

Leukaemia Section

Mini Review

t(9;9)(q34;q34)

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Clinics and pathology

Disease

Acute undifferentiated leukemia (AUL).

Epidemiology

Only one case described so far.

Cytogenetics

Note: the only SET-NUP214 positive case described so far had a normal karyotype; on the cytogenetic level it is unclear if 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.

Cytogenetics morphological

The single case described so far had a normal karyotype; may be overlooked.

Genes involved and Proteins

SET

Location: 9q34

DNA / RNA

8 exons spanning 6,81 kb of genomic DNA; 2559 bp mRNA.

Protein

Two isoforms; isoform 1 (TAF1 alpha) 290 amino acids, 33.5 kDa; isoform 2 (TAF1 beta) - 277 amino acids, 23.1 kDa; DNA associated, transcription factor; multitasking protein, involved in apoptosis, transcription, nucleosome assembly and histone binding; cytoplasmic and nuclear; in the cytoplasm,

found both in the cytosol and associated with the endoplasmic reticulum.

NUP214

Location: 9q34

DNA / RNA

36 exons encompassing about 108 kb of genomic DNA; 6.6 kb mRNA.

Protein

2090 amino acids, 213.8 kDa; forms homodimers, contains two leucine zipper dimerization domains, and FG repeats at the C-terminus, which are homologous to those observed in other members of the nucleoporin family; expressed in thymus, spleen, bone marrow, kidney, brain and testis, but hardly in all other tissues or in whole embryos during development; may serve as a docking site in the receptor-mediated import of substrates across the nuclear pore complex (NPC); intracellular localization, cytoplasmic face of the NPC.

Results of the chromosomal anomaly

Hybrid gene

Transcript

5' SET - 3' NUP214

Fusion protein

Description

The SET-NUP214 fusion protein consists of almost the whole SET protein fused to the C-terminus of NUP214; 155 kDa.

Oncogenesis

SET-NUP214 leads to disorganization of nuclear export.

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