Case Report

Alveolar type of rhabdomyosarcoma of maxilla—A case report

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Abstract

Rhabdomyosarcoma (RMS) is the most common sarcoma among children and accounts for 20% of soft tissue sarcomas. In children, close to 50% of rhabdomyosarcomas arise in the head and neck. RMS of the oral cavity is rare and is seen in only 10–12% of all head and neck lesions and the involvement of the jaws is extremely rare. Histopathologically, the various types are pleomorphic type, botryoid type, spindle cell type, embryonal, and alveolar type of RMS. The alveolar variant accounts for almost 30% of all rhabdomyosarcomas and tends to arise in patients of the age group 10–25 years. We present a case of orofacial RMS in a young adult who was referred to our Institution for the management of an odontogenic lesion of the maxilla. The clinicopathological aspects and poor survival rate as a consequence of delayed diagnosis are discussed. We dentists may misdiagnose it as an odontogenic tumour due to its location in the oral and maxillofacial region. Careful clinical history and examination and investigations may help to narrow down the diagnosis. Expert opinion and referrals to oral pathologists and oncologists are essential to arrive at early diagnosis and to initiate the treatment.

Keywords: Alveolar RMS, non-odontogenic malignant tumour of the maxilla, RMS-Rhabdomyosarcoma

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INTRODUCTION

Rhabdomyosarcoma (RMS) is a malignant soft tissue tumour of skeletal muscle origin. It was first described by Weber in 1854 and is most common in children accounting for 6% of all malignancies in children under the age of 15 years.[1]

It commonly involves the head and neck, genitourinary tract, retroperitoneum, and extremities.[2] Head and neck RMS are further divided into Orbital, parameningeal, and non-orbital non-parameningeal, based on anatomical distribution. RMS of the oral cavity is rare accounting for 10–12% of all head and neck tumours.

Clinically orofacial RMS presents as a cutaneous nodule, rapidly growing facial mass, paresthesia, and facial palsy. Histopathological subtypes include pleomorphic type, botryoid type, spindle cell type, embryonal, and alveolar type of RMS.[3] The prognosis and survival rate depend on various factors which include age, site, delayed diagnosis, distant metastasis, and the histopathological sub-type. Alveolar RMS has the worst prognosis among the histopathological subtypes of RMS.[4]

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In this case report, we present a case of alveolar rhabdomyosarcoma involving the maxilla in a 23-year-old female patient who was referred to our dental college, suspecting it as an odontogenic tumour. We have described elaborately the clinical, radiological, histopathological, and immunohistochemical features of alveolar rhabdomyosarcoma.

CASE HISTORY

A 23-year-old female patient complained of swelling and pain in the right maxilla for the past 2 months.

The pain was gradual in onset, intermittent in nature, pricking type, radiating, aggravated on mastication, and relieved on medication. The patient has consulted a physician and underwent investigations suggesting odontogenic lesion on the right maxilla. The past medical history revealed no relevant medical, surgical, and another dental history.

Examination revealed facial asymmetry due to single diffuse swelling on the right middle third of the face extending superiorly from infraorbital rim and inferiorly 1 cm below the ala tragus line, anteriorly extending up to commissure of the lip, posteriorly 5 cm anterior to the pinna of the ear. Diplopia of the right eye was present and interpupillary distance was 30 mm.

Inspectory findings were confirmed during palpation. The swelling was tender, firm in consistency, non-fluctuant, and fixed to the underlying bone. The surface of the swelling was glossy and non-pinchable.

Level II B nodes were approximately 1.5 * 2 cm in size, mobile, soft, and tender. Level III nodes were palpable.

The mouth opening was 36 mm. Intraoral examination revealed missing of 16, 17, and 18 and no mobility of remaining teeth present. Single diffuse swelling over the right maxilla extending from anteriorly from 13 regions posteriorly to soft palate and medially 5 cm from the mid palatine region and laterally extending beyond the bucco-gingival sulcus and was firm in consistency and tender.

A provisional diagnosis of a malignant tumour of the maxilla was given. Investigations like routine blood examination and radiographs were taken. Orthopantomogram (OPG) revealed an irregular radiolucency over the right maxilla. Computed tomography (CT) scan revealed an expansile and osteolytic lesion in the same area.

A biopsy was done for histopathological examination. The haematoxylin and eosin-stained section showed malignant neoplastic cells with features of poorly differentiated small round cell tumour in the form of nests, sheets and alveolar patterns with perithelial rosettes at places. Extensive necrosis, high cellularity, and atypical features were noted. Connective tissue showed bundles of collagen fibres, areas of hyalinization, adipose tissue, blood vessels, haemorrhagic areas, and bony spicules [Figure 1]. A provisional diagnosis of round cell tumour was made. Differential diagnoses were Ewing's sarcoma, non-Hodgkin's lymphoma, mesenchymal chondrosarcoma, rhabdomyosarcoma, and PNET. (primitive neuroectodermal tumour).

Further, immunohistochemistry was done for definite typing and it was positive for vimentin, desmin [Figure 2], and myogenin [Figure 3] and it was confirmed as alveolar rhabdomyosarcoma. The patient was referred to the oncology department for management and follow-up.

DISCUSSION

Rhabdomyosarcoma is a high-grade malignant neoplasm whose lineage is from mesenchymal cells and is related to skeletal muscle cells.[5] It was first described in the English literature in 1937 and in 1946 Stout described rhabdomyosarcoma as a tumour with rhabdomyoblasts of round, strap, racquet, and spider forms.[6]

RMS commonly involves the head and neck (35%), followed by the genitourinary tract (23%) and extremities (17%). The most common location is the head and neck area, which is further subdivided into orbital, parameningeal sites (nasopharynx, nasal cavity, paranasal sinuses, temporal bone, pterygopalatine fossa, and the infratemporal fossa), and non-parameningeal sites (neck, parotid region, oropharynx, cheek, masseter muscle, scalp, oral cavity, and larynx).[2]

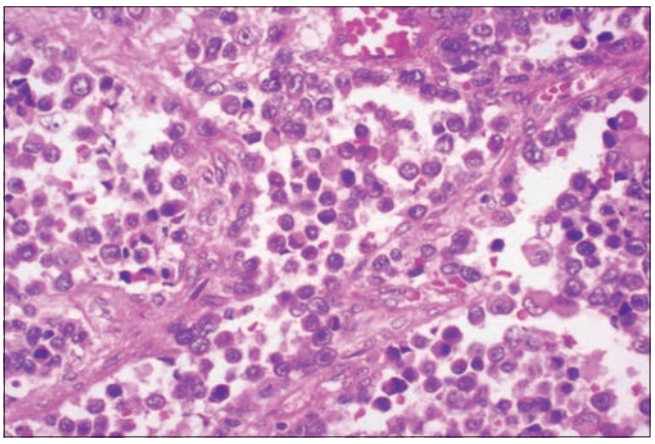

Figure 1: The haematoxylin and eosin-stained section showed malignant neoplastic cells with features of poorly differentiated small round cell tumour cells in the form of nests, sheets, and alveolar pattern. (H and E stain, ×40 magnification)

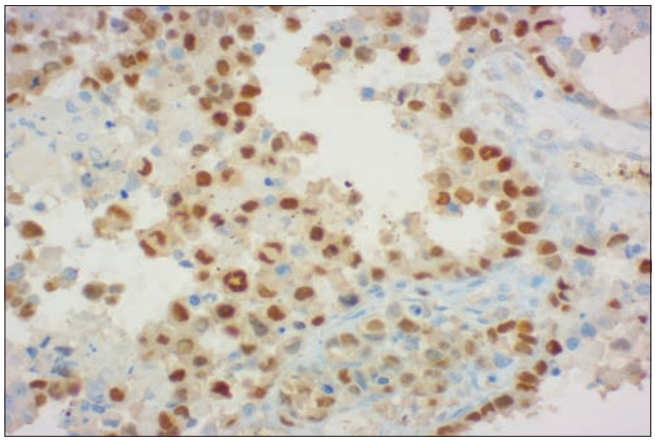

Figure 2: Positive immunohistochemical staining for desmin, IHC, ×40 magnification

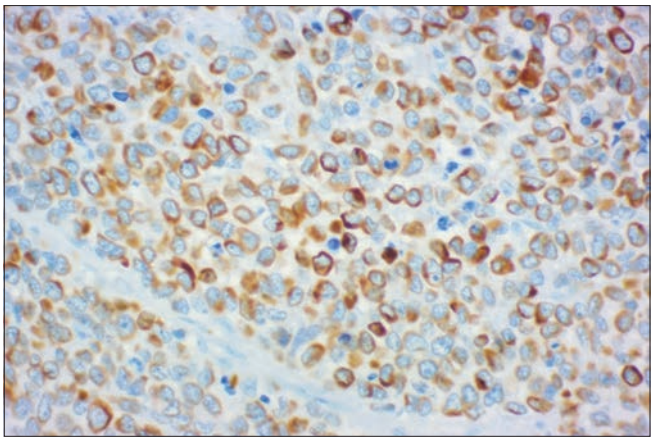

Figure 3: Positive immunohistochemical staining for myogenin, IHC × 40 magnification

The 2013 World Health Organization (WHO) classification of skeletal muscle tumours modified the histologic classification of RMS and included Sclerosing RMS which is separated from embryonal RMS. The current WHO classification includes pleomorphic, alveolar, spindle cell/sclerosing, and embryonal, subtypes of RMS and does not separate the botryoid subtype.[5]

The 2020 WHO classification of soft tissue tumours

Skeletal muscle tumours.

Benign

Rhabdomyoma

Malignant

Embryonal rhabdomyosarcoma

Alveolar rhabdomyosarcoma

Pleomorphic rhabdomyosarcoma

Spindle cell/sclerosing rhabdomyosarcoma

Ectomesenchymoma.

RMS is the most common childhood and adolescent soft tissue sarcomas, most common in children accounting for 6% of all malignancies in children under the age of 15 years.[1]

RMS comprises a heterogeneous into two major histologic subtypes, embryonal (ERMS) occurring in children less than 10 years of age with a generally favourable prognosis and alveolar (ARMS) occurring in adolescents and young age with a poor prognosis. These clinical and pathologic differences between ARMS and ERMS are hypothesized to be the result of different molecular alterations in myogenic precursors and different biological mechanisms of tumorigenesis.[7]

Chromosomal analyses of RMS cases demonstrated a translocation involving chromosomes 2 and 13, t(2;13) (q35;q14), that was detected in 70% of published ARMS cases. In addition, there have been several reports of a t (1;13) (p36;q14) variant translocation. These two translocations have not been associated with any other tumour and thus appear to be specific markers for ARMS.[8]

RMS has bimodal peak incidence. The first peak occurs in children, the second peak occurs in adolescence with a slight male predilection.[9]

In our case, the patient was a female and 23 years old falling into the adolescent age.

Children and young adolescents have more head and neck involvement and are usually of Embryonal type. Involvement of extremities is more common in adolescents and usually of Alveolar type. The pleomorphic type occurs in extremities and is common among adults. Botryoid RMS is common in children and involves the hollow viscera.[10] Our patient was 23 years old and was found to be diagnosed with an alveolar type of rhabdomyosarcoma in the head and neck region.

Clinical signs and symptoms are highly variable and depend on the site, age, local invasion, and metastasis. Tumours of head and neck present as a nodule or polypoid lesion, painless or painful mass, nasal block, rhinorrhoea, recurring otitis media,[11] eye proptosis,[12] and facial nerve palsy[13] and often spread to contiguous sites including the base of the skull and temporal bones.

In general, most patients have an advanced disease even at the stage of initial presentation because RMSs are known to show rapid growth and the patients generally tend to delay medical consultation.[1] Oral RMSs often have a rapidly enlarging painless mass, usually larger than 1 cm in diameter at the time of presentation.[14]

The most prevalent sites in the oral cavity are the palate, buccal mucosa, tongue,[15] lip, gingiva, mandible, and maxilla which can be associated with pain, trismus,[16] and paresthesia, loss of a tooth, ankyloglossia.

In our case, the patient had facial asymmetry due to single diffuse swelling on the right middle third of the face, diplopia of the right eye and restricted mouth opening and it involved the right maxilla, hard palate, and soft palate.

Distant metastasis most commonly involves lymph nodes, bones, bone marrow, and the lungs.[17] In our patient, there was the involvement of level II and level III lymph nodes. In the head and neck RMS, lumbar puncture with the cytological examination, skeletal survey, bone scan and bone marrow biopsy, and PET scan should be carried out to rule out metastasis.

Among the histopathological subtypes, pleomorphic type is very rare in children.[18] Embryonal occurs in infants and young children and often involves the head and neck and urogenital tract.[19] Alveolar rhabdomyosarcoma occurs in children and young adults between the ages of 2 and 25 years. Alveolar rhabdomyosarcoma typically has a characteristic alveolar growth pattern, and consists of small cells with round nuclei and a scant cytoplasm as well as larger cells with more eosinophilic cytoplasm and round, eccentric nuclei and they are usually nested with fibrovascular septa. Sometimes cells are seen with cross striations.[20] similar to that seen in our case which had sheets of round cells in an alveolar pattern with perithelial rosettes.

Differential diagnosis includes Ewing's sarcoma, non-Hodgkin's lymphoma, mesenchymal chondrosarcoma, and small cell variant of osteosarcoma which are more common among the younger age group. Tumour cells are usually round and morphologically similar in appearance. The pattern of infiltration or the components of tumour may differ which could be tumour osteoid in small cell variants of osteosarcoma and hyaline cartilage as in mesenchymal chondrosarcoma. But the features like multinucleated tadpole-like or racquet cells, intracytoplasmic vacuoles and cross striation of tumour cells seen in RMS would suggest the differentiation lineage.[6] The cytoplasm of soft-part sarcoma is granular or finely vacuolated rather than fibrillar and nuclei are prominent, unlike the Alveolar RMS.

Immunohistochemical studies reveal that RMS is usually positive for desmin, myoglobin, myogenin myosin, vimentin, muscle-specific actin (HHF 35), sarcomeric actin, smooth muscle actin, and troponin-T. Occasionally, S100 protein and cytokeratin may also be positive.

Desmin is among the earliest muscle structural gene to be expressed in the myotome of the embryo. It has been regarded as the best single marker which has high sensitivity but is not very specific for skeletal muscle for the diagnosis of poorly differentiated RMS. Myogenin and MyoD1 nuclear expression was noted in 91% of RMS, and offers the advantage of high sensitivity and specificity in identifying RMS.[14] Our case showed positivity for vimentin, desmin, and smooth muscle actin confirming the diagnosis of rhabdomyosarcoma. Other recent diagnostic panel includes flow cytometry and genetic analysis with regard to gene fusion status.

The International Classification of Rhabdomyosarcoma includes botryoid and spindle cell RMS as superior-risk groups, embryonal RMS as an intermediate-risk group, and alveolar RMS as an unfavourable-risk group.[5]

Since the clinical manifestations are non-specific diagnosis, therapy may be delayed and could decrease the morbidity rate.

Multimodal management comprising surgical, chemotherapy, and radiation therapy may improve the survival rate. The survival rate varies depending on the tumour location, age, clinical, biologic, and pathologic characters, stage, and risk group.

Adults have poor 5-year overall survival (27%) compared with children (61%). The overall survival of metastatic rhabdomyosarcoma patients is low and typically does not exceed 25%. Parameningeal rhabdomyosarcoma and rhabdomyosarcoma of the extremities tend to have a worse prognosis compared to other sites.[10] Alveolar pattern with nodal involvement has a less favourable outcome.[21,22]

We dentists may misdiagnose it as an odontogenic lesion due to its location in the oral and maxillofacial region. Careful clinical history and examination and investigations may help to narrow down the diagnosis. Expert opinion and referrals to oral pathologists and oncologists are essential to arrive at an early diagnosis and to initiate the treatment. RMS is managed by a multidisciplinary approach consisting of all the concerned specialists.

Further study and research are needed for early diagnosis, and management of RMS and to improve the survival rate. The morbidity and mortality rates should be reduced and the quality of life should be improved.

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Conflicts of interest
There are no conflicts of interest.

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