

SET (SET translocation (myeloid leukemia-associated))

Identity

Other names	I2PP2A, I-2PP2A (Phosphatase 2A inhibitor) TAF-IBETA (Template activating factor I) PHAPII (HLA-DR associated protein II) IGAAD (Inhibitor of granzyme A-activated Dnase)
Hugo	<u>SET</u>
Location	9q34 from centromere to telomere: SET, ABL1 , NUP214 (alias CAN), NOTCH1 (alias TAN1)

DNA/RNA

Description	SET encompasses 6.81 kb on the genomic DNA; 8 exons
Transcription	2577 bp mRNA

Protein

Description	two isoforms; isoform 1 (TAF1 alpha) 290 amino acids, 33.5 kDa; isoform 2 (TAF1 beta) - 277 amino acids, 23.1 kDa
Expression	widely expressed; highly expressed in Wilms' tumor
Localisation	cytoplasmic and nuclear; in the cytoplasm, found both in the cytosol and associated with the endoplasmic reticulum
Function	multitasking protein, involved in apoptosis, transcription, nucleosome assembly, and histone binding; potent inhibitor of protein phosphatase 2A; inhibits also EP300/CREBBP and PCAF-mediated acetylation of histones (HAT) - predominantly H4 - and nucleosomes; HAT inhibition leads to silencing of HAT-dependent transcription and prevents active demethylation of DNA
Homology	belongs to the nucleosome assembly protein (NAP) family

Implicated in

Entity	t(9;9)(q34;q34) --> SET- NUP214 (alias CAN)
Disease	acute undifferentiated leukemia (AUL); only one case described so far
Cytogenetics	normal karyotype; may be overlooked
Hybrid/Mutated	5' SET - 3' NUP214

Gene

Abnormal Protein the SET-NUP214 (alias CAN) fusion protein consists of almost the whole SET protein fused to the C-terminus of NUP214

Oncogenesis SET-NUP214 leads to disorganization of nuclear export

External links

Nomenclature

[Hugo](#) [SET](#)

[GDB](#) [SET](#)

[Entrez Gene](#) [SET 6418](#) SET translocation (myeloid leukemia-associated)

Cards

[GeneCards](#) [SET](#)

[Ensembl](#) [SET](#)

[CancerGene](#) [SET](#)

[Genatlas](#) [SET](#)

[GeneLynx](#) [SET](#)

[eGenome](#) [SET](#)

[euGene](#) [6418](#)

Genomic and cartography

[GoldenPath](#) [SET - 9q34 chr9:128531415-128538221 + 9q34.11](#) (hg17-May_2004)

[Ensembl](#) [SET - 9q34.11 \[CytoView\]](#)

[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#) [Disease map \[OMIM\]](#)

[HomoloGene](#) [SET](#)

Gene and transcription

[Genbank](#) [AL356481](#) [SRS] [AL356481](#) [ENTREZ]

[Genbank](#) [AY349172](#) [SRS] [AY349172](#) [ENTREZ]

[Genbank](#) [BC032749](#) [SRS] [BC032749](#) [ENTREZ]

[Genbank](#) [CR536543](#) [SRS] [CR536543](#) [ENTREZ]

[Genbank](#) [CR542050](#) [SRS] [CR542050](#) [ENTREZ]

[RefSeq](#) [NM_003011](#) [SRS] [NM_003011](#) [ENTREZ]

[RefSeq](#) [NT_086756](#) [SRS] [NT_086756](#) [ENTREZ]

[AceView](#) [SET](#) AceView - NCBI

[TRASER](#) [SET](#) Traser - Stanford

[Unigene](#) [Hs.436687](#) [SRS] [Hs.436687](#) [NCBI] [HS436687](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [Q01105](#) [SRS] [Q01105](#) [EXPASY] [Q01105](#) [INTERPRO]

[Interpro](#) [IPR002164 NAP family](#) [SRS] [IPR002164 NAP family](#) [EBI]
[CluSTr](#) [Q01105](#)
[Pfam](#) [PF00956 NAP](#) [SRS] [PF00956 NAP](#) [Sanger] [pfam00956](#) [NCBI-CDD]
[Blocks](#) [Q01105](#)

Polymorphism : SNP, mutations, diseases

[OMIM](#) [600960](#) [[map](#)]
[GENECLINICS](#) [600960](#)
[SNP](#) [SET](#) [dbSNP-NCBI]
[SNP](#) [NM_003011](#) [SNP-NCI]
[SNP](#) [SET](#) [GeneSNPs - Utah] [SET](#) [SNP - CSHL] [SET1](#) [HGBASE - SRS]

General knowledge

[Family Browser](#) [SET](#) [UCSC Family Browser]
[SOURCE](#) [NM_003011](#)
[SMD](#) [Hs.436687](#)
[SAGE](#) [Hs.436687](#)
[Amigo](#) [process|DNA replication](#)
[Amigo](#) [component|endoplasmic reticulum](#)
[Amigo](#) [function|histone binding](#)
[Amigo](#) [process|negative regulation of histone acetylation](#)
[Amigo](#) [process|nucleocytoplasmic transport](#)
[Amigo](#) [process|nucleosome assembly](#)
[Amigo](#) [process|nucleosome disassembly](#)
[Amigo](#) [component|nucleus](#)
[Amigo](#) [component|perinuclear region](#)
[Amigo](#) [function|protein phosphatase inhibitor activity](#)
[Amigo](#) [function|protein phosphatase type 2A regulator activity](#)
[BIOCARTA](#) [Granzyme A mediated Apoptosis Pathway](#)
[PubGene](#) [SET](#)

Other databases

Probes

[Probe](#) [SET Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [16 Pubmed reference\(s\) in LocusLink](#)

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Medline [1630450](#)

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Medline [8131851](#)

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Medline [15239126](#)

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Written 08- Sabine Strehl
 2005

Citation

This paper should be referenced as such :

Strehl S . SET (SET translocation (myeloid leukemia-associated)). Atlas Genet Cytogenet Oncol Haematol. August 2005 .

URL :

<http://www.infobiogen.fr/services/chromcancer/Genes/SETID42272ch9q34.html>

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