

SEPT5 (septin 5)

Identity

Other names	hCDCRel-1 (human cell division cycle regulation 1) PNUTL1 (peanut (drosophila)- like 1) CDCREL AF22 (ALL1 fused gene from chromosome 22)
Hugo	SEPT5
Location	22q11.2

DNA/RNA

Description	the gene spans 13 kb; 13 exons; one large intron (3.7kb) between exons 2 and 3
Transcription	two major splicings in 5' (exons 1 and 2 versus exon 3; 2032 bp mRNA; coding sequence: 1109 bp; the gene is just 5' of GPIb beta (platelet membrane glycoprotein I beta precursor), and GPIb beta is co-expressed with hCDCRel-1; this is due to a non consensus polyadenylation signal in 3' of hCDCRel-1

Protein

Description	369 amino acids; GTPase activity
Expression	high level expression in brain, heart, platelets; low level expression in some other tissues
Localisation	cytoplasm
Function	see below; may have a role in the synaptic function of neurones in the brain
Homology	belong to the septin family: filament forming proteins implicated in the cytoskeleton organization; nucleotide binding proteins; hCDCRel-1 is closely related to AF17q25/MSF , also found involved in fusion protein with MLL in leukemia

Implicated in

Entity	t(11;22)(q23;q11) acute non lymphocytic leukemia (ANLL) --> MLL - hCDCRel-1
Disease	M4, M2, and M1 ANLL
Hybrid/Mutated Gene	5' MLL - 3' hCDCRel, with fusion of MLL exon 7 to hCDCRel exon 3
Abnormal Protein	NH2 - AT hook and DNA methyltransferase from MLL fused to hCDCREL-1 - COOH

External links

Nomenclature

Hugo	SEPT5
GDB	SEPT5
Entrez_Gene	SEPT5 5413 septin 5

Cards

Atlas	hCDCRel-1ID220
GeneCards	SEPT5
Ensembl	SEPT5
Genatlas	SEPT5
GeneLynx	SEPT5
eGenome	SEPT5
euGene	5413

Genomic and cartography

GoldenPath	SEPT5 - 22q11.2 chr22:18081987-18092297 + 22q11.21 (hg18-Mar_2006)
Ensembl	SEPT5 - 22q11.21 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	SEPT5

Gene and transcription

Genbank	AF061154 [ENTREZ]
Genbank	AK056273 [ENTREZ]
Genbank	BC025261 [ENTREZ]
Genbank	BC030503 [ENTREZ]
Genbank	CR456545 [ENTREZ]
RefSeq	NM_002688 [SRS] NM_002688 [ENTREZ]
AceView	SEPT5 AceView - NCBI
TRASER	SEPT5 Traser - Stanford
Unigene	Hs.517410 [SRS] Hs.517410 [NCBI] HS517410 [spliceNest]

Protein : pattern, domain, 3D structure

SwissProt	Q59GE1 [SRS] Q59GE1 [EXPASY] Q59GE1 [INTERPRO]
Interpro	IPR000038 Cell_Div_GTP_bd [SRS] IPR000038 Cell_Div_GTP_bd [EBI]
CluSTr	Q59GE1
Pfam	PF00735 GTP_CDC [SRS] PF00735 GTP_CDC [Sanger]] pfam00735 [NCBI-CDD]
Blocks	Q59GE1

Protein Interaction databases

DIP	Q59GE1
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IntAct	Q59GE1
	Polymorphism : SNP, mutations, diseases
OMIM	602724 [map]
GENECLINICS	602724
SNP	SEPT5 _[dbSNP-NCBI]
SNP	NM_002688 _[SNP-NCI]
SNP	SEPT5 _[GeneSNPs - Utah] SEPT5 _[HGBASE - SRS] SEPT5 _[SNP - HAPMAP]
	General knowledge
Family Browser	SEPT5 _[UCSC Family Browser]
SOURCE	NM_002688
SMD	Hs.517410
SAGE	Hs.517410
Amigo	nucleotide binding
Amigo	cytokinesis
Amigo	cytokinesis
Amigo	GTPase activity
Amigo	GTPase activity
Amigo	structural molecule activity
Amigo	structural molecule activity
Amigo	protein binding
Amigo	protein binding
Amigo	GTP binding
Amigo	plasma membrane
Amigo	plasma membrane
Amigo	cell cycle
Amigo	synaptic vesicle
Amigo	synaptic vesicle
Amigo	synaptic vesicle targeting
Amigo	synaptic vesicle targeting
Amigo	regulation of exocytosis
Amigo	regulation of exocytosis
BIOCARTA	Role of Parkin in the Ubiquitin-Proteasomal Pathway [Genes]
PubGene	SEPT5
	Other databases
	Probes
Probe	SEPT5 Related clones (RZPD - Berlin)
	PubMed

[PubMed](#) [21 Pubmed reference\(s\) in LocusLink](#)

Bibliography

A human gene similar to *Drosophila melanogaster* peanut maps to the DiGeorge syndrome region of 22q11.

McKie JM, Sutherland HF, Harvey E, Kim UJ, Scambler PJ.

Hum Genet 1997; 101: 6-12.

Medline [98046335](#)

Alternative expression of platelet glycoprotein Ib-beta mRNA from an adjacent 5-prime gene with an imperfect polyadenylation signal sequence.

Zieger B, Hashimoto Y, Ware J.

J Clin Invest 1997; 99: 520-525.

Localization of a novel septin protein, hCDCrel-1, in neurons of human brain.

Caltagarone J, Rhodes J, Honer WG, Bowser R.

Neuroreport 1998; 9: 2907-2912.

Medline [98430944](#)

Structure and expression of the human septin gene HCDCREL-1.

Yagi M, Zieger B, Roth GJ, Ware J.

Gene 1998; 212: 229-236.

Medline [98278839](#)

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Contributor(s)

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URL : <http://AtlasGeneticsOncology.org/Genes/hCDCRel-1ID220.html>

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