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CRTC1/MAML2 fusion transcript in central mucoepidermoid carcinoma of mandible—diagnostic and histogenetic implications☆

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Abstract Intraosseous salivary gland carcinomas are extremely rare, comprising only 2% to 3% of all mucoepidermoid carcinomas (MECs) reported. The t(11;19) translocation and its CRTC1/MAML1 fusion transcript have been identified in MEC at different sites and are believed to be associated with the development of a subset of these tumors. However, the status of the fusion transcript has not been reported in intraosseous MEC. Here, we report 3 examples of central MEC of the mandible, including a case with a history of primary retromolar MEC. Reverse transcriptase–polymerase chain reaction and DNA sequencing analyses of the microdissected components of these tumors were used for the detection and verification of the fusion transcript. We identified, for the first time, the t(11;19) fusion gene transcript in central MEC, including in the previous primary retromolar MEC. No fusion transcript was detected in the second primary noncentral MEC or in another central MEC. The results indicate that central MEC can manifest the fusion transcript. This finding may have diagnostic and histogenetic roles in the future analysis of this entity.
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Keywords: Intraosseous mucoepidermoid carcinoma; Fusion gene; Histogenesis

1. Background

Mucoepidermoid carcinoma (MEC) typically arises from major or minor salivary glands and comprises 5% to 10% of all salivary gland tumors [1-3]. Intraosseous salivary gland carcinomas are extremely rare, comprising 2% to 3% of all MECs reported [1,2,4,5]. Approximately 108 examples of MEC arising in the mandible have been reported [4,6-8]. Central MEC affects females twice more frequently than males and involves the mandible twice more often than the maxilla. The most common site of occurrence is the premolar-molar-angle region of the mandible. It has been reported in all ages ranging from 1 to 78 years, but the overwhelming majority of cases occur in the fourth and fifth decades of life. Eversole et al [1] found that approximately 50% of mandibular tumors were associated with dental cysts and/or impacted teeth, whereas Brookstone and Huvos [4] reported an association rate of 32% [9]. In children, these tumors are rare, with a sex ratio similar to that in adults and a mandible-to-maxilla ratio of 1:1 [10].

The main symptoms of central MEC of the mandible are swelling and pain, with trismus, paresthesia, and tooth mobility being noted occasionally. The disease is more common in patients with a history of a cyst or impacted tooth, which gives credence to the theory that odontogenic epithelium is capable of giving rise to mucous secretory cells that may undergo neoplastic transformation to MEC. The radiographic features of central MEC of the mandible are usually a well-circumscribed unilocular/multilocular radiolucency; however, these lesions may be initially confused with odontogenic benign and malignant tumors [1,4,11–16].

The t(11;19) translocation and its CRTC1/MAML1 fusion transcript have been identified in MEC at different sites [17-22] and are believed to be associated with the development of a subset of these tumors. To date, the

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status of the fusion transcript has not been reported in intraosseous MEC.

We report 3 cases of central MEC of the mandible, including 1 case with a history of primary retromolar MEC. To assess the involvement of the fusion transcript in central MEC and to determine the relationship between the 2 tumors in 1 patient, we used reverse transcriptase–polymerase chain reaction (RT-PCR) and DNA sequencing analyses to study the microdissected components of these tumors.

2. Materials and methods

A search of the database of the Department of Pathology at The University of Texas MD Anderson Cancer Center from 1998 to 2010 for the diagnosis central MEC yielded 3 tumors that had paraffin-tissue blocks available, and these formed the major materials of our study. Two cases were treated at MD Anderson (one case has been previously reported [23]), and one of the authors (AEN) was a consultant on the third case.

Microdissection, RNA extraction, RT-PCR assay for the CRTC1/MAML2 transcript, and DNA sequencing were done as previously described in detail [18,22].

3. Results

The clinicopathologic features of the retrieved cases are summarized in Table 1.

3.1. Case presentation

Case 3 was a 49-year-old man, who in 2002 was diagnosed with a mucoepidermoid low-grade carcinoma of the retromolar trigone. The patient did not receive any further treatment because it was considered to be a low-grade lesion with complete excision at the time of tooth removal. In August 2009, the patient noticed a painless lump in the angle of his left mandible. A computed tomographic scan indicated cystic expansion of the mandible (Fig. 1). An expansile lytic lesion, 3.5 × 2.2 cm and multiloculated with thin internal septations, was present at the left mandibular angle extending into the body and ramus. The lesion caused cortical thinning, was not associated with a tooth, did not show any mineralization, and was without soft tissue extension. The patient underwent incision and curettage of the left mandibular cystic mass. The pathologic assessment was consistent with a well-differentiated MEC. He underwent a left composite resection, including left hemimandibulectomy, partial resection of adjacent floor of mouth, partial resection of adjacent buccal mucosa, and left selective neck dissection of levels 1 through 3. The patient is free of disease after 5 months.

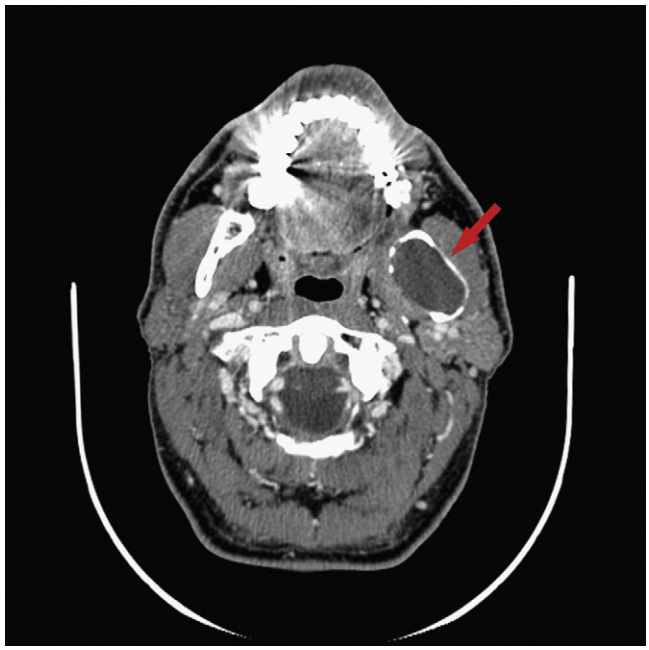

Fig. 1. Computed tomography of the neck with intravenous contrast showing an expansive lesion in the mandible (case 3).

Gross evaluation of cut sections of the bone revealed a 2.8-cm cystic cavity with smooth lining filled with partially hemorrhagic and gelatinous material (Fig. 2). Final histopathologic examination revealed low-grade cystic mucinous adenocarcinoma, which favored the diagnosis of a second primay cystic mucinous adenocarcinoma, probably arising from a glandular odontogenic cyst, although the possibility of multifocal MEC with dominant mucinous component cannot be excluded (Fig. 3A-C).

Case 1 was initially reported as an adenocarcinoma arising in a squamous mucosa-lined cyst wall (radiological impression of dentigerous cyst), with a final diagnosis of

Table 1
Clinicopathologic features of reported cases

Case	Age (y)/Sex	Histology	Location	Cyst/Extraction associated	Treatment	Follow-up
1	18/Male	MEC, low grade	Mandibular body	Yes	Segmental mandibulectomy, suprahyoid neck dissection	Lost to follow-up
2	20/Male	MEC, intermediate grade	Mandibular ramus	Yes	Curettage	NA
3	49/Male	MEC, low grade	Mandibular body	NA	Hemimandibulectomy, neck dissection levels I-III, partial resection of floor of mouth	NED × 3 mo

NA indicates not available; NED, no evidence of disease.

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