

CASE REPORT



Spindle cell embryonal rhabdomyosarcoma mimicking innocuous lesion – A case report

S. Muthu Kumar¹, B. Suman², K. R. Vijaylakshmi², C. Ramanna³

¹Department of Oral Medicine and Radiology, Dental Health Officer, Community Health Care Center, Masur, Maharashtra, India, ²Department Oral Medicine and Radiology, Government Dental College and Research Institute, Bengaluru, Karnataka, India, ³Department of Oral and Maxillofacial Pathology, Consultant Oral Pathologist, Bengaluru

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Correspondence:

S. Muthu Kumar, Department of Oral Medicine and Radiology, Dental Health Officer, Community Health Care Center, Masur, Maharashtra, India. Phone: +91-7829497673. E-mail: muthukumardoc@gmail.com

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Abstract

Rhabdomyosarcomas (RMSs) are rare malignant neoplasms that originate from the embryonal cells called rhabdomyoblasts. Hence, they tend to be much more common in children, although rarely occurring in adults. A 19-year-old adult female patient reported with a complaint of painful growth in the lower anterior jaw. The patient gave a history of growth in the lower anterior labial vestibule 2 months ago which was insidious in onset and was first noticed 2 months ago for which patient had visited a private dental hospital where it was excised, records of which were not traceable. The clinical features with radiological, histopathological, and immunohistochemical findings confirmed the diagnosis of spindle cell embryonal RMS (ERMS) which are discussed in this paper in detail. Early diagnosis plays a pivotal role in the prognosis of spindle cell ERMS which is a fatal, rapidly proliferating, and malignant neoplasm. A thorough knowledge of the pathognomonic presentation of RMS can aid in differentiating from various innocuous soft tissue growths. Hence, the oral physician plays a key role in prompt diagnosis of such lesions which ensures better prognosis.

Introduction

Spindle cell embryonal rhabdomyosarcoma (ERMS) is also known as myosarcoma, malignant RMS, botryoid sarcoma, rhabdopoietic sarcoma, rhabdosarcoma, and embryonal sarcoma.

ERMS represents approximately 4% of all malignancies among children and adolescents, and has an annual incidence of approximately 4 per million. Oral spindle cell ERMSs are rare, malignant, and neoplasias derived from proliferation of embryonic mesenchymal tissue, corresponding to 10–12% of all head and neck RMSs.

Weber first reported RMS in 1854 and Arthur Purdy Stout first delineated RMS as a distinct entity.^[1] It shows a characteristic age distribution, most of the patients being within the second decade of life. RMSs are associated with a high rate of recurrence; an aggressive neoplastic behavior characterized by highly invasive, immature cells, and generalized metastasis through hematogenic and/or lymphatic routes.^[2]

The diagnosis of RMS is based on thorough clinical, radiological, and histopathological analysis. However, special techniques such as immunohistochemistry and chromosomal

studies play a very important role in diagnosis. Differential diagnosis of reactive, inflammatory lesion, and malignant neoplastic growths should be borne in mind when oral clinicians encounter rapidly progressing soft tissue growths in a young adult.

Case Report

A 19-year-old female presented to the department of oral medicine and radiology, at our institution, with a painful growth in the lower jaw in the anterior region for 1 month.

The patient gave a history of growth in the lower anterior labial vestibule 2 months ago which was insidious in onset, which was first noticed 2 months ago, gradually progressed in size, associated with pain, bleeding, and mobility of teeth, with no history of trauma or fever. The patient had visited a private dental hospital where it was excised, records of which were not traceable. However, following excision recurrence of growth was noted in same site after a week, which was rapidly progressive.

Medical history and family history were noncontributory with no history of any deleterious habits. The general physical

examination of the patient did not reveal any sign of a pathological state associated with other systems. Vital parameters were within normal limits.

Bilateral submandibular lymph nodes were palpable, soft, mobile, and tender. Gross facial asymmetry was noted in the lower 1/3rd of face due to the roughly oval-shaped, diffuse, smooth-surfaced, firm, and non-tender swelling measuring about 3 to 4 cm in greatest dimensions, skin over the swelling appeared stretched with no evidence of discharge or ulceration [Figure 1].

Intraorally, an oval, lobulated, and sessile growth measuring about 3.5 cm × 2.5 cm in its greatest dimension in the anterior region of the lower jaw, obliterating the labial vestibule, and remarkably pushing the tongue lingually was noted. It extended anteriorly from distal right lower lateral incisor, crossing the midline posteriorly up to mesial aspect of the left first molar. Mucosa over the growth appeared bluish with indentations of teeth in the lingual aspect of the growth [Figure 2]. On palpation, the growth was soft, fleshy, tender, and bleed on gentle pressure, and associated teeth numbers 31, 32, and 33 were Grade III mobile. Electric pulp test revealed delayed response w.r.t 42, 43, and 44 and no response w.r.t 41, 31, 32, and 33.

A provisional diagnosis of aggressive giant cell granuloma was considered based on the history and clinical presentation. Differential diagnosis of pyogenic granuloma, sarcoma (fibrosarcoma), and Ewing's sarcoma was considered.

Intraoral periapical radiograph revealed a wide area of irregular radiolucency involving the alveolar bone from the left lower canine to the right lateral incisor with floating teeth appearance suggesting the diagnosis of an aggressive malignant tumor.

CT imaging revealed an ill-defined, heterogeneously enhancing soft tissue attenuation mass with central areas of necrosis in the left anterior buccal spaces, and vestibule with destruction of the left hemimandible and alveolar process. The lesion was carrying mass effect in the form of the displacement of the muscles of tongue to contralateral side [Figure 3]. Routine blood investigations were within normal limits and posteroanterior chest view appeared normal.

The histopathology of the incisional biopsy specimen showed stratified squamous epithelium with focal ulceration. The connective tissue showed destroyed bony spicules with an infiltrating malignant neoplasm arranged in fascicles composed of spindle-shaped cells in "herringbone" pattern with inflammatory infiltrate and vascular spaces in between them. There was no rhabdomyoblasts differentiation seen. The histological diagnosis was established as spindle cell variant of RMS with a differential diagnosis of fibrosarcoma; monophasic synovial sarcoma and malignant peripheral nerve sheath tumor were considered.

Further, it was definitively delineated using immunohistochemical markers by polymer IHC technique using MM: Mouse monoclonal and RM: Rabbit monoclonal antibodies. Markers were done (with clones) included Vimentin (Vg MM) [Figure 4], Desmin (33 MM), Myogenin (F5D MM), S100 (15E2E2 MM), Ki67 (Ki88MM), SMA (IA4 MM), h-caldesmon (h-CD MM), Alk (SP8 RM), CD34

(OBEnd 10 MM), Bcl2 (100 MM), CD99 (12E7 MM), CK (AE1+AE3 MM), and Melan A (A103 MM). The neoplastic cells were strongly positive for vimentin, desmin (2-3+, 90%), Bcl2 (3+, >90%), CD99 (2-3+, 50-60%), weakly positive for S100 (1+, 50-60%), and focal myogenin positivity (2-3+, 5%) with high Ki67 proliferative index of 60-70%. Rests of the markers were negative. The morphology and immunoprofile was consistent with spindle cell ERMS.

Subtotal mandibulectomy and radical modified neck dissection were performed, but unfortunately, we lost the patient after a week due to postsurgical complications.

Discussion

ERMS is defined as a primitive, malignant soft tissue sarcoma that recapitulates the phenotypic and biological features of embryonic skeletal muscle.

Although no known etiologic factors have been identified to account for the occurrence of this malignant neoplasm, genetic abnormalities have been proposed like loss in heterozygosity in chromosome 11p15 in most ERMSs, imprinting abnormalities, and heterogeneous expression pattern on gene expression. Cytogenetic features found are gains of chromosomes 2, 7, 8, 11, 12, 13q21, and 20 and losses of 1p35-36.3, 6, 9q22, 14q21-32, and 17.^[3]

RMS can occur at any site but is most commonly observed either in the genitourinary region or the head and neck region with an incidence of 36%. The occurrence of ERMS in the head and neck region is 28%, chiefly from the facial (orbital, tonsil, soft palate, mastoid, internal ear and parotid, zygoma, and temporal) and cervical musculature. Cheek, mandible, and gingiva are found to be additional sites of occurrence.^[4,5]

Clinically, presentation of ERMS is variable, influenced by age, site, and the presence or absence of distant metastases. Since ERMS is very rare in adults, the data on clinical presentation are not clear. However, few studies have described that these tumors are more aggressive in adults compared to children and adolescents. ERMS tends to form expansile, rapidly growing, hemorrhagic, soft, and fleshy tumors ranging in size from 2 to 8 cm.^[6]

Spindle cell ERMS presented in a young 19-year-old female who reported to us with a rapid and painful growth in the mandibular alveolar ridges with obliteration of the labial and lingual vestibule. The definitive diagnosis was confirmed with immunohistochemical panel.

Differential diagnosis comprised rapidly growing growth such as aggressive giant cell granuloma, fibrosarcoma, and Ewing's sarcoma.

Aggressive giant cell granuloma may be seen in all age groups, it is much more common in the young, especially those under 30 years of age with female predilection and anterior segment of mandible is more often affected.

Fibrosarcoma occurs in any location but mainly affects long bones and 5% occur in head and neck region, mandible being the most common site, clinically, the lesion can cause



Figure 1: Extraoral clinical photograph showing swelling in submental region



Figure 2: Fleshy, oval, and sessile mass in anterior region of the lower jaw obliterating labial vestibule

pain, swelling, paresthesia and occasionally loss of teeth and ulceration of the overlying mucosa. However, it has been reported in all age groups, most commonly in the 3rd–6th decade of life and occurs slightly more commonly in men than in women.

Ewing's sarcoma is most common in the second decade of life and manifests with intermittent pain, rapidly progressive swelling that may often ulcerate. Furthermore, low-grade fever, facial neuralgia, and lip paresthesia can occur. Radiographically, it appears as an irregular radiolucency with a characteristic onion skin appearance.

Radiographically, ERMS appears as ill-defined, ragged bordered radiolucencies with grossly displaced teeth that appear to be floating in radiolucencies.

Histopathologically, in many instances, the tumors characteristically show the presence of spindle-shaped cells arrayed in a "herringbone" pattern. Similar features were noted in our case. Depending on their differentiation, the cytoplasm

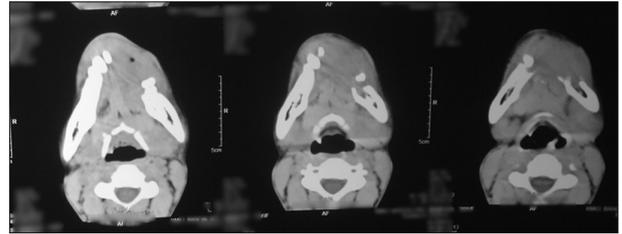


Figure 3: Computed tomography imaging revealing an ill-defined, heterogeneously enhancing soft tissue attenuation mass in the left anterior buccal spaces, and vestibule with destruction of the left hemimandible and alveolar process

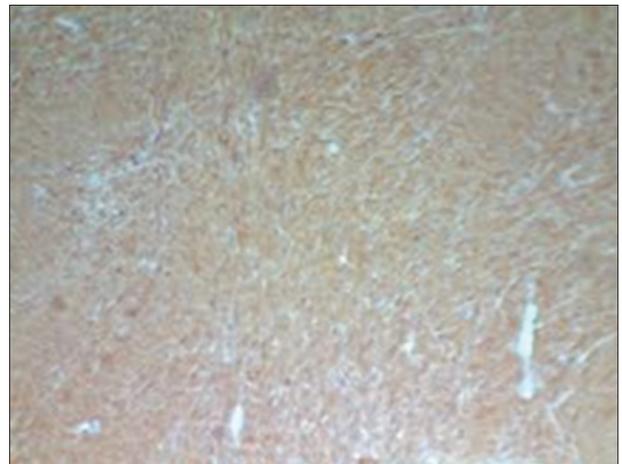


Figure 4: Immunohistochemical markers were done (with clones) showing strongly positive for vimentin

may be scant and indistinct or more abundant and strongly eosinophilic. Immunomarkers used to confirm RMS include desmin, myoglobin, myosin, vimentin, muscle-specific actin (HHF 35), sarcomeric actin, smooth muscle actin, and troponin T3. A positive reaction for vimentin confirms a mesenchymal-derived tumor and desmin indicates muscular differentiation.^[7,8] Desmin expressed in myotome of embryo is identified in 75–100% of RMSs. It is regarded as best single marker for diagnosis of poorly differentiated RMS. Myoglobin appears to be specific for skeletal muscle but it has average sensitivity. In our presented case, vimentin, desmin, Bcl2, and CD99 were strongly positive.^[9,10]

ERMS is generally treated surgically followed with a chemoradiotherapy regimen.

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Dr. Mubeen. B.D.S, M.D.S
Professor and Head of the Department
Oral medicine and radiology
Government Dental College and Research Institute, Bengaluru.

Conclusion

ERMSS are extremely rare in the oral cavity and if they occur appears as aggressive soft tissue lesions with high recurrence rate. Hence, oral physicians should be alert with such clinical presentation in young patients and always consider RMS for differential diagnosis to ensure early diagnosis and better prognosis.

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