classified into four broad categories based on the most recent World Health Organization (WHO) classification: (1) alveolar RMS, (2) embryonal RMS (ERMS), (3) pleomorphic RMS, (4) spindle cell/sclerosing RMS (S-ScRMS).[1] The spindle cell variant was originally proposed in 1992 by Cavazzana et al,[2] while the sclerosing variant was first described in 2000 by Mentzel and Katenkamp[6]. Both variations

DOI https://doi.org/10.1055/s-0044-1791946 ISSN 2278-330X

How to cite this article: Poojari M, Madabhavi I. Spindle Cell Variant of Rhabdomyosarcoma: An Aggressive Clinical Case of the Oral Cavity. South Asian J Cancer 2024;00(00):00–00.

Thieme Medical and Scientific Publishers Pvt. Ltd., A-12, 2nd Floor, Sector 2, Noida-201301 UP, India

previously were subcategorized under the classification of ERMS; however, because the spindle cell and sclerosing variants share similar clinical, histopathological, and genetic features that are separate from the remaining three categories, they were classified as a stand-alone variant in the most recent (2013) edition of the WHO classification system.[1]

S-ScRMS comprises 5 to 13% of the cases of RMS and can be seen in both children and adults.[1] RMS is most common in the head and neck but tends to have a poorer prognosis and more aggressive clinical course when found in these locations due to proximity to vital structures. Because of the recent reclassification of S-ScRMS, minimal data on prognosis and clinical course of this rare variant are available. Here, we present an aggressive clinical course in a 7-year-old male child who came with a complaint of left cheek swelling for 15 to 20 days. He was diagnosed as S-ScRMS. He received chemotherapy but the disease was aggressive and progressed on chemotherapy. He received chemoradiation but still the disease was inoperable, hence was kept on supportive care.

Case Report

A 7-year-old male child presented with chief complaints of left cheek swelling since 15 to 20 days. There was no history of fever and weight loss. The patient had consulted at other hospitals and was evaluated with positron emission tomography computed tomography, which showed FDG AVID soft tissue mass with few cystic components noted involving left masticator space and retromolar trigone, extending into high infratemporal fossa measuring $\sim 6.6 \times 6.5 \times 4.8$ cm with SUVmax of 12.49. There was extension into pterygomaxillary fissure and pterygopalatine fossa. There was cortical erosion of the upper alveolar process, left ramus of mandible, and lateral pterygoid plate. There was scalloping of posterior wall of maxillary antrum seen, and also widening of the temporomandibular joint space noted. Few enlarged left cervical level IB and II nodes with low-grade FDG uptake, with maintained fatty hila, were seen—indeterminate. No e/o metabolically active disease elsewhere was noted. On biopsy and immunohistochemistry, it showed as MyoD1-positive; negative for Desmin, Myogenin, SMA, and S100p; was diagnosed as S-ScRMS.

He received one cycle of VAC chemotherapy: Inj Vincristine 1.5 mg/m^2 on Day 1, 8, 15; Inj Cyclophosphamide 1.2 g/m^2 on Day 1; Inj Actinomycin D 0.045 mg/kg on Day 1 at another hospital. As there was rapid increase in the size of tumor, as per parents, they came for a second opinion at our center. On examination, there was intra-orally bulky mass filling in whole oral cavity and extra-orally huge swelling covering the entire left side of cheek. Skin of the left cheek was tense and shiny (►Fig. 1). It was associated with pain. The patient had compromised nutrition so Ryle's tube was inserted for alternate feeding procedure. The case was discussed in Tumor Board Meeting and decided to continue chemotherapy and to be reassessed by a surgical oncologist.

He received one cycle of VAC protocol chemotherapy—Inj Vincristine 1.5 mg/m^2 on Day 1, 8, 15; Inj Cyclophosphamide 1.2 g/m^2 on Day 1; Inj Actinomycin D 0.045 mg/kg on Day 1. After one cycle of chemotherapy, on clinical examination there was progression of disease, and it was inoperable. He was referred to Radiation Oncology department for opinion on radiation therapy. He was treated with external beam radiotherapy at a dose of 50.4 Gy in 28 fractions for 5 days a week at 1.8 Gy per fraction using helical image-guided radiotherapy—tomotherapy. During the treatment, after 5 fractions there was reduction in size of the disease but on 6th fraction next week again the disease was back to its original size. Postradiation treatment he was assessed by a surgical oncologist and the disease was still inoperable clinically as well as on magnetic resonance imaging scan. Hence, he was advised supportive care.

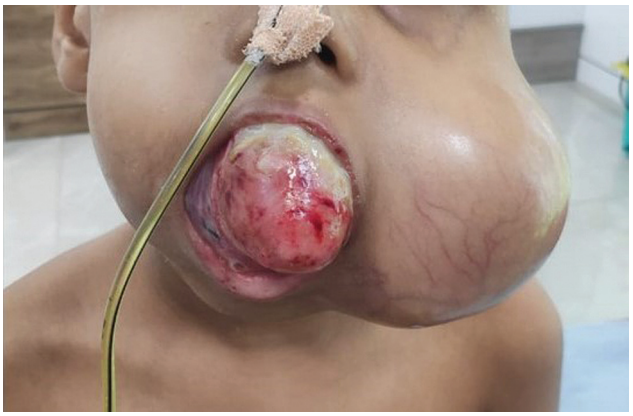


Fig. 1 Clinical picture pretreatment showing intra-orally bulky mass filling in whole oral cavity and extra-orally swelling in left cheek with tense and shiny skin.

Discussion

Approximately 7% of all malignancies occurring in patients under the age of 20 represent sarcomas, and of these sarcomas, RMS is the most prevalent, comprising 40 to 50%. The spindle cell variant has a predilection for the paratesticular region, followed by the head and neck, whereas the sclerosing variant mostly involves extremities, then the head and neck.[3] As with all types of RMS, S-ScRMS exhibits a male predilection.[2,3] Both children and adults are affected with this variant, with a range of 0.3 to 79 years.[3] This tumor is usually fast-growing and infiltrative, and often appears as an enlarging, painless mass.

Microscopically, S-ScRMS typically reveals a proliferation of spindle to ovoid cells with long and intersecting fascicles. An important diagnostic clue is the presence of rhabdomyoblastic features, including "strap cells" with cross-striations or ganglion cell-like rhabdomyoblasts, which demonstrate abundant granular, eosinophilic cytoplasm with eccentrically placed nuclei. Immunohistochemical stains demonstrate distinct positivity for desmin and vimentin. Variable expressivity of myogenin (Myf-4), Myo-D1 (Myf-3), myoglobin, smooth muscle actin (SMA), and muscle specific actin (MSA) has been reported. MDM2, Bcl-2, p53, calponin, and CD56 rarely have been expressed in published cases.[3] RMS is negative for S-100, epithelial membrane antigen, cytokeratins, glial fibrillary acidic

protein, caldesmon, vascular markers, neuroendocrine markers, and melanoma markers. Immunohistochemistry is most helpful in differentiating RMS from other malignancies.

Heterogenic genetic alterations are demonstrated in patients with S-ScRMS which have particular importance on prognosis. Tumors diagnosed in infancy frequently exhibit recurrent gene fusions implicating VGLL2, SRF, TEAD1, or NCOA2 and appear to be associated with outcomes that are more favorable.[4] Conversely, a subset of S-ScRMS demonstrates mutations in MYOD1 (some with accompanying PIK3CA mutations). These latter mutations exhibit aggressive clinical courses with higher mortality rates unrelated to the age of the patient. Molecular evaluation of MYOD1 mutations has been suggested for risk stratifications in patients with S-ScRMS as it is associated with poorer prognosis. In our case, we could not do genetic study due to financial reasons.

Most RMSs are treated with conventional surgery, chemotherapy, and radiotherapy. Chemotherapeutic agents typically consist of actinomycin D, doxorubicin, ifosfamide, cyclophosphamide, etoposide, or vincristine. Despite efforts at various treatment techniques, the prognosis for RMS, especially S-ScRMS, remains poor.[5]

Conclusion

Due to the recent reclassification of S-ScRMS and rarity of cases in the literature, case reports and series are especially valuable to the literature. Here, we are reporting a case of a 7-year-old male child with aggressive clinical course of S-ScRMS who progressed on chemotherapy. He received chemoradiation but still the disease was inoperable and was kept on supportive care. Hence, additional case reports of this RMS variant may help us not only to better understand the clinical behavior and also the diagnostic challenges, but also to develop more targeted treatment therapies to improve long-term survival and quality of life.

Conflicts of Interest

None declared.

References

1 Parham DM, Barr FG, Montgomery E, Nascimento AF. Skeletal muscle tumors. In: Fletcher CDM, Bridge JA, Hogendoorn PCW, Mertens F, eds. WHO Classification of Tumours of Soft Tissue and Bone. 4th ed. Lyon: IARC Press; 2013:123–135

2 Cavazzana AO, Schmidt D, Ninfo V, et al. Spindle cell rhabdomyosarcoma. A prognostically favorable variant of rhabdomyosarcoma. Am J Surg Pathol 1992;16(03):229–235

3 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(04):494–500

4 Alaggio R, Zhang L, Sung YS, 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(02):224–235

5 Turner JH, Richmon JD. Head and neck rhabdomyosarcoma: a critical analysis of population-based incidence and survival data. Otolaryngol Head Neck Surg 2011;145(06):967–973

6 Mentzel T, Katenkamp D. Sclerosing, pseudovascular rhabdomyosarcoma in adults. Clinicopathological and immunohistochemical analysis of three cases. Virchows Arch 2000;436(04):305–11

