A Rare Case : Spindle Cell Rhabdomyosarcoma of Mandible

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Summary

Rhabdomyosarcoma is the commonest soft tissue sarcoma in 15-45 years.[1] We report a case of aggressive rhabdomyosarcoma of mandible (spindle cell variant) in a 27 year female patient, describing the clinicopathological findings and radiological findings of the patient and its management in our centre.

Keywords: Oral spindle cell variant rhabdomyosarcoma, mesenchymal tumour

Introduction

Rhabdomyosarcoma is aggressive malignancy and commonest malignant soft tissue sarcoma of children and young adults.[1] RMS primarily involves the head and neck. It is soft tissue malignancy which arises from mesenchymal tissue. Its incidence is more in within 20 years of age.[1] The present incidence of RMS in oral cavity is 0.041 cases per 100,000 people.[2] This is a case of a 27-year female with spindle cell rhabdomyosarcoma wherein clinico-pathological and clinicoradiological correlation led to the diagnosis.

Case Report

A 27-year-old female with the chief complaints of progressive swelling and growth over the mandibular region presented to us with a rapid increase in size over the previous four weeks. She also had a history of significant loss of weight. Inspection showed protrusion of lower lip with exophytic mass resting over it. It measured 6x5x3cm. on palpation localized tenderness was present. Skin overlying the chin was involved (Figure 1). On intraoral inspection, the ulceroproliferative mass was noted in anterior part of oral cavity, involving the anterior part of mandible, gingivobuccal sulcus and buccal mucosa.

Contrast Enhanced Computerized Tomography-Paranasal sinuses, Neck, Thorax revealed a 7.7x7x7.3 cm lesion arising from the central arch along with its erosion. Lesion involved bilateral body of mandible, mandibular canal, lower buccal space and gingivobuccal space. Lesion involved angle of mouth and overlying skin and shows exophytic growth with soft tissue defect. It involved bilateral gingivolabial and bilateral gingivolingual. The lesion extended into floor of mouth and involved mylohyoid muscle, ventral surface of tongue, and intrinsic component of tongue. There is reduced distensibility of adjacent buccal space, with bilateral level IB and II necrotic nodes (Figure 2).

A biopsy was performed and showed a malignant spindle cell tumour with moderate pleomorphism with >20 mitosis /hpf.

Figure 1: Preoperative picture of case

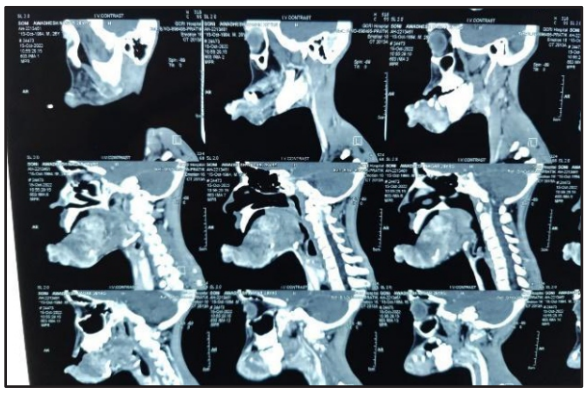


Figure 2: Preoperative CECT of the case

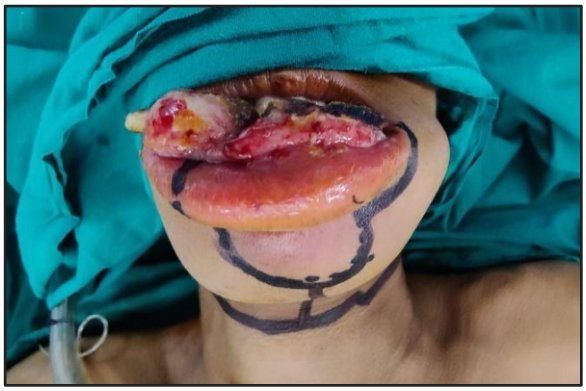


Figure 3: Intra operative picture of the case

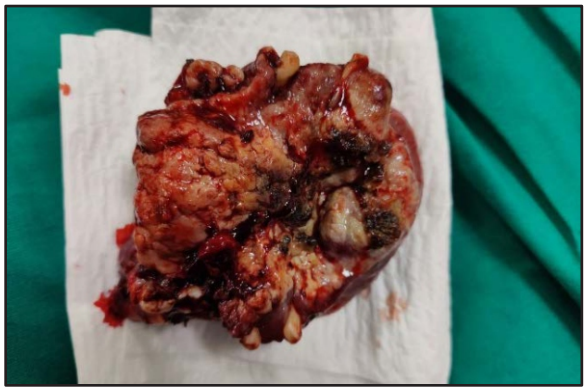
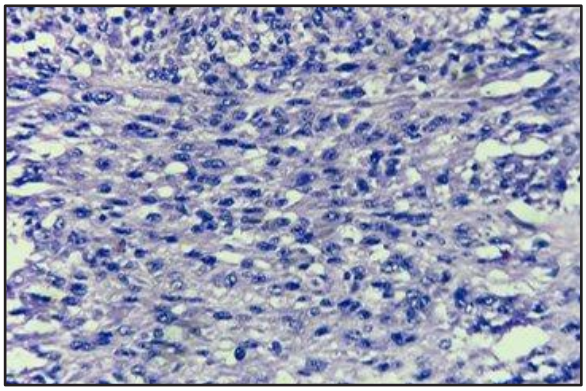


Figure 4: Wide local excision of the lesion



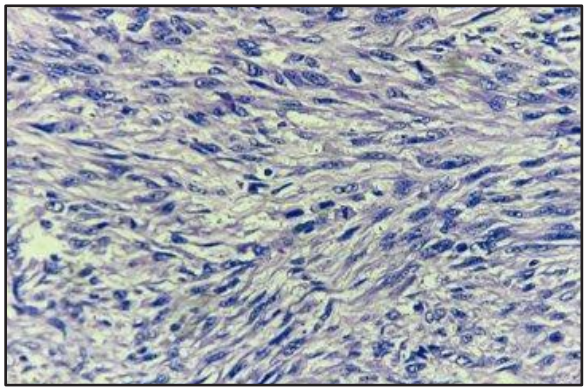


Figure 5 a, b: The haematoxylin and eosin sections show proliferation of predominantly spindle cells with moderately pleomorphic nuclei, distinct nucleoli and eosinophilic cytoplasm. 40x objective

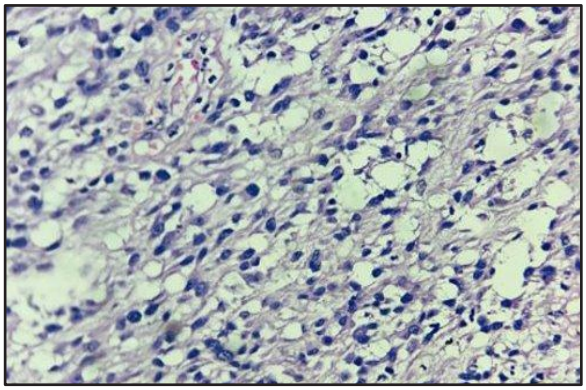


Figure 5c: Scattered rhabdoid cells are seen with eccentrically pushed hyperchromatic nuclei having amphophilic cytoplasm. 40x

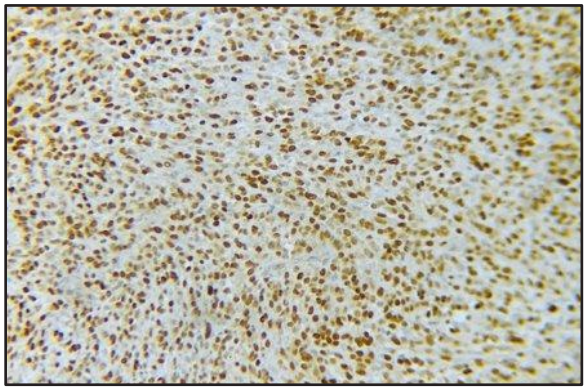


Figure 5d: MyoD1 immunostain shows diffuse nuclear positivity in all the tumour cells, 40x

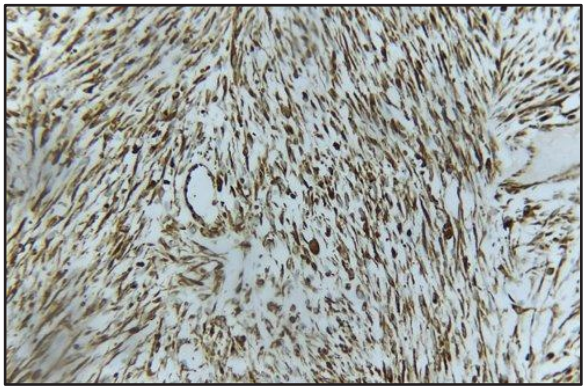


Figure 5e: Vimentin immunostain shows diffuse cytoplasmic positivity in all the tumour cells, 40x

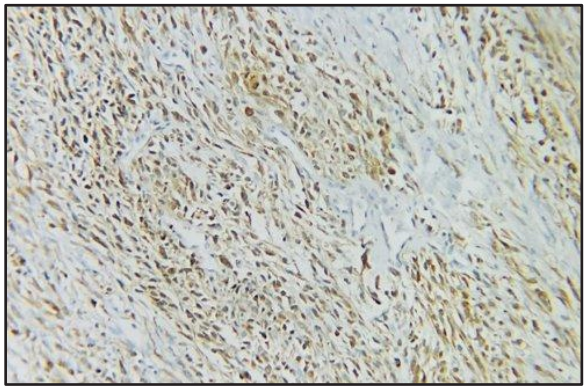


Figure 5f: EMA (epithelial membrane antigen) immunostain shows diffuse cytoplasmic positivity in all the tumor cells, 40x

Immunohistochemistry was performed and it was positive.

Patient was planned for neoadjuvant chemotherapy, and was started on VACM regimen. After taking one cycle, she developed severe vomiting, generalized weakness and diarrhoea. Since she was unable to tolerate neoadjuvant chemotherapy, she was planned for salvage surgery. Wide local excision of central arch with segmental mandibulectomy and bilateral modified neck dissection type II was done. Reconstruction with a bilobed pectoralis major myocutaneous flap was performed (Figure 3). Later, patient developed skin necrosis, hence debridement followed by deltopectoral flap cover was done (Figure 4). Final histopathology showed high grade spindle cell sarcoma – spindle cell Rhabdomyosarcoma of size 8x6x4 cm histologic grade 3, mitotic rate 4-5/10 hpf, 20% necrosis due to effect of chemotherapy, multifocal tumour, no lymphovascular invasion and perineural invasion and anterior cutaneous margin was 0.3cm away from tumour rest margins greater than 0.5cm and free of tumour. pT4aN0. Patient was subsequently referred for completion of chemotherapy after removal of sutures. However, before chemotherapy could be started, she developed a nodule over right cheek. Core biopsy from the same showed recurrent spindle cell rhabdomyosarcoma. Disease free survival was 2 months. Patient was subsequently sent for palliative systemic therapy. Final IHC suggestive of spindle cell rhabdomyosarcoma with AE1 weak positive and MyoD1 positive, EMA focal positive and occasional positive (Figure 5).

Discussion

Spindle cell rhabdomyosarcoma (RMS) is a rare type of RMS. It can affect any age group, more common children and in male with a ratio of 6:1. Histological types of RMS are: alveolar,embryonal, and pleomorphic.[3] It's the commonest soft-tissue sarcomas of childhoodand embryonalhas the highest incidence.[4] It accounts only 5-10% of all solid tumours cases and 4-8% of all malignancies.

Spindle cell, rare type of RMS, was initially grouped under embryonal RMS which was more common in head-and-neck region. Some of the embryonal tumours showed hyaline sclerosis and pseudovascular growth pattern, as found in sclerosing RMS. Both embryonal and spindle cell tumours demonstrate recurrent mutations of the MYOD1 gene and are therefore classified as a single type in WHO classification.[5] The Spindle cell variant of embryonal RMS was first recognized as a rare one in 1992 by German-Italian Cooperative STS Study.[6] Patients comes with painless firm swelling most commonly.[7] The size of tumor may range from as small as 1.5 cm to around 35 cms.[8] Histologically, RMS shows small, round-to-spindle-shaped cells having moderate nuclear pleomorphism. Large rhabdomyoblasts having an eccentric nucleus and eosinophilic cytoplasm.[9]

Previously classification was based on collagen density in between the tumor cells and was collagen rich or collagen poor type.[10] Cellular type spindle-cell tumours may be similar to leiomyosarcomas, MPNSTs and fibrosarcomas. Similarly, desmoplastic melanoma and spindle cell carcinoma are the first differentials in adults, inflammatory myofibroblastic tumour and synovial sarcoma are other differential diagnoses.

Immunohistochemistry helps to confirm the diagnosis. Morphology and immunoprofile of leiomyosarcomas and RMS are indistinguishable. Presence of rhabdomyoblasts confirms RMS, other than this IHC markers such as desmin, myogenin and MyoD1, indicative of skeletal muscle differentiation helps to conclude diagnosis.[11] Spindle cell variant of RMS is negative for S100 .[7]

These tumours have a more aggressive course in adults. Their prognosis relies on the size, resectability and staging of the tumour.[7] In our case also, complete resection was done with histologically free margins but the patient came with recurrence within two months of resection, showing the aggressiveness of the disease.

Conclusion

This case demonstrates that spindle cell rhabdomyosarcoma is a rare but aggressive malignancy and it requires upfront chemotherapy and completion surgery and additional nutrition therapy is essential in order to tolerate treatment.

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