

## NUP214 (nucleoporin 214kDa)

### Identity

Other names

**CAN**

**CAIN**

**D9S46E**

**NUP214 (nuclear pore complex protein 214 kDa)**

Hugo

[NUP214](#)

Location

9q34.3

from centromere to telomere: SET, , NUP214 (alias CAN), NOTCH1 (alias [TAN1](#))



[NUP214](#) (9q34.3) - Courtesy Mariano Rocchi, [Resources for Molecular Cytogenetics](#). Laboratories willing to validate the probes are welcome : contact [rocchi@biologia.uniba.it](mailto:rocchi@biologia.uniba.it)

### DNA/RNA

Description 36 exons encompassing about 108 kb of genomic DNA

Transcription 6.6 kb mRNA

### Protein

Description 2090 amino acids; 214 kDa; dimerization domains (2 leucine zippers) and FG repeats; forms homodimers

Expression thymus, bone marrow, spleen, kidney, testis, brain; apparently not in other tissues

Localisation nuclear membrane; cytoplasmic face of nucleopore

Function Nucleoporins are the main components of the [nuclear pore complex](#)(NPC) in eukaryotic cells. The nuclear pore complex is a massive structure that extends across the nuclear envelope, forming a gateway that regulates the flow of macromolecules between the nucleus

and the cytoplasm. NUP214 may serve as a docking site in the receptor mediated import of substrates across the NPC, and plays a role in nuclear protein import, mRNA export, and cell cycle progression; interacts with DDX19, [NUP88](#), and XPO1.

Homology NUP214 is a member of the FG-repeat-containing nucleoporins

### Implicated in

**Entity** [t\(6;9\)\(p23;q34\)](#) --> [DEK](#) - NUP214  
**Disease** M2, M4 [ANLL](#) or [MDS](#)  
**Prognosis** remission difficult to obtain  
**Cytogenetics** this chromosome anomaly may be over looked  
**Hybrid/Mutated Gene** 5' DEK - 3' NUP214; chromosome 6 breakpoint clusters in a single intron  
**Abnormal Protein** head to tail DEK/NUP214 fusion protein (the alternative SET/NUP214 is exceptional); almost the entire DEK protein is fused to the C-terminal two-thirds of the NUP214 protein; nuclear localization

**Entity** [t\(9;9\)\(q34;q34\)/AUL](#) --> [SET](#)-NUP214

**Note** the only SET-NUP214 positive case described so far had a normal karyotype; on the cytogenetic level it is unclear whether the SET-NUP214 fusion is generated by a t(9;9)(q34;q34) or an interstitial deletion at 9q34; the latter is supported by the centromere-telomere orientation of both genes and their local order: centromere ' SET - NUP214' telomere

**Disease** only one case to date; acute undifferentiated leukemia

**Cytogenetics** may be overlooked

**Hybrid/Mutated Gene** 5' SET 3' NUP214

**Entity** amplification --> NUP214-[ABL1](#)

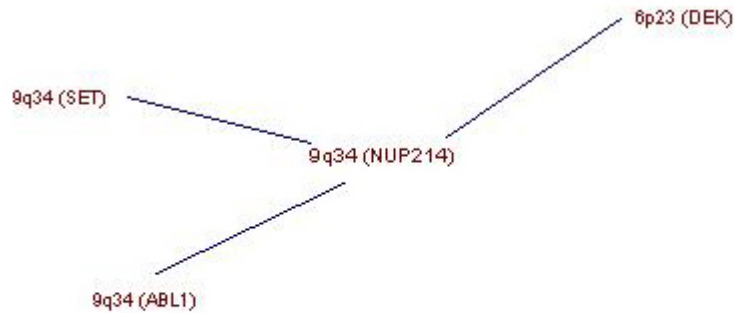
**Disease** 5-6% of childhood and adult [T-ALL](#)

**Prognosis** aggressive course of disease

**Cytogenetics** found in T-ALL with various karyotypes

**Hybrid/Mutated Gene** episomal amplification of the 5' NUP214 3' ABL1 fusion gene

### Breakpoints



NUP214 (CAN) and partners. Editor 08/2005.

## External links

### Nomenclature

<a href="#">Hugo</a>	<a href="#">NUP214</a>
<a href="#">GDB</a>	<a href="#">NUP214</a>
<a href="#">Entrez_Gene</a>	<a href="#">NUP214</a> <a href="#">8021</a> nucleoporin 214kDa

### Cards

<a href="#">Atlas</a>	<a href="#">CAN</a>
<a href="#">GeneCards</a>	<a href="#">NUP214</a>
<a href="#">Ensembl</a>	<a href="#">NUP214</a>
<a href="#">GenAtlas</a>	<a href="#">NUP214</a>
<a href="#">GeneLynx</a>	<a href="#">NUP214</a>
<a href="#">eGenome</a>	<a href="#">NUP214</a>
<a href="#">euGene</a>	<a href="#">8021</a>

### Genomic and cartography

<a href="#">GoldenPath</a>	<a href="#">NUP214</a> - <a href="#">9q34.3</a> <a href="#">chr9:132990802-133098911</a> + <a href="#">9q34.13</a> (hg18-Mar_2006)
<a href="#">Ensembl</a>	<a href="#">NUP214</a> - <a href="#">9q34.13</a> [CytoView]
<a href="#">NCBI</a>	<a href="#">Genes Cyto</a> <a href="#">Gene Seq</a> [Map View - NCBI]
<a href="#">OMIM</a>	<a href="#">Disease map</a> [OMIM]
<a href="#">HomoloGene</a>	<a href="#">NUP214</a>

### Gene and transcription

<a href="#">Genbank</a>	<a href="#">AB159230</a> [ENTREZ]
<a href="#">Genbank</a>	<a href="#">AJ420413</a> [ENTREZ]
<a href="#">Genbank</a>	<a href="#">AL832609</a> [ENTREZ]
<a href="#">Genbank</a>	<a href="#">BC012500</a> [ENTREZ]
<a href="#">Genbank</a>	<a href="#">BC045620</a> [ENTREZ]
<a href="#">RefSeq</a>	<a href="#">NM_005085</a> [SRS] <a href="#">NM_005085</a> [ENTREZ]

[AceView](#) [NUP214](#) AceView - NCBI  
[TRASER](#) [NUP214](#) Traser - Stanford  
[Unigene](#) [Hs.461860](#) [SRS] [Hs.461860](#) [NCBI] [HS461860](#) [spliceNest]

### Protein : pattern, domain, 3D structure

[SwissProt](#) [P35658](#) [SRS] [P35658](#) [EXPASY] [P35658](#) [INTERPRO]  
[Interpro](#) [IPR001680 WD40](#) [SRS] [IPR001680 WD40](#) [EBI]  
[Interpro](#) [IPR011046 WD40 like](#) [SRS] [IPR011046 WD40 like](#) [EBI]  
[CluSTr](#) [P35658](#)  
[Smart](#) [SM00320 WD40](#) [EMBL]  
[Blocks](#) [P35658](#)  
[HPRD](#) [P35658](#)

### Protein Interaction databases

[DIP](#) [P35658](#)  
[IntAct](#) [P35658](#)

### Polymorphism : SNP, mutations, diseases

[OMIM](#) [114350;601626](#) [map]  
[GENECLINICS](#) [114350;601626](#)  
[SNP](#) [NUP214](#) [dbSNP-NCBI]  
[SNP](#) [NM\\_005085](#) [SNP-NCI]  
[SNP](#) [NUP214](#) [GeneSNPs - Utah] [NUP214](#) [HGBASE - SRS]  
[HAPMAP](#) [NUP214](#) [HAPMAP]

### General knowledge

[Family Browser](#) [NUP214](#) [UCSC Family Browser]  
[SOURCE](#) [NM\\_005085](#)  
[SMD](#) [Hs.461860](#)  
[SAGE](#) [Hs.461860](#)  
[Amigo](#) [transporter activity](#)  
[Amigo](#) [protein binding](#)  
[Amigo](#) [molecular function unknown](#)  
[Amigo](#) [nucleus](#)  
[Amigo](#) [nuclear pore](#)  
[Amigo](#) [protein export from nucleus](#)  
[Amigo](#) [transport](#)  
[Amigo](#) [porin activity](#)  
[Amigo](#) [integral to membrane](#)  
[Amigo](#) [outer membrane](#)  
[PubGene](#) [NUP214](#)

## Other databases

### Probes

[Probe](#) [Cancer Cytogenetics \(Bari\)](#)

[Probe](#) [NUP214 Related clones \(RZPD - Berlin\)](#)

### PubMed

[PubMed](#) [33 Pubmed reference\(s\) in LocusLink](#)

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Medline [1630450](#)

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Medline [92195315](#)

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Medline [95273090](#)

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Medline [8896451](#)

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Medline [10546895](#)

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Boer J, Bonten-Surtel J, Grosveld G.

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Stergianou K, Fox C, Russell NH.

Leukemia. 2005; 19: 1680-1681.

Medline [16015385](#)

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

[BiblioGene - INIST](#)

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URL : <http://AtlasGeneticsOncology.org/Genes/CAN.html>

**Strehl S** . NUP214 (nucleoporin 214kDa). Atlas Genet Cytogenet Oncol Haematol. September 2005 .

URL : <http://AtlasGeneticsOncology.org/Genes/CAN.html>

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