Head and neck: Pleomorphic salivary gland adenoma with ins(8)(q12;q11q11) (TCEA1-PLAG1)

Julia Asp, Göran Stenman

Molecular Cell Biology and Regenerative Medicine, Department of Clinical Chemistry and Transfusion Medicine, Institute of Biomedicine, Bruna Straket 16, Sahlgrenska University Hospital, 413 45 Goteborg, Sweden.

Published in Atlas Database: April 2007

Online updated version: http://AtlasGeneticsOncology.org/Tumors/SalivAdenTCEA1PLAG1ID5430.html

DOI: 10.4267/2042/16971

This work is licensed under a Creative Commons Attribution-Non-commercial-No Derivative Works 2.0 France Licence.

Clinics and pathology

Disease
Pleomorphic salivary gland adenomas (PA) are benign, slow-growing tumors, which show a remarkable degree of morphological diversity. They constitute the most common form of all salivary gland neoplasms and the majority of the PAs occur in the parotid gland, while the remaining tumors are found in the submandibular and minor salivary glands. Although PAs are benign tumors, subsets of these tumors have a tendency to recur and/or undergo malignant transformation.

Cytogenetics

Genes involved and Proteins

TCEA1 (Transcription elongation factor A 1)

Note: An intronless, ubiquitously expressed pseudogene designated TCEA1P2 or SII is located at 3p22-p21.3.

DNA/RNA
The gene spans about 56 kb and includes 10 exons. Two alternative splicing forms, with and without exon 2, have been detected, yielding transcript sizes of 2784 bp and 2721 bp, respectively.

Protein
The gene codes for two proteins of 301 amino acids (aa) and 280 aa. They contain an N-terminal conserved TFII-S domain, a TFS2M domain, and a C-terminal TFII-S domain.

PLAG1 (Pleomorphic Adenoma Gene 1)

Location: 8q12.1

DNA/RNA
The gene spans about 50 kb and includes 5 exons. The size of the transcript is about 7.3 kb. Two splicing forms of RNA have been found, with or without exon 2.

Protein
500 aa, 74 kDa. The gene encodes a zinc finger protein with two putative nuclear localization signals. It contains a conserved SFP1 domain (aa 58-139), which is a putative transcriptional repressor regulating G2/M transition.

Result of the chromosomal anomaly

Hybride Gene

Note: The fusion occurs as a result of a cryptic, intrachromosomal rearrangement in tumors with apparently normal karyotypes.

Description
The TCEA1-PLAG1 fusion transcript is formed by fusion of exon 1 of TCEA1 to exon 2 or 3 of PLAG1.

Detection protocol
1) RT-PCR using total RNA extracted from frozen tumor tissue. The TCEA1-PLAG1 fusion transcript was amplified by nested RT-PCR using the first round primers SII-UP,
Head and neck: Pleomorphic salivary gland adenoma with ins(8)(q12;q11q11) (TCEA1-PLAG1)  

Asp J, Stenman G

Map of the 8q11.2-q12 region including the PLAG1 and TCEA1 genes (not drawn to scale). Exons are shown as boxes and the start and stop codons are shown as asterisks and arrowheads, respectively. Reprinted partially from publication CHCHD7-PLAG1 and TCEA1-PLAG1 gene fusions resulting from cryptic, intrachromosomal 8q rearrangements in pleomorphic salivary gland adenomas, Genes Chromosomes Cancer, Vol. 45, No. 9, 2006, 820-828. Copyright 2006 Wiley-Liss, Inc. Reprinted with permission of Wiley-Liss, Inc.

Fusion Protein

Expression Localisation

Nucleus.

References


This article should be referenced as such:  