

Gene Section

Short Communication

AKAP9 (A kinase (PRKA) anchor protein (yotiao) 9)

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Identity

Hugo: AKAP9

Other names: AKAP350; AKAP450; CG-NAP; HYPERION; KIAA0803; MU-RMS-40.16A; PRKA9; YOTIAO

Location: 7q21.2

DNA/RNA

Description

AKAP9 gene consists of 51 encoding exons with a total gene size of 169797 bp.

Transcription

The AKAP9 transcript has an open reading frame (ORF) of 11724 bp. Four different transcript variants have been identified (NM147171, NM005751, NM147185, NM147166).

Protein

Description

The 11.7 kb ORF encodes for a 3908 aa protein with a predicted size of 453 kDa. Four different protein products have been described (NP005742.4, NP671695.1, NP671700.1, NP671714.1).

Localisation

Centrosomes, Golgi apparatus.

Function

AKAP9 belongs to the family of A-Kinase Anchor Proteins (AKAPs), which are scaffold proteins able to bind the type II regulatory subunit (RII) of cAMP dependent Protein Kinase A (PKA) and several other protein kinases and phosphatases and to anchor them to specific intracellular compartments. AKAP9 shows

centrosome and Golgi compartmentalization. Various splice variants of the transcript are found in different human tissues. The AKAP9 protein contains the PKA binding domain, a large coiled-coil domain and C-terminal centrosome binding domain.

Implicated in

Papillary Thyroid Carcinoma (PTC)

Cytogenetics

inv(7)(q21-22q34) with AKAP9-BRAF fusion.

Breakpoints

Note: Breakpoint in AKAP9-BRAF fusion is located within intron 8 of the gene. In this fusion, exons 1-8 of AKAP9 are fused with last 10 exons 9-18 of BRAF. In the fusion, AKAP9 lacks the centrosome binding domain and, as a result, the AKAP9-BRAF protein loses its cytoplasmic compartmentalization and appears to be diffusely distributed in the cytoplasm, as detected by immunofluorescence.

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