t(4;11)(q21;p15)
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Identity
Note
Not to be confused with a variant of the classical t(4;11)(q21;q23) translocation.

Clinics
No notable particular aspect.

Prognosis
Probably unfavorable, median survival below 18 months; improved by allogeneic bone marrow transplantation.

Cytogenetics
Cytogenetics morphological
In approximately 2/3 of cases; 2 cases with 12p-.

Cytogenetics molecular
Two BAC clones 290A12 and 118H17 (California Institute of Technology BAC library) encompasses all NUP98 gene and are split by translocation.

Variants
Not described.

Genes involved and proteins
RAP1GDS1
Location
4q22.3
Protein
SmgGDS, 558 amino acids; stimulates GDP --> GTP transition in a series of small GTP-binding proteins (g proteins) including rap1a, rap1b, K-ras, rac1, rac2, rhoA and ralB.

Somatic mutations
Not involved in other known clonal rearrangement associated with tumoral proliferation.

NUP98
Location
11p15.4
Protein
Nucleoporin 98, a 98 kDa component of the nuclear pore complex implicated in nucleo-cytoplasmic transport.

Somatic mutations
involved in different types of acute myeloid leukemia, as fusion gene with HOX A9, DDX10, HOX D13, TOP1, PMX1 and LEDGF, resulting respectively from t(7;11)(p15;p15), inv(11)(p15q22), t(2;11)(q31;p15), t(11;20)(p15;q11), t(1;11)(q23;p15) and t(9;11)(p22;p15).

Result of the chromosomal anomaly

Hybrid gene
Description
NUP98 breakpoint in the intron between exons B and C; 5'-part of NUP98 is fused in frame with the whole coding sequence of RAP1GDS1; fusion gene called NRG: 5'-NUP-RAP1GDS1-3'. Variant described with breakpoint in NUP98 before exon A.

Fusion protein
Description
t(4;11) generates only one chimeric protein 5' - NUP98 - RAP1GDS1 - 3' which contains a variable part of NUP98 and the totality of smgGDS except for the initial methionine.

References
Hussey DJ, Nicola M, Moore S, Peters GB, Dobrovic A. The (4;11)(q21;p15) translocation fuses the NUP98 and RAP1GDS1 genes and is recurrent in T-cell acute lymphocytic leukemia. Blood. 1999 Sep 15;94(6):2072-9

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