

## SS18L1 (synovial sarcoma translocation gene on chromosome 18-like 1)

### Identity

Other names	<b>CREST</b> <b>KIAA0693</b> <b>MGC26711</b> <b>SYT homolog 1</b>
Hugo	<b><u>SS18L1</u></b>
Location	20q13.3 chr20:60,152,245-60,190,935 (UCSC latest release: May 2004)

### DNA/RNA

Note	Member of the SS18 family.
Description	11 exons with similar splice sites as SS18. The promoter region lacks CAAT and TATA boxes but contains CpG islands, suggesting that SS18L1 is a housekeeping gene.
Pseudogene	SS18L2 (3p21)

### Protein

Description	396 amino acids, 42990 Da. The SS18L1 protein, similarly to the SS18 protein, exhibits two domains: a SYT N-terminal homology domain found in a wide variety of species ranging from plants to humans and the QPGY domain at the COOH-terminal part, rich in glutamine, proline, glycine, and tyrosine. The QPGY domain of the SS18 protein may activate transcription when coupled to a DNA-binding domain.
Expression	Ubiquitous; with lowest levels in spleen.
Localisation	Nuclear?
Function	Calcium-responsive transactivator: CREST is a SYT -related nuclear protein that interacts with <a href="#">CREB-binding protein (CBP)</a> and is expressed in the developing brain.
Homology	SS18, SS18L2

### Implicated in

Disease	<a href="#">Synovial sarcoma</a>
Prognosis	Unknown
Cytogenetics	t(X;20)(p11;q13.3)

Hybrid/Mutated Gene In the SS18L1/ SSX1 transcript detected in the synovial sarcoma, the exon 10 of SS18L1, which corresponds to exon 10 of SS18, was fused to exon 6 of SSX1

Abnormal Protein In the putative SS18L1/SSX1 chimeric protein, the last 8 amino acid residues of the SS18L1 protein are replaced by 78 amino acids from the COOH-terminal part of SSX1. By analogy with what is presumed to be the case for the SS18/SSX fusion protein, SS18L1/SSX1 is likely to show an altered transcriptional pattern with the COOH-terminal SSX domain, redirecting the SS18L1 activation domain to new target promoters.

## External links

### Nomenclature

[Hugo](#) [SS18L1](#)  
[GDB](#) [SS18L1](#)  
[Entrez\\_Gene](#) [SS18L1\\_26039](#) synovial sarcoma translocation gene on chromosome 18-like 1

### Cards

[GeneCards](#) [SS18L1](#)  
[Ensembl](#) [SS18L1](#)  
[CancerGene](#) [SS18L1](#)  
[Genatlas](#) [SS18L1](#)  
[GeneLynx](#) [SS18L1](#)  
[eGenome](#) [SS18L1](#)  
[euGene](#) [26039](#)

### Genomic and cartography

[GoldenPath](#) [SS18L1](#) - [20q13.3](#) [chr20:60152217-60190961](#) + [20q13.33](#) (hg17-May\_2004)  
[Ensembl](#) [SS18L1 - 20q13.33 \[CytoView\]](#)  
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]  
[OMIM](#) [Disease map \[OMIM\]](#)  
[HomoloGene](#) [SS18L1](#)

### Gene and transcription

[Genbank](#) [AL078633](#) [SRS] [AL078633](#) [ENTREZ]  
[Genbank](#) [AB014593](#) [SRS] [AB014593](#) [ENTREZ]  
[Genbank](#) [AK125656](#) [SRS] [AK125656](#) [ENTREZ]  
[Genbank](#) [AY203931](#) [SRS] [AY203931](#) [ENTREZ]  
[Genbank](#) [BC034494](#) [SRS] [BC034494](#) [ENTREZ]  
[RefSeq](#) [NM\\_015558](#) [SRS] [NM\\_015558](#) [ENTREZ]  
[RefSeq](#) [NM\\_198935](#) [SRS] [NM\\_198935](#) [ENTREZ]

<a href="#">RefSeq</a>	<a href="#">NT_086910</a> [SRS] <a href="#">NT_086910</a> [ENTREZ]
<a href="#">AceView</a>	<a href="#">SS18L1</a> AceView - NCBI
<a href="#">TRASER</a>	<a href="#">SS18L1</a> Traser - Stanford
<a href="#">Unigene</a>	<a href="#">Hs.154429</a> [SRS] <a href="#">Hs.154429</a> [NCBI] <a href="#">HS154429</a> [spliceNest]

### Protein : pattern, domain, 3D structure

<a href="#">SwissProt</a>	<a href="#">O75177</a> [SRS] <a href="#">O75177</a> [EXPASY] <a href="#">O75177</a> [INTERPRO]
<a href="#">Interpro</a>	<a href="#">IPR007726 SSXT</a> [SRS] <a href="#">IPR007726 SSXT</a> [EBI]
<a href="#">CluSTr</a>	<a href="#">O75177</a>
<a href="#">Pfam</a>	<a href="#">PF05030 SSXT</a> [SRS] <a href="#">PF05030 SSXT</a> [Sanger] <a href="#">pfam05030</a> [NCBI-CDD]
<a href="#">Blocks</a>	<a href="#">O75177</a>

### Polymorphism : SNP, mutations, diseases

<a href="#">OMIM</a>	<a href="#">606472</a> [ <a href="#">map</a> ]
<a href="#">GENECLINICS</a>	<a href="#">606472</a>
<a href="#">SNP</a>	<a href="#">SS18L1</a> [dbSNP-NCBI]
<a href="#">SNP</a>	<a href="#">NM_015558</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">NM_198935</a> [SNP-NCI]
<a href="#">SNP</a>	<a href="#">SS18L1</a> [GeneSNPs - Utah] <a href="#">SS18L1</a> [SNP - CSHL] <a href="#">SS18L1</a> [HGBASE - SRS]

### General knowledge

<a href="#">Family Browser</a>	<a href="#">SS18L1</a> [UCSC Family Browser]
<a href="#">SOURCE</a>	<a href="#">NM_015558</a>
<a href="#">SOURCE</a>	<a href="#">NM_198935</a>
<a href="#">SMD</a>	<a href="#">Hs.154429</a>
<a href="#">SAGE</a>	<a href="#">Hs.154429</a>
<a href="#">PubGene</a>	<a href="#">SS18L1</a>

### Other databases

### Probes

<a href="#">Probe</a>	<a href="#">SS18L1 Related clones (RZPD - Berlin)</a>
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### PubMed

<a href="#">PubMed</a>	<a href="#">6 Pubmed reference(s) in LocusLink</a>
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Ishikawa K, Nagase T, Suyama M, Miyajima N, Tanaka A, Kotani H, Nomura N, Ohara O.

DNA Res 1998; 5: 169-176.

Medline [9734811](#)

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### **human SS18-like genes and a mouse Ss18 pseudogene.**

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Medline [11435705](#)

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Proc Natl Acad Sci U S A 2002; 9: 16899-16903. Epub 2002 Dec 11.

Medline [12477932](#)

### **A novel fusion gene, SS18L1/SSX1, in synovial sarcoma.**

Storlazzi CT, Mertens F, Mandahl N, Gisselsson D, Isaksson M, Gustafson P, Domanski HA, Panagopoulos I.

Genes Chromosomes Cancer 2003; 37: 195-200.

Medline [12696068](#)

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Science 2004; 303: 197-202.

Medline [14716005](#)

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                 2005      Panagopoulos

### **Citation**

*This paper should be referenced as such :*

**Storlazzi CT, Mertens F, Panagopoulos I .** SS18L1 (synovial sarcoma translocation gene on chromosome 18-like 1). Atlas Genet Cytoenet Oncol Haematol. March

2005 .

URL :

<http://www.infobiogen.fr/services/chromcancer/Genes/SS18L1D474ch20q13.html>

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