

NSD1 (Nuclear receptor-binding, su(var), enhancer-of-zeste and trithorax domain-containing protein 1

Identity

Hugo [NSD1](#)

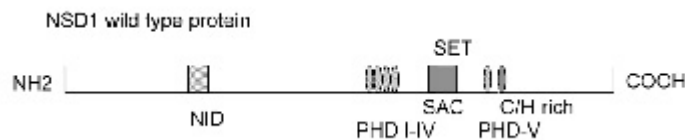
Location 5q35

DNA/RNA

Description At least 23 exons. cDNA is 8552 bp, 8088 bp open reading frame

Transcription Two transcripts: 9.0 and 10 kb

Protein



Description 2696 amino acids. Murine Nsd1 is a nuclear protein containing SET, proline-tryptophan-tryptophan-proline (PWWP) and plant homeodomain protein (PHD) finger domains. The protein has two distinct nuclear receptor (NR)-interaction domains (NID-L, NID+L). Human NSD1 shows 86% identity to the murine Nsd1 at the nucleotide level and 83% at the amino acid level, retaining the nuclear interaction domains (NID) as well as the SET/SAC and PHD finger domains.

Expression Widely expressed

Function Features of a basic transcription factor, also of a bifunctional transcriptional regulator, (similar to murine Nsd1)

Homology NSD2: (Wolf-Hirschhorn syndrome critical region on 4p); NSD3: expressed in tumour cell lines

Implicated in

Entity [t\(5;11\)\(q35;p15.5\)](#)/ acute non lymphoblastic leukemia (ANLL)

Disease De novo childhood ANLL

Prognosis Only 5 cases reported. All had poor response to treatment/short survival

Cytogenetics Cryptic: associated with [del\(5q\)](#) (sole cytogenetic abnormality) or a normal karyotype

Hybrid/Mutated Gene 5' NUP98- 3' NSD1 ; NSD1-NUP98 also present in all cases tested

Abnormal NH2 NUP98- COOH NSD1: Fuses the FXFG portion of NUP98 to

Protein the SET, SAC and PHD finger domains of NSD1. NSD1-NUP98:
Fuses the RNA-binding domain of NSD1 to the NID domain NUP98

External links

Nomenclature

[Hugo](#) [NSD1](#)
[GDB](#) [NSD1](#)
[Entrez Gene](#) [NSD1_64324](#) nuclear receptor binding SET domain protein 1

Cards

[Atlas](#) [NSD1ID356](#)
[GeneCards](#) [NSD1](#)
[Ensembl](#) [NSD1](#)
[CancerGene](#) [ARA267](#)
[Genatlas](#) [NSD1](#)
[GeneLynx](#) [NSD1](#)
[eGenome](#) [NSD1](#)
[euGene](#) [64324](#)

Genomic and cartography

[GoldenPath](#) [NSD1 - 5q35](#) [chr5:176493532-176655367 + 5q35.2](#) (hg17-May_2004)
[Ensembl](#) [NSD1 - 5q35.2 \[CytoView\]](#)
[NCBI](#) [Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]
[OMIM](#) [Disease map \[OMIM\]](#)
[HomoloGene](#) [NSD1](#)

Gene and transcription

[Genbank](#) [AF085858](#) [SRS] [AF085858](#) [ENTREZ]
[Genbank](#) [AF322907](#) [SRS] [AF322907](#) [ENTREZ]
[Genbank](#) [AF380302](#) [SRS] [AF380302](#) [ENTREZ]
[Genbank](#) [AF395588](#) [SRS] [AF395588](#) [ENTREZ]
[Genbank](#) [AK025916](#) [SRS] [AK025916](#) [ENTREZ]
[RefSeq](#) [NM_022455](#) [SRS] [NM_022455](#) [ENTREZ]
[RefSeq](#) [NM_172349](#) [SRS] [NM_172349](#) [ENTREZ]
[RefSeq](#) [NT_086683](#) [SRS] [NT_086683](#) [ENTREZ]
[AceView](#) [NSD1](#) AceView - NCBI
[TRASER](#) [NSD1](#) Traser - Stanford
[Unigene](#) [Hs.208961](#) [SRS] [Hs.208961](#) [NCBI] [HS208961](#) [spliceNest]

Protein : pattern, domain, 3D structure

[SwissProt](#) [Q96L73](#) [SRS] [Q96L73](#) [EXPASY] [Q96L73](#) [INTERPRO]

Prosite	PS50868 POST SET [SRS]	PS50868 POST SET [Expasy]	
Prosite	PS50812 PWWP [SRS]	PS50812 PWWP [Expasy]	
Prosite	PS50280 SET [SRS]	PS50280 SET [Expasy]	
Prosite	PS01359 ZF PHD 1 [SRS]	PS01359 ZF PHD 1 [Expasy]	
Prosite	PS50016 ZF PHD 2 [SRS]	PS50016 ZF PHD 2 [Expasy]	
Interpro	IPR006560 AWS [SRS]	IPR006560 AWS [EBI]	
Interpro	IPR003616 PostSET [SRS]	IPR003616 PostSET [EBI]	
Interpro	IPR000313 PWWP [SRS]	IPR000313 PWWP [EBI]	
Interpro	IPR001214 SET [SRS]	IPR001214 SET [EBI]	
Interpro	IPR001965 Znf PHD [SRS]	IPR001965 Znf PHD [EBI]	
Interpro	IPR001841 Znf ring [SRS]	IPR001841 Znf ring [EBI]	
CluSTr	Q96L73		
Pfam	PF00628 PHD [SRS]	PF00628 PHD [Sanger]	pfam00628 [NCBI-CDD]
Pfam	PF00855 PWWP [SRS]	PF00855 PWWP [Sanger]	pfam00855 [NCBI-CDD]
Pfam	PF00856 SET [SRS]	PF00856 SET [Sanger]	pfam00856 [NCBI-CDD]
Smart	SM00570 AWS [EMBL]		
Smart	SM00249 PHD [EMBL]		
Smart	SM00508 PostSET [EMBL]		
Smart	SM00293 PWWP [EMBL]		
Smart	SM00184 RING [EMBL]		
Smart	SM00317 SET [EMBL]		
Blocks	Q96L73		

Polymorphism : SNP, mutations, diseases

OMIM	606681 [map]		
GENECLINICS	606681		
SNP	NSD1 [dbSNP-NCBI]		
SNP	NM_022455 [SNP-NCI]		
SNP	NM_172349 [SNP-NCI]		
SNP	NSD1 [GeneSNPs - Utah]	NSD1 [SNP - CSHL]	NSD1 [HGBASE - SRS]

General knowledge

Family Browser	NSD1 [UCSC Family Browser]
SOURCE	NM_022455
SOURCE	NM_172349
SMD	Hs.208961
SAGE	Hs.208961
Amigo	function DNA binding
Amigo	component nucleus

[Amigo](#) [process|protein ubiquitination](#)
[Amigo](#) [process|regulation of transcription, DNA-dependent](#)
[Amigo](#) [component|ubiquitin ligase complex](#)
[Amigo](#) [function|ubiquitin-protein ligase activity](#)
[Amigo](#) [function|zinc ion binding](#)
[PubGene](#) [NSD1](#)

Other databases

Probes

[Probe](#) [HSP2301A4](#)
[Probe](#) [NSD1 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [13 Pubmed reference\(s\) in LocusLink](#)

Bibliography

Two distinct nuclear receptor interaction domains in NSD1, a novel SET protein that exhibits characteristics of both corepressors and coactivators.

Huang N, vom Baur E, Garnier JM, Lerouge T, Vonesch JL, Lutz Y, Chambon P, Losson R.

EMBO J 1998; 17(12): 3398-3412.

Medline [9628876](#)

WHSC1, a 90 kb SET domain-containing gene, expressed in early development and homologous to a Drosophila dysmorphy gene maps in the Wolf-Hirschhorn syndrome critical region and is fused to IgH in t(4;14) multiple myeloma.

Stec I, Wright TJ, van Ommen GJ, de Boer PA, van Haeringen A, Moorman AF, Altherr MR, den Dunnen JT.

Hum Mol Genet 1998; 7(7): 1071-1082.

Medline [9618163](#)

A new recurrent translocation, t(5;11)(q35;p15.5), associated with del(5q) in childhood acute myeloid leukemia.

Jaju RJ, Haas OA, Neat M, Harbott J, Saha V, Boulwood J, Brown JM, Pirc-Danoewinata H, Krings BW, Muller U, Morris, SW, Wainscoat JS, Kearney L.

Blood 1999; 94(2): 773-780.

Medline [10397745](#)

NSD3, a new SET domain-containing gene, maps to 8p12 and is amplified in human breast cancer cell lines.

Angrand PO, Apiou F, Stewart AF, Dutrillaux B, Losson R, Chambon P.

Genomics 2001; 74(1): 79-88.

Medline [11374904](#)

A novel gene, NSD1, is fused to NUP98 in the t(5;11)(q35;p15.5) in de novo

childhood acute myeloid leukemia.

Jaju RJ, Fidler C, Haas OA, Strickson AJ, Watkins F, Clark K, Cross NC, Cheng JF, Aplan PD, Kearney L, Boulton J, Wainscoat JS.

Blood 2001; 98(4): 1264-1267

Medline [11493482](#)

Molecular characterization of NSD1, a human homologue of the mouse Nsd1 gene.

Kurotaki N, Harada N, Yoshiura K, Sugano S, Niikawa N, Matsumoto N.

Gene 2001; 279(2): 197-204

Medline [11733144](#)

NUP98 gene fusions in hematologic malignancies.

Lam DH, Aplan PD.

Leukemia 2001; 15(11): 1689-1695.(REVIEW)

Medline [11681408](#)

A cryptic t(5;11)(q35;p15.5) in two AML children with apparently normal karyotypes, identified by a multiplex FISH telomere assay.

Brown M, Jawad M, Eils R, Twigg SFR, Saracoglu K, Sauerbrey A, Thomas AE, Harbott J, Kearney L.

Blood (2002) (in press)

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

[BiblioGene - INIST](#)

Contributor(s)

Written 03-2002 Lyndal Kearney

Citation

This paper should be referenced as such :

Kearney L . NSD1 (Nuclear receptor-binding, su(var), enhancer-of-zeste and trithorax domain-containing protein 1. Atlas Genet Cytogenet Oncol Haematol. March 2002 .

URL : <http://www.infobiogen.fr/services/chromcancer/Genes/NSD1ID356.html>

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