

## Gene Section

### Short Communication

## SEPT5 (septin 5)

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### Identity

**Other names:** hCDCRel-1 (human cell division cycle regulation 1); PNUTL1 (peanut (drosophila)- like 1); CDCREL; AF22 (ALL1 fused gene from chromosome 22)

**HGNC (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 Ib 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

***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.

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