

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

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

Cytogenetics Molecular *ins(8)(q12.1q11.23q11.23)*.

Genes involved and Proteins

Gene Name [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, has 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 TFIIIS-I domain, a TFS2M domain, and a C-terminal TFIIIS domain.

Gene Name [PLAG1 \(Pleomorphic Adenoma Gene 1\)](#)

Location 8q12.1

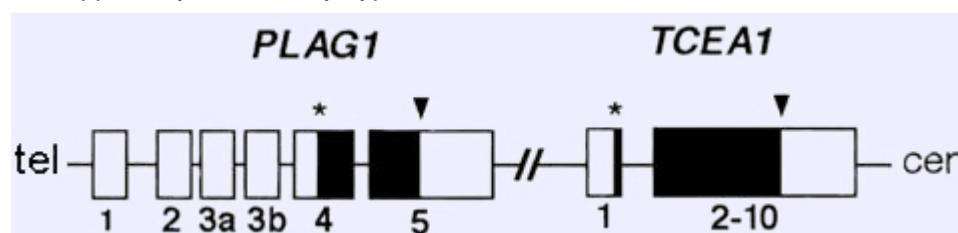
Dna / Rna The gene spans about 50 kb and includes 5 exons. The size of the transcript is about 7.3 kb. Two splicing forms of RNA have been found, with or without exon 2.

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

Hybrid Gene

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



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.

- Description The TCEA1-PLAG1 fusion transcript is formed by fusion of exon 1 of TCEA1 to exon 2 or 3 of PLAG1.
- Detection 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, 5'-CATGCGGTGGTGGGGTTGCT-3', and MV5, 5'-CAGGAGAATGAGTAGCCATGTGC-3', and the second round primers S2-764S, 5'-GGGGTCGCTCCTGCTGTGTCT3' and MV6, 5'-TGCACCTGTAGGGCCTCTCTCCTG-3'. Fusion transcripts of 557 bp and 662 bp were detected.
- 2) Dual-color FISH and fiber-FISH on metaphase chromosomes using the BAC clones RP11-140I16 (PLAG1) and RP11-410P17 (TCEA1) as probes.

Fusion Protein

Expression Localisation Nucleus.

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