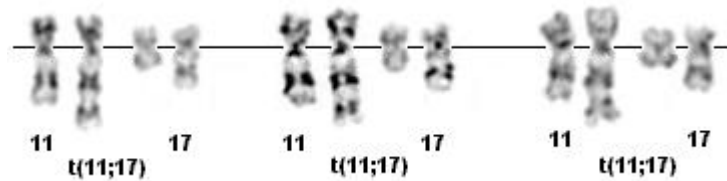


t(11;17)(q23;q12-21) MLL/AF17

Identity

Note not to be confused with the [t\(11;17\)\(q23;q12-21\)](#) involving [MLL](#) and [LASP1](#) or the t(11;17)(q23;q12-21) involving MLL and ACACA



t(11;17)(q23;q12) G-banding - Courtesy Cytogenetics Laboratory of the CCRI, Children's Cancer Research Institute, Vienna.

Clinics and Pathology

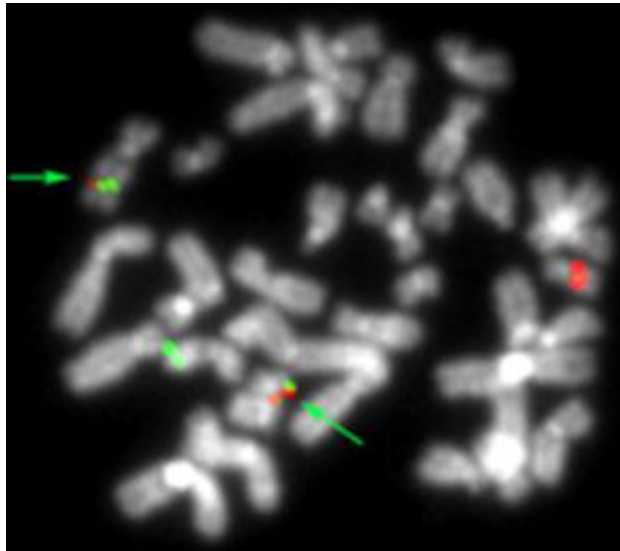
Disease [Acute myeloid leukemia](#) (AML)

Epidemiology not sufficient cases to date

Cytogenetics

Note so far three MLL fusion partners, namely LASP1, AF17 (alias MLLT6), and ACACA 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 chromosomes (arrows).
Molecular



FISH using a combination of the MLL-specific PACs 217a21 and 167k13 (green signals) and the AF17-specific BAC RP11-25H10 (red signals) results in two fusion signals on the der(11) and the der(17)

Probes AF17-specific probes: BACs RP11-25H10 and RP11-58E17

Additional anomalies +8

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 -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 [MLLT6 \(alias AF17\)](#)

Location 17q12

Note previously LASP1 and AF17 (alias MLLT6) were mapped to 17q21, but according to the most recent genome assembly built by the Genome Bioinformatics Group of the University of California Santa Cruz and recent FISH data both genes are localized in 17q12 and proximal to [RARA](#)

Dna / Rna encompasses 19.97 kb of genomic DNA; 4914 bp mRNA; 20 exons, 3282 bp coding sequence

Protein 1023 amino acids, 112 kDa; MLLT6 (alias AF17), [MLLT10](#) (alias AF10), and BRPF1 (alias BR140) belong to a small evolutionary highly

conserved family of putative nuclear transcription factors, which contain amino-terminal PHD fingers, a highly conserved octapeptide, and a classical leucine zipper dimerization motif; nuclear localization

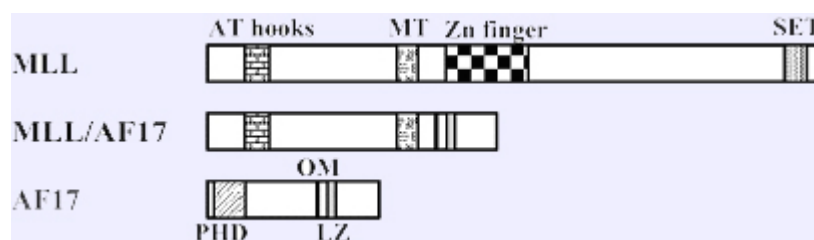
Result of the chromosomal anomaly

Hybrid gene

5' MLL - 3' AF17

Transcript

Fusion Protein



Schematic representation of MLL, AF17 (alias MLLT6), and the putative MLL-AF17 fusion protein. MT, methyltransferase domain; Zn finger, Zinc finger domain; SET-domain; PHD, Zinc finger PHD-type; OM, octapeptide motif; LZ, leucine-zipper dimerization motif.

Description the AT-hook DNA-binding domain and the methyltransferase motif including the CXXC zinc-finger (Zn) domain of MLL are fused to the highly conserved octapeptide (OM) and the leucine-zipper (LZ) dimerization motif of AF17 (alias MLLT6).

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](#).

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Contributor(s)

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