Leukaemia Section
Mini Review

\textbf{t(11;17)(q23;q12-21) MLL/AF17}

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**Identity**

\textbf{Note:} Not to be confused with the \textbf{t(11;17)(q23;q12-21)} involving MLL and LASP1 or the \textbf{t(11;17)(q23;q12-21)} involving MLL and ACACA.

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{t1117q23q12.png}
\caption{t(11;17)(q23;q12) G-banding - Courtesy Cytogenetics Laboratory of the CCRI, Children's Cancer Research Institute, Vienna.}
\end{figure}

**Clinics and pathology**

\textbf{Disease}

Acute myeloid leukemia (AML).

\textbf{Epidemiology}

Not sufficient cases to date.

**Cytogenetics**

\textbf{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.

\textbf{Cytogenetics molecular}

Chromosomes (arrows on the figure below).

\textbf{Probes}


\textbf{Additional anomalies}

+8.

**Genes involved and Proteins**

\textbf{MLL}

\textbf{Location:} 11q23

\textbf{DNA / RNA}

37 exons, spanning over 100 kb; transcription in a centromeric to telomeric direction; 13 -15 kb mRNA; coding sequence 11.9 kb.

\textbf{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.

\textbf{MLLT6 (alias AF17)}

\textbf{Location:} 17q12

\textbf{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.
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).

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.

**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.

**Fusion protein**
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).

**Results of the chromosomal anomaly**

**Hybrid gene**

**Transcript**
3′ MLL - 3′ AF17

**References**


This article should be referenced as such: