

原文題目(出處)：	Central mucoepidermoid carcinoma, a case report with molecular analysis of the TORC1/MAML2 gene fusion Head and Neck Pathol 2010;4:261-4
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內文：

### 1. Abstract

- (1) Mucoepidermoid carcinoma is the most common salivary gland malignancy
  - i. Majorities arise in parotid and minor salivary glands
  - ii. Rarely develop intraosseously
- (2) This case report is an example of intraosseous mucoepidermoid carcinoma with positive TORC1/MAML2 gene fusion transcript and discusses the clinical implications

### 2. Introduction

- (1) Mucoepidermoid carcinoma (MEC) is the most common malignant tumor of salivary glands.
  - i. Intraosseous development is rare: difficult to diagnose and manage.
  - ii. Histogenesis of MEC is uncertain, but an origin from ectopic salivary tissue or metaplastic transformation of odontogenic epithelium has been proposed.
- (2) MEC occurs at relatively young age
  - i. More female than male patients
  - ii. Most common symptoms: pain and swelling
  - iii. Radiologically: unilocular or multilocular well-circumscribed radiolucency

### 3. Case report

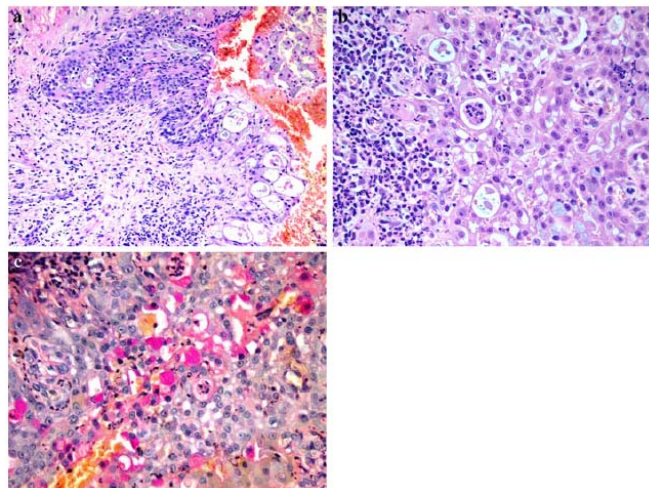
- (1) Patient: 20 year old male with a history of pain, swelling, and discharge in the left mandible of one and a half years duration.
- (2) CT scan: circumscribed intraosseous lytic lesion with solid and cystic components involving ramus and part of the left mandible body



**Fig. 1** Radiologically a circumscribed intraosseous lytic lesion with solid and cystic components involving ramus and part of body of the *left* mandible

- i. Lesion was displacing unerrupted 3<sup>rd</sup> molar → thickening of overlying bone and soft tissue
  - ii. Differential diagnosis (at this stage): odontogenic keratocyst and ameloblastoma
- (3) Incisional biopsy: consisted of multiple hemorrhagic soft tissue fragments
- i. Microscopic examination: epithelial lined cyst wall with infiltrating nests of squamoid and mucin producing cells
  - ii. Presence of mucin was confirmed by mucicarmine stain
  - iii. Cytokeratin AE1/AE3 positively proved epithelial nature of cells
  - iv. Diagnosis of mucoepidermoid carcinoma was made

**Fig. 2** a Low power histologic image showing epithelial lined cyst wall with invasive tumor nest. (H&E stain, 200x). b High power histologic image showing squamoid and mucous cells. (H&E stain, 400x). c High power view, highlighting mucin production by some of the tumor cells. (Mucicarmine stain, 400x)



- (4) RNA was extracted from the tissue
- i. Positive control: RNA from MEC that had been previously identified as having the transcript
  - ii. Negative control: RNA from MEC lacking the transcript
  - iii. Gel electrophoresis was performed and visualized with ethidium bromide and staining under UV light
  - iv. The CRTC1/MAML2 fusion transcript was detected in the mucoepidermoid carcinoma.

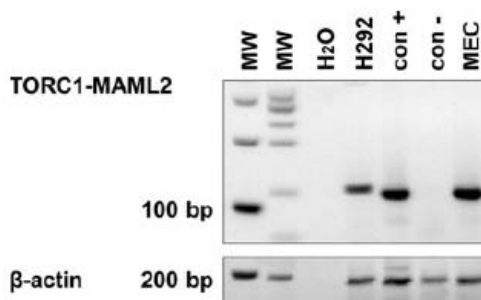


Fig. 3 Nested PCR gel

#### 4. Discussion

- (1) The detection of TORC1/MAML2 fusion gene in this case is the first documentation of this event in these tumors
- (2) Studies of mucoepidermoid carcinomas in salivary glands and at unconventional sites (breast, cervix, lung, thyroid, and skin) have detected the same fusion gene.
- (3) TORC1/MAML2 fusion gene results from: translocation of long arm of chromosome 11 with the long arm of chromosome 19, where exon 1 of the TORC1 gene and exons 2-5 of the MAML2 are linked
  - i. Leads to altered Notch signaling → tumorigenesis
- (4) Recent data suggests that this fusion in MEC is fairly common
  - i. Can help as a diagnostic marker especially in poorly differentiated tumors
- (5) Prognosis of patients with intraosseous MEC is good, especially for patients with radiologically cystic or rarefying type lesion.
- (6) Studies of salivary MEC with fusion positive have also reported a better survival advantage
- (7) Whether similar findings hold true for intraosseous MEC's must await additional cases and studies with long-term follow up.

題號	題目
1	關於 mucoepidermoid carcinoma (MEC), 下列敘述何者為非? (A) MEC 的好發年齡是 10 歲以下的病患 (B) MEC 較常發生在女性 (C) MEC 是最常見的惡性唾液腺腫瘤 (D) MEC 在 parotid gland 最常見
答案(A)	出處：Oral & Maxillofacial Pathology, Second Edition. p.420
題號	題目
2	何者是 intraosseous mucoepidermoid carcinoma 最常見的好發部位? (A) 上顎前牙 (B) 上顎後牙 (C) 下顎前牙 (D) 下顎後牙
答案(D)	出處：Oral & Maxillofacial Pathology, Second Edition. p.422