t(6;22)(p21;q12) in mucoepidermoid carcinoma of the salivary glands

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Published in Atlas Database: January 2010

Online updated version: http://AtlasGeneticsOncology.org/Tumors/t0622p21q12MucoepidID6282.html

DOI: 10.4267/2042/44903

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Clinics and pathology

Disease
Mucoepidermoid carcinoma is the most common type of malignant salivary gland tumor, mostly located to the parotid gland; it is associated with a t(11;19)(q21;p13) translocation with expression of chimeric genes 5' CRTC1 - 3' MAML2 in about half of the cases, mainly associated with a highly or moderately differentiated histology and an excellent outcome.

Epidemiology
One case to date, an 85-year-old male patient; the patient died of an unrelated disease one year after diagnosis (Behboudi et al., 2006; Moller et al., 2008).

Pathology
The mucoepidermoid carcinoma was poorly differentiated.

Genes involved and proteins

POU5F1
Location
6p21
Protein
Homeobox protein (homeodomain in amino acids 230-289 in the 360 aa isoform) with a POU domain (in aa 138-212). Binds the sequence 5'-ATTTCAT-3'. Transcription factor.

EWSR1
Location
22q12
Protein
From N-term to C-term: a transactivation domain (TAD) containing multiple degenerate hexapeptide repeats, 3 arginine/glycine rich domains (RGG regions), a RNA recognition motif, and a RanBP2 type Zinc finger. Role in transcriptional regulation for specific genes and in mRNA splicing.

Result of the chromosomal anomaly

Hybrid Gene
Description
5' EWSR1 - 3' POU5F1. EWSR1 exon 6 is fused in frame to POU5F1 exon 2.

Fusion Protein
Description
Fusion of the N terminal transactivation domain of EWSR1 to the POU and the homebox (DNA binding domain) of POU5F1.

References


Möller E, Stenman G, Mandahl N, Hamberg H, Mölne L, van den Oord JJ, Brosjö O, Mertens F, Panagopoulos I. POU5F1, encoding a key regulator of stem cell pluripotency, is fused to EWSR1 in hidradenoma of the skin and mucoepidermoid carcinoma of the salivary glands. J Pathol. 2008 May;215(1):78-86

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