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Case Report

An atypical case-spindle cell variant of embryonal rhabdomyosarcoma of mandible

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ABSTRACT

Rhabdomyosarcoma (RMS) is an aggressive malignancy and commonest malignant soft tissue Sarcoma predominantly seen in children and young adults**.** This study presents a case of a spindle cell embryonal type RMS involving the mandible of a 17-year-young male with a localized disease who underwent a R0 resection with tumor free margins followed by adjuvant chemotherapy, now with recurrence and metastatic disease, currently on palliative therapy. This report emphasizes the significance of early diagnosis and accurate immunohistochemical analysis in achieving successful treatment outcomes for RMS.

Keywords: Spindle cell embryonal type rhabdomyosarcoma, Mandible, Immunohistochemical analysis

INTRODUCTION

Rhabdomyosarcoma (RMS) is a malignant neoplasm of striated muscle. RMS primarily involves the head and neck. It is a soft tissue malignancy which arises from mesenchymal tissue. It was classified by Horn and Enterline based on clinical and histological features into 4 types-pleomorphic, alveolar, embryonal, and botryoid.[2] Embryonal RMS shows a distinct predilection for visceral sites, such as the genitourinary tract, followed by the head and neck, and extremities.[3] In the recent times, other variations have been identified, including spindle cell and sclerosing RMS.

The spindle cell variant of embryonal RMS was first recognized as a rare one in 1992 by German-Italian Coopera.[4,5] Compared with other subtypes, the spindle cell variant in children is associated with a favorable outcome. Even though embryonal RMS is common, the spindle cell variant is considered rare (only 3% of all RMS cases in intergroup RMS study).[6]

CASE REPORT

A 17 year old young male presented with a history of swelling in the right side of the jaw for the past 4 months, which was rapidly growing in size and associated with intermittent pricking type of pain. Biopsy from the right-side mass done in his home country was suggestive of malignant spindle cell neoplasm.

On clinical examination a huge nodular mass was noted involving the right side of the mandible crossing the midline with widening of the lower alveolus (Figure 1).

A contrast enhanced computed tomography (CECT) neck was done which showed large exophytic heterogeneously enhancing soft tissue density mass with necrotic areas involving mandible (from angle of right hemi mandible to body of left hemimanible). The mass measured 11.5 (AP)×9.7(T)×11 (CC) cm. The mass was seen involving the right masticator space, gingivo-buccal sulcus, retro-molar trigone and bilateral submandibular space. Posteriorly, abutting the right parotid gland with

indistinct fat planes. Medially the mass was extending intra orally and abutting the bilateral mylohyoid, hyoglossus and digastric muscles with indistinct fat planes. There was an extensive destruction of mandible with involvement of inferior alveolar process. Bilateral orbicularis oris and Depressor anguli oris muscles were seen separately from the mass. Multiple prominent to enlarged bilateral cervical nodes were noted (Figures 2).

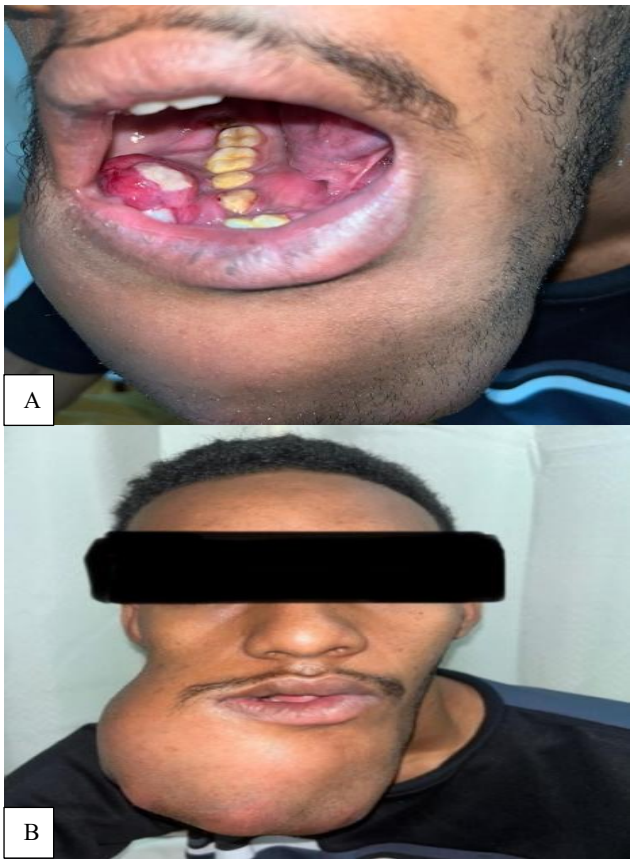
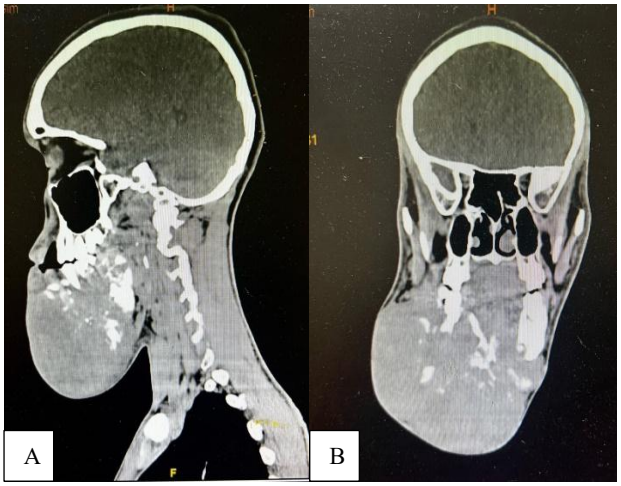


Figure 1 (A and B): On clinical examination a huge nodular mass.



Figures 2 (A and B): CECT showing a large exophytic heterogeneously enhancing soft tissue density mass with necrotic areas involving mandible.

Furthermore, an ultrasound guided trucut biopsy from the right side mass was done which showed spindle cells arranged in the form of cords and singly scattered cells having hyperchromatic nuclei with conspicuous nucleoli in some of them displaying mild to moderate atypia.

Immunostains were positive for pancytokeratin, vimentin, MyoD1, Desmin, scattered cells were positive for SMA and MSA. Hence histopathological findings and immunohistochemistry pattern was suggestive of RMS, spindle cell type. The differential diagnosis considered were leiomyosarcoma, Ewing sarcoma which on immunohistochemical staining, turned out to be negative.

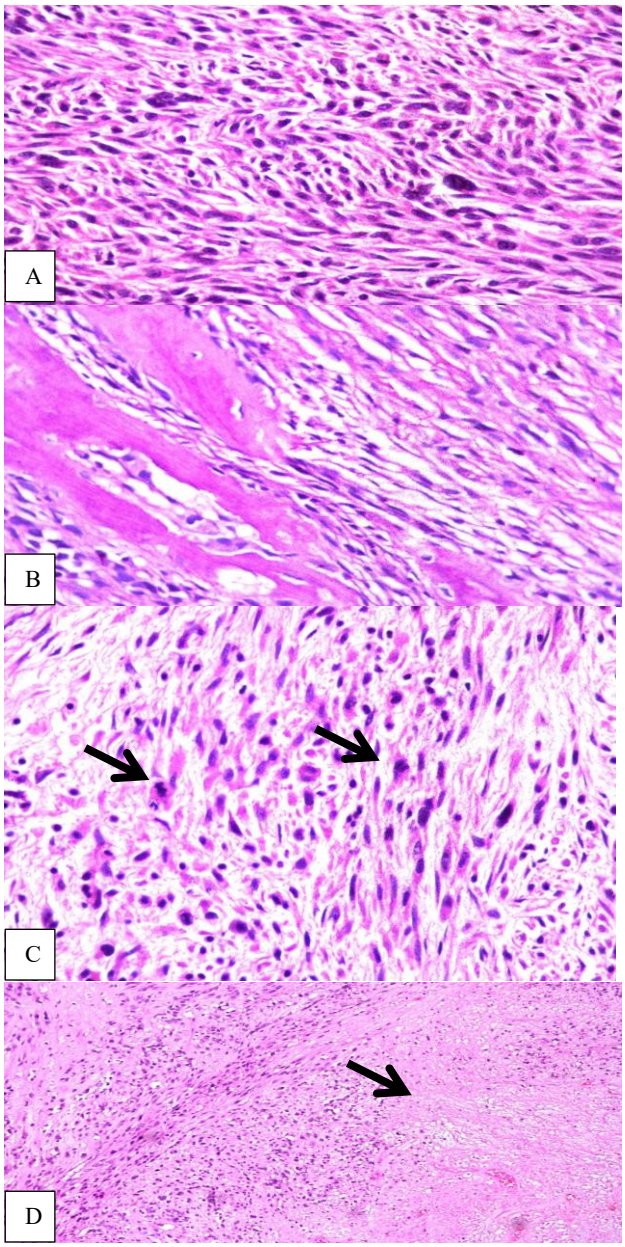


Figure 3 (A-D): Cellular tumor composed of fascicles of spindle cells HE ×100. B: Spindle cells infiltrating the bony trabeculae HE ×100. C: Marked atypia within the tumour cells HE ×100. D: Areas of necrosis HE ×100.

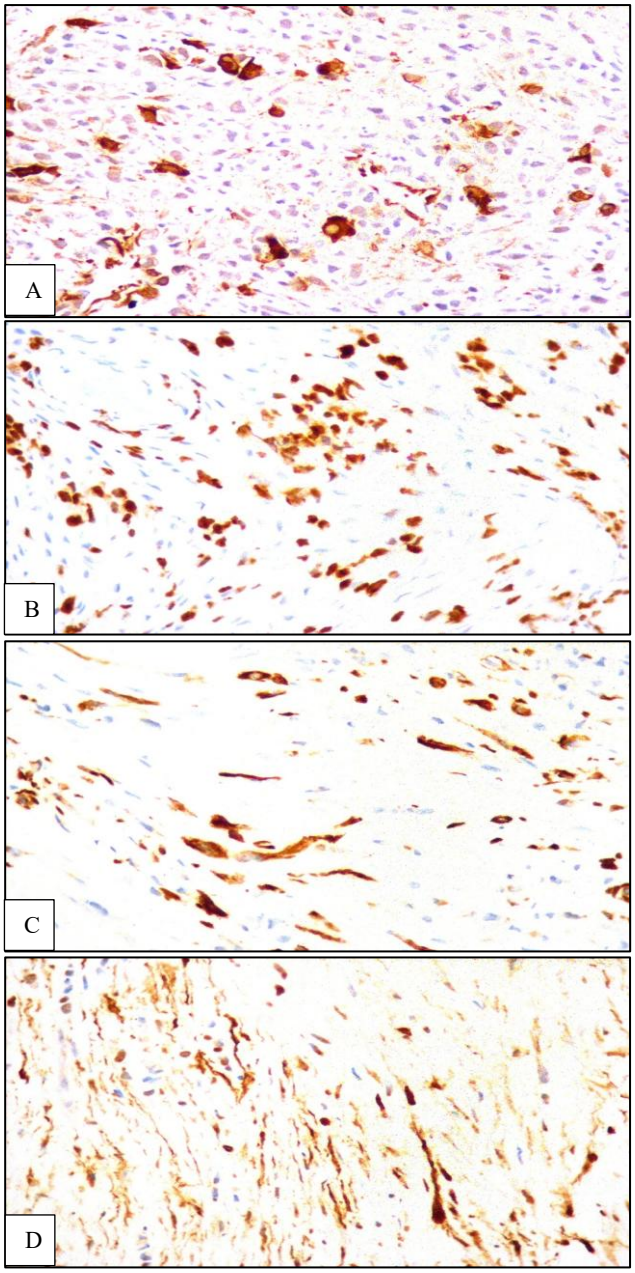


Figure 4 (A-D): Positive immunostaining with Desmin ×100. B: Positive immunostaining with MyoD1× 100. C: Positive immunostaining with SMA ×100. D: Positive immunostaining with MSA ×100.

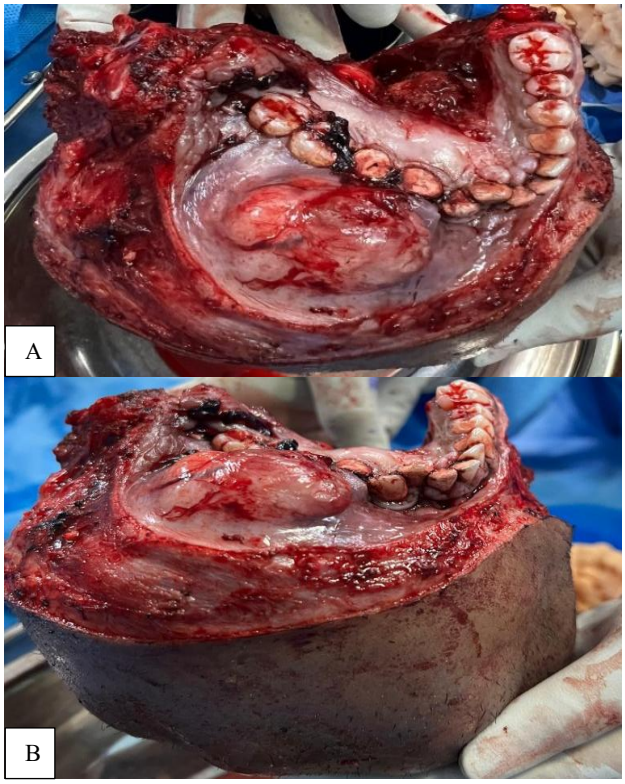
PET CT whole body was done which showed FDG avid destructive lytic lesion involving mandible (R>L) with associated large heterogeneously enhancing soft tissue component and extensions which was consistent with biopsy proven spindle cell neoplasm. No other metabolically active disease elsewhere in whole body survey.

The case was then discussed in the tumour board committee, and decided that if R0 resection is amenable, surgery followed by adjuvant chemotherapy could be proceeded if not to start with neoadjuvant chemotherapy.

Since a complete excision without any major disfigurement or decreased function was possible, he was taken up for surgery and segmental mandibulectomy with wide local excision of overlying skin with tracheostomy was performed and the defect was reconstructed with right fibular osteomyocutaneous flap and left anterolateral thigh flap. The Tumor was classified based on the intergroup RMS study group as stage 1, group 1.

His postoperative period was uneventful. The final histopathology showed a high-grade Spindle cell variant of embryonal RMS with mitotic rate of 24-26 per 10 high power field, 10-15% necrosis, no lymphovascular invasion and the resected margins were free of tumour.

Microscopic examination showed fibrocollagenous and fibromuscular tissue showing infiltration by a malignant tumour comprising of short and long fascicles of spindle cells admixed with areas of necrosis and myxoid change. Additionally, FOXO1 (FKHR) (13Q14) re arrangement by FISH was done to rule out alveolar type RMS, which was reported negative.



Figures 5 (A and B): Wide local excision of the lesion with the overlying skin.

Patient did not have chemotherapy as advised and came for review 3 months after surgery. At the time he had necrotic lesions in the lower lip and multiple tender subcutaneous nodules in the face and the neck.

He then underwent 2 cycles of chemotherapy (C1-Actinomycin D 1.6 mg+vincristine 2 mg+cyclophosphamide (Endoxan) 1800 mg+Peg

GCSF+mesna 600 mg) after which, he was reviewed again.

PET CT was done and there was increase in size, number and metabolic activity of multiple subcutaneous necrotic lesions in head and neck, increased metabolic activity of FDG avid prominent enlarged necrotic level IA, bilateral level II and III nodes and in the marrow of right mid shaft of femur which was likely metastatic.

In view of disease progression, he was started on a second line chemotherapy that consisted of carboplatin 600 mg +VP 16 and inj. Peg-religrast for 3 cycles. The patient is still on follow up.

DISCUSSION

RMS is the most common childhood and adolescent sarcoma showing features of skeletal muscle differentiation.[7]

Head and neck are the primary location for RMS and accounts for 36% of tumours. RMSs are further subdivided into the parameningeal and non-parameningeal types**.** The tumours of mandible and oral cavity are included in the non parameningeal type which has a favorable outcome. In the oral region, a site predilection has not been stated clearly.[8] Some authors report that oral RMS more frequently occurs in the soft palate. In the present patient, the structures primarily involved were the mandible extending intraorally and posteriorly abutting the parotid gland, which is noteworthy as it is not recognized as a common site for occurrence within the mouth.

Histologically, the most prevalent kind of RMSs in the head and neck are of the embryonal type, as seen in this case. RMS shows small, round-to-spindle-shaped cells having moderate nuclear pleomorphism.[9] In this particular case, the diagnosis of spindle cell variant RMS was supported by the presence of spindle cell fascicles with hyperchromatic nuclei.

Spindle cell RMS is a rare type of RMS which is categorized under embryonal RMS. Following the updated WHO classification, 5-13% of cases have been documented to have a spindle cell subtype of RMS in the oral cavity.[10] Although it is more common in the paratesticular area, the spindle cell type can also be seen in the head and neck, particularly in the orbit and the extremities. In this report the primary site being the mandible and its extension to the oral cavity makes it a rare presentation. Tumor sizes can vary from as small as 1.5 cm to around 35 cm, as in this case report the size was 16 cm.[11,12]

Immunohistochemistry plays a crucial role in diagnosing the type of RMS. The WHO classifies spindle cell and embryonal tumors as a single category since they both have recurrent mutations of the MyoD1 gene.[13] In this case IHC showed positive for Desmin which is highly sensitive in 75-100% of RMSs. Similarly, IHC markers that show skeletal muscle development, such as Desmin, myogenin, and MyoD1, support in the diagnosis of RMS, as seen in this case study MyoD1, Desmin and SMA (Smooth muscle actin) were all positive.

Recently, molecular biology approaches have further characterized RMS by the presence or absence of fusion proteins.[14] FISH can also be used to differentiate embryonal RMS from other RMS subtypes; a FOX01 rearrangement will be identified in alveolar RMS, and a NCOA2 rearrangement will be identified in spindle ell/sclerosing RMS. Like in this case study the FOX01 rearrangement was negative.

The prognosis can be determined based on the tumor size, margin status following surgical resection, and presence of metastases. Although age and histological subtype seem to be significant prognostic factors in children, there is controversy over their effect in adults.

The patient in our case presented with a recurrence within three months following a R0 resection and ongoing chemotherapy, demonstrating the aggressiveness of the diseases, despite the fact the margins resected were histologically free.

CONCLUSION

This is case of spindle cell embryonal RMS in a 17-year-old male involving the mandible. This is rare and has an aggressive nature with a high recurrence potential therefore accurate and timely diagnosis becomes crucial in the treatment.

It is important to distinguish between tumours with favorable histology (all embryonal RMSs) and unfavorable histology (alveolar and anaplastic RMS, undifferentiated sarcoma), because a specific histopathologic type determines treatment and predicts survival and prognosis.

Therefore, a multidisciplinary strategy should be used for the treatment of RMS. Due to RMS's propensity to spread to the bone marrow, it should involve surgical resection of tumour, multi agent chemotherapy with or without radiation. In certain cases, it may also require neoadjuvant chemotherapy based on the staging of the tumour.

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REFERENCES

1. Enzinger F, Enzinger WS. Weiss's: Soft tissue tumors. Mosby. Elsevier Science Health Science. 2019.

2. Parham DM, Barr FG. Classification of rhabdomyosarcoma and its molecular basis. Adv Anatomic Pathol. 2013;20(6):387-97.
3. Nascimento AF. Rhabdomyosarcomas in adults: classification and differential diagnosis. Diagnostic Histopathol. 2008;14(11):538-45.
4. Kaur P, Kaur A, Suri AK, Malik H. Spindle cell variant of embryonal rhabdomyosarcoma: A rare entity with diagnostic challenges. J Clin Diagnost Res. 2016;10(8):ED17.
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(3):229-35.
6. Kaseb H, Kuhn J, Gasalberti DP. Rhabdomyosarcoma. StatPearls. Treasure Island (FL): StatPearls Publishing. 2024.
7. Bergeron C, Jenney M, De Corti F, Gallego S, Merks H, Glosli H, et al. Embryonal rhabdomyosarcoma completely resected at diagnosis: The European paediatric Soft tissue sarcoma Study Group RMS2005 experience. Eur J Cancer. 2021;146:21-9.
8. Arya K, Vij H, Vij R, Rao NN. Rhabdomyosarcoma of mandible: A diagnostic predicament. J Oral Maxillof Pathol. 2011;15(3):320-5.
9. 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(1):S103-6.
10. 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(4):494-500.
11. Carroll SJ, Nodit L. Spindle cell rhabdomyosarcoma: a brief diagnostic review and differential diagnosis. Arch Pathol Lab Med. 2013;137(8):1155-8.
12. Nascimento AF, Fletcher CD. Spindle cell rhabdomyosarcoma in adults. Am J Surg Pathol. 2005;29(8):1106-13.
13. Agaram NP, Chen CL, Zhang L, LaQuaglia MP, Wexler L, Antonescu CR. Recurrent MYOD1 mutations in pediatric and adult sclerosing and spindle cell rhabdomyosarcomas: evidence for a common pathogenesis. Genes Chromosomes Cancer. 2014;53(9):779-87.
14. Zarrabi A, Perrin D, Kavoosi M, Sommer M, Sezen S, Mehrbod P, et al. Rhabdomyosarcoma: current therapy, challenges, and future approaches to treatment strategies. Cancers. 2023;15(21):5269.

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