Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Gene Section
Mini Review

NUP214 (nucleoporin 214kDa)

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Identity

Hugo: NUP214
Other names: CAN; CAIN; D9S46E; NUP214 (nuclear pore complex protein 214 kDa)
Location: 9q34.3
Local order: from centromere to telomere: SET, ABL1, NUP214 (alias CAN), NOTCH1 (alias TAN1).

NUP214 (9q34.3) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

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../Deep/NuclearPoreComplID20048.html 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

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.

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

Amplification ⟶ NUP214- ABL1

Disease

5-6% of childhood and adult T-ALL.

Prognosis

Aggressive course of disease.

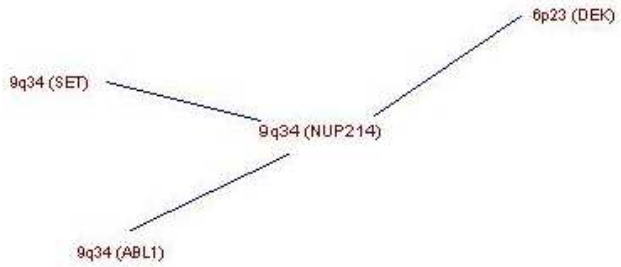
Cytogenetics

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.

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