

ASPSCR1 (Alveolar soft part sarcoma critical region 1)

Identity

Other names **ASPL (Alveolar soft part sarcoma locus)**
 Hugo [ASPSCR1](#)
 Location 17q25

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

Entity [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 heterozygosity 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

Entity [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

External links

Nomenclature
[Hugo](#) [ASPSCR1](#)
[GDB](#) [ASPSCR1](#)
[Entrez Gene](#) [ASPSCR1_79058](#) alveolar soft part sarcoma chromosome region, candidate 1

Cards

Atlas	ASPSCR1ID358
GeneCards	ASPSCR1
Ensembl	ASPSCR1
CancerGene	ASPSCR1
Genatlas	ASPSCR1
GeneLynx	ASPSCR1
eGenome	ASPSCR1
euGene	79058
	Genomic and cartography
GoldenPath	ASPSCR1 - 17q25 chr17:77528715-77568569 + 17q25.3 (hg17-May_2004)
Ensembl	ASPSCR1 - 17q25.3 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	ASPSCR1
	Gene and transcription
Genbank	AF324219 [SRS] AF324219 [ENTREZ]
Genbank	AK057403 [SRS] AK057403 [ENTREZ]
Genbank	AK057851 [SRS] AK057851 [ENTREZ]
Genbank	BC006152 [SRS] BC006152 [ENTREZ]
Genbank	BC018722 [SRS] BC018722 [ENTREZ]
RefSeq	NM_024083 [SRS] NM_024083 [ENTREZ]
RefSeq	NT_086886 [SRS] NT_086886 [ENTREZ]
AceView	ASPSCR1 AceView - NCBI
TRASER	ASPSCR1 Traser - Stanford
Unigene	Hs.298351 [SRS] Hs.298351 [NCBI] HS298351 [spliceNest]
	Protein : pattern, domain, 3D structure
SwissProt	Q96M40 [SRS] Q96M40 [EXPASY] Q96M40 [INTERPRO]
Prositate	PS50033 UBX [SRS] PS50033 UBX [Expasy]
Interpro	IPR001012 UBX [SRS] IPR001012 UBX [EBI]
CluSTr	Q96M40
Pfam	PF00789 UBX [SRS] PF00789 UBX [Sanger] pfam00789 [NCBI-CDD]
Blocks	Q96M40
	Polymorphism : SNP, mutations, diseases
OMIM	606236 [map]
GENECLINICS	606236
SNP	ASPSCR1 [dbSNP-NCBI]
SNP	NM_024083 [SNP-NCI]
SNP	ASPSCR1 [GeneSNPs - Utah] ASPSCR1 [SNP - CSHL] ASPSCR1 [HGBASE - SRS]
	General knowledge
Family Browser	ASPSCR1 [UCSC Family Browser]
SOURCE	NM_024083
SMD	Hs.298351
SAGE	Hs.298351
Amigo	process biological_process unknown
Amigo	component cellular_component unknown
Amigo	function molecular_function unknown
PubGene	ASPSCR1
	Other databases
	Probes

[Probe](#) [Cancer Cytogenetics \(Bari\)](#)
[Probe](#) [ASPSCR1 Related clones \(RZPD - Berlin\)](#)

[PubMed](#)

[PubMed](#) [6 Pubmed reference\(s\) in LocusLink](#)

Bibliography

The der(17)t(X.17)(p11;q25) of human alveolar soft part sarcoma fuses the TFE3 transcription factor gene to ASPL, a novel gene at 17q25.

Ladanyi M, Lui MY, Antonescu CR, Krause-Boehm A, Meindl A, Argani P, Healey JH, Ueda T, Yoshikawa H, Meloni-Ehrig A, Sorensen PHB, Mertens F, Mandahl N, van den Berghe H, Sciot R, dal Cin P, Bridge J.

Oncogene 2001; 20: 48-57.

Medline [21140288](#)

Primary renal neoplasms with the ASPL-TFE3 gene fusion of alveolar soft part sarcoma: a distinctive tumor entity previously included among renal cell carcinomas of children and adolescents.

Argani P, Antonescu CR, Illei PB, Lui MY, Timmons CF, Newbury R, Reuter VE, Garvin AJ, Perez-Atayde AR, Fletcher JA, Beckwith JB, Bridge JA, Ladanyi M.

Am J Pathol 2001; 159: 179-192.

Medline [21331068](#)

[REVIEW articles](#) *automatic search in PubMed*

[Last year publications](#) *automatic search in PubMed*

[BiblioGene - INIST](#)

Contributor(s)

Written 08-2001 Jean-Loup Huret

Citation

This paper should be referenced as such :

Huret JL . ASPSCR1 (Alveolar soft part sarcoma critical region 1). Atlas Genet Cytogenet Oncol Haematol. August 2001 .

URL : <http://www.infobiogen.fr/services/chromcancer/Genes/ASPSCR1ID358.html>

© Atlas of Genetics and Cytogenetics in Oncology and Haematology
