t(11;17)(q23;q12-21) MLL/LASP1

Clinics and Pathology

Disease	infant acute myeloid leukemia AML-M4
Epidemiology	only one case described so far
Prognosis	insufficient data; of note: the only patient described, remains in complete remission >8 years

Cytogenetics

Note	so far three MLL fusion partners, namely LASP1 (in the t(11;17) herein described), MLLT6 (alias AF17) (in another t(11;17)(q23;q12-21), and ACACA (also in another t(11;17)(q23;q12-21) have been identified in 17q12-21; these translocations cannot be distinguished cytogenetically and the accurate detection of the specific fusion gene requires RT-PCR or refined FISH analysis
Cytogenetics Morphological	sole abnormality

Genes involved and Proteins

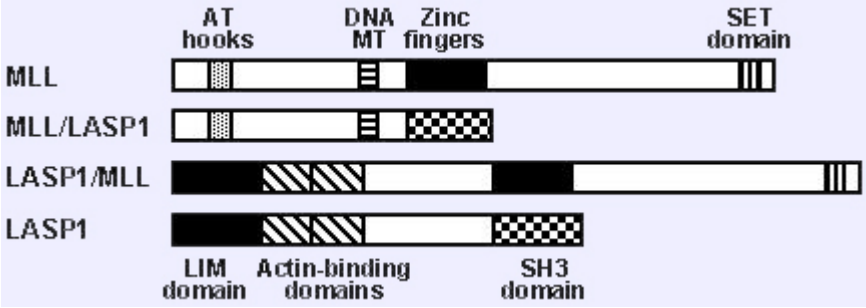
Gene Name	MLL
Location	11q23
Dna / Rna	37 exons, spanning over 100 kb; transcription in a centromeric to telomeric direction; 13 and 15 kb mRNA; coding sequence: 11.9 kb
Protein	431 kDa; contains two DNA binding motifs (an AT hook, and Zinc fingers), a DNA methyl transferase motif, and a bromodomain; transcriptional regulatory factor; nuclear localization
Gene Name	LASP1
Location	17q12
Note	previously LASP1 and MLLT6 (alis AF17) were mapped to 17q21, but according to the most recent genome assembly built and recent FISH data both genes are localized in 17q12 and proximal to RARA
Dna / Rna	7 exons spanning about 50 kb of genomic DNA; 3845 bp mRNA, 783 bp coding sequence; ubiquitous expression
Protein	LASP1 encodes a member of a LIM protein subfamily; contains a LIM motif, two actin-binding domains, and an SH3 domain; cytoplasmic localization

Result of the chromosomal anomaly

Hybrid gene
Transcript

5'MLL - 3'LASP1; also the reciprocal 5'LASP1 - 3'MLL is present

Fusion Protein



Schematic representation of MLL, LASP1, and the putative MLL-LASP1 and LASP1-MLL fusion proteins.

Description the C-terminal SH3 domain of LASP1 is fused to the N-terminal portion of MLL retaining the AT-hook DNA-binding domain and the DNA methyltransferase motif (MT)

External links

Other database t(11;17)(q23;q12-21) MLL/LASP1 — CancerChromosomes (NCBI)

Other database Genome Bioinformatics Group of the University of California Santa Cruz

To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:
you are welcome to submit a paper to our new Case Report section.

Bibliography

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Contributor(s)

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URL : http://AtlasGeneticsOncology.org/Anomalies/t1117q23q21LASP1ID1367.html