Leukaemia Section
Mini Review

\textbf{t(1;21)(p36;q22)}

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\textbf{Identity}

Note: Subtle cytogenetic abnormality, easy to confuse with a del(21)(q22), may be missed in poor quality metaphases.

\begin{figure}
\centering
\includegraphics[width=\textwidth]{t121p36q22.png}
\end{figure}

Partial GTG-banded karyotype of t(1;21)(p36;q22).
Disease

Acute non lymphocytic leukemia (ANLL or AML: acute myeloid leukemia) and myelodysplastic syndromes (MDS); 2 of 5 cases at least are secondary to toxic exposure.

Note: Only 5 cases described so far one with features identical to a case of TXT t(18;21)(q21;q22), and a case of t(19;21)(q13.4;q22).

Etiology

Two of the reported cases are therapy-related, in another case, ANLL occurred about 50 years after radiation exposure from nuclear explosions.

Prognosis

Poor; median survival 6 months.

Cytogenetics

Probes

AML1/ETO dual-color, dual-fusion probe.

Additional anomalies

-7, del(7q).
Schematic representation of RUNX1 and PRDM16 (fusion) genes.
Upper panel: normal genomic structures of PRDM16 and RUNX1 (non-coding parts in blue). A cryptic exon, residing within intron 1 of PRDM16, is indicated in green (speckled).
Lower panel: structure of RUNX1-PRDM16 fusion transcripts. Exons are numbered on the basis of consensus gene sequences. Exon sizes are not to scale.

Genes involved and Proteins

**PRDM16**
Location: 1p36
Note: This gene is also involved in the t(1;3)(p36;q21) (AML/MDS).
Protein
Zinc-finger protein, containing two DNA binding domains and a PRDI-BF1 (positive regulatory domain I binding factor 1/ RIZ (retinoblastoma-interacting zinc finger protein) homologous (PR) domain at the N-terminus.

**RUNX1/AML1**
Location: 21q22
DNA / RNA
Transcription is from telomere to centromere.
Protein
Contains a Runt domain and, in the C-term, a transactivation domain; forms heterodimers; widely expressed; nuclear localisation; transcription factor (activator) for various hematopoietic-specific genes.

Results of the chromosomal anomaly

**Hybrid gene**
Note: Two different chimeric transcripts have been identified. One contains the exon 1 to 6 of RUNX1 including the runt domain, fused to sequences derived from intron 1 of PRDM16. The other fusion transcript contains exons 1 to 6 of RUNX1 and almost the entire PRDM16 coding region (see figure above).
Description
5' RUNX1 - 3' PRDM16.

References

This article should be referenced as such: