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Gene Section
Short Communication

ASPSCR1 (alveolar soft part sarcoma critical region 1)

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Identity

Other names: ASPL (Alveolar soft part sarcoma locus)

HGNC (Hugo): ASPSCR1

Location: 17q25

Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

DNA/RNA

Description

1872 bp cDNA.

Transcription

Alternative splicing of 47 bp from exon 2 in the 5' untranslated region.

Protein

Description

476 amino acids; contains an UBX domain, which may be related to the ubiquitylation pathway.

Expression

Widely expressed in the adult; low expression in fetal tissues.

Implicated in

Alveolar soft part sarcoma with ASPSCR1 -TFE3 fusion

Cytogenetics

der(X)t(X;17)(p11;q25) is consistently involved; it implicates: 1- the formation of a hybrid gene at the breakpoint, and also, 2- gain in Xp11-pter sequences, and loss of heterozygocity in 11q25-qter, with possible implications.

Hybrid/Mutated gene

5' ASPSCR1-3' TFE3; the reciprocal 5' TFE3 - 3' ASPSCR1 is most often absent. ASPSCR1 is fused in frame to TFE3 exon 3 or 4.

Abnormal protein

NH2 term ASPSCR1, fused to the C term of TFE3.

Oncogenesis

Might combine the effect of a fusion protein to that of gene(s) dosage.

Primary renal ASPSCR1-TFE3 tumour

Disease

A subset of renal cell carcinoma, which presents with a combination of alveolar soft part sarcoma-like features and epithelial features is found to carry this anomaly.

Cytogenetics

Balanced t(X;17)(p11.2;q25), in contrast with what is found in the alveolar soft part sarcoma (see above).

Hybrid/Mutated gene

5' ASPSCR1-3' TFE3.

Abnormal protein

NH2 term ASPSCR1, fused to the C term of TFE3.

References

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