

Leukaemia Section

Mini Review

t(2;11)(q11;q23)

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Published in Atlas Database: August 2005

Online updated version: <http://AtlasGeneticsOncology.org/Anomalies/t0211q11q23ID1196.html>

DOI: 10.4267/2042/38266

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Clinics and pathology

Disease

Acute lymphocytic leukemia (ALL) with proB phenotype.

Epidemiology

Three cases reported to date, two infants and one two-year-old child.

Prognosis

Both cases of infant ALL showed a poor survival, 2 and 9 months respectively. The third case, a two-year-old child, achieved complete remission.

Cytogenetics

Cytogenetics molecular

The three reported cases carried different rearrangements involving chromosomes 2 and 11: t(2;11)(p15;p14), t(2;11)(q11;q23) and ins(11;2)(q23;q11.2q11.2).

Probes

LAF4 specific PACs: RP6-44B23 and RP6-226I23.

Genes involved and Proteins

MLL

Location: 11q23

DNA / RNA

13-15 kb mRNA.

Protein

431 kDa; contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyl transferase motif, a bromodomain; transcriptional regulatory factor; nuclear localisation.

LAF4

Location: 2q11-q12

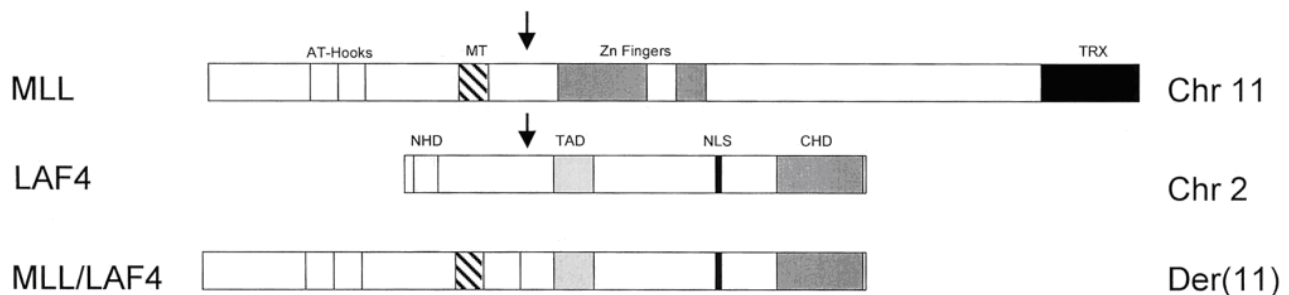
Note: AF4 and AF5q31, also known as MLL fusion partners, belong to the same gene family.

DNA / RNA

22 exons, transcript length: 3855 bp.

Protein

LAF4 protein (Lymphoid nuclear protein related to AF4) 1226 amino acids; 133734 Da.



Schematic representation of MLL, LAF4, and the putative MLL-LAF4 fusion protein. MT, methyltransferase domain; TRX, Drosophila trithorax homology; NHD, N-terminal homology domain; TAD, transactivation domain; NLS, nuclear localization sequence; CHD, C-terminal homology domain.

Results of the chromosomal anomaly

Hybrid gene

Transcript

5' MLL - 3' LAF4

Fusion protein

Description

The MLL-LAF4 fusion protein includes the transactivation domain of LAF4 that is part of the AF4/LAF4/FMR2 homology domain.

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This article should be referenced as such:

von Bergh ARM. t(2;11)(q11;q23). *Atlas Genet Cytogenet Oncol Haematol.*2006; 10(1):12-13.
