

Gene Section

Short Communication

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

Clelia Tiziana Storlazzi, Fredrik Mertens, Ioannis Panagopoulos

Department of Genetics and Microbiology, University of Bari, Bari, Italy (CTS); Department of Clinical Genetics, Lund University Hospital, Lund, Sweden (FM, IP)

Published in Atlas Database: March 2005

Online updated version: <http://AtlasGeneticsOncology.org/Genes/SS18L1D474ch20q13.html>

DOI: 10.4267/2042/38186

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2005 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Identity

Other names: CREST; KIAA0693; MGC26711; SYT homolog 1

HGNC (Hugo): SS18L1

Location: 20q13.3

Local order: 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 CREB-binding protein (CBP) and is expressed in the developing brain.

Homology

SS18, SS18L2.

Implicated in

Disease

Synovial sarcoma.

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.

References

Ishikawa K, Nagase T, Suyama M, Miyajima N, Tanaka A, Kotani H, Nomura N, Ohara O. Prediction of the coding sequences of unidentified human genes. X. The complete sequences of 100 new cDNA clones from brain which can code for large proteins in vitro. *DNA Res.* 1998 Jun 30;5(3):169-76

de Bruijn DR, Kater-Baats E, Eleveld M, Merx G, Geurts Van Kessel A. Mapping and characterization of the mouse and human SS18 genes, two human SS18-like genes and a mouse Ss18 pseudogene. *Cytogenet Cell Genet.* 2001; 92(3-4):310-9

Strausberg RL, Feingold EA, Grouse LH, Derge JG, Klausner RD, Collins FS, Wagner L, Shenmen CM, Schuler GD, Altschul SF, Zeeberg B, Buetow KH, Schaefer CF, Bhat NK, Hopkins RF, Jordan H, Moore T, Max SI, Wang J, Hsieh F, Diatchenko L, Marusina K, Farmer AA, Rubin GM, Hong L, Stapleton M, Soares MB, Bonaldo MF, Casavant TL, Scheetz TE, Brownstein MJ, Usdin TB, Toshiyuki S, Carninci P, Prange C, Raha SS, Loquellano NA, Peters GJ, Abramson RD, Mullahy SJ, Bosak SA, McEwan PJ, McKernan KJ, Malek JA,

Gunaratne PH, Richards S, Worley KC, Hale S, Garcia AM, Gay LJ, Hulyk SW, Villalon DK, Muzny DM, Sodergren EJ, Lu X, Gibbs RA, Fahey J, Helton E, Kettelman M, Madan A, Rodrigues S, Sanchez A, Whiting M, Madan A, Young AC, Shevchenko Y, Bouffard GG, Blakesley RW, Touchman JW, Green ED, Dickson MC, Rodriguez AC, Grimwood J, Schmutz J, Myers RM, Butterfield YS, Krzywinski MI, Skalska U, Smailus DE, Schnerch A, Schein JE, Jones SJ, Marra MA. Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences. *Proc Natl Acad Sci U S A.* 2002 Dec 24;99(26):16899-903

Storlazzi CT, Mertens F, Mandahl N, Gisselsson D, Isaksson M, Gustafson P, Domanski HA, Panagopoulos I. A novel fusion gene, SS18L1/SSX1, in synovial sarcoma. *Genes Chromosomes Cancer.* 2003 Jun;37(2):195-200

Aizawa H, Hu SC, Bobb K, Balakrishnan K, Ince G, Gurevich I, Cowan M, Ghosh A. Dendrite development regulated by CREST, a calcium-regulated transcriptional activator. *Science.* 2004 Jan 9;303(5655):197-202

This article should be referenced as such:

Storlazzi CT, Mertens F, Panagopoulos I. SS18L1 (synovial sarcoma translocation gene on chromosome 18-like 1). *Atlas Genet Cytogenet Oncol Haematol.* 2005; 9(2):143-144.
