Leukaemia Section
Short Communication

\[ t(6;11)(q13;q23) \]

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Clinics and pathology

\textbf{Disease}

Acute leukemia
\textbf{Note:} Only 2 cases to date; one of which was not further described.

\textbf{Phenotype / cell stem origin}

One case was a M4 acute myeloid leukaemia.

\textbf{Epidemiology}

The patient was a 14-year-old girl.

\textbf{Prognosis}

Survival was 18 months in the only documented case.

Cytogenetics

\textbf{Additional anomalies}

Sole anomaly.

\textbf{Genes involved and Proteins}

\textbf{SMAP1}

\textbf{Location:} 6q13

\textbf{Protein}

SMAP1 is a GTPase-activating protein (GAP) for Arf6. Vesicle formation requires clathrin, its adaptors, and an enzymatic activity. This is given by small GTPase ADP-ribosylation factors (Arf; there is six Arfs, Arf1, Arf3, Arf2/Arf4, Arf5 and Arf6). Arf switches between a GTP-bound active state (regulated by guanine-nucleotide exchange factor (GEF)) and a GDP-bound inactive state (regulated by GTPase-activating protein (GAP)). Arf6/SMAP1 regulates the clathrin dependent endocytosis of vesicles from the plasma membrane and the recycling of endosome to the plasma membrane.

\textbf{MLL}

\textbf{Location:} 11q23

\textbf{Protein}

Transcriptional regulatory factor; MLL may have yin-yang functions though actions of MLL-N and MLL-C (e.g. desacetylation/acyetlation); MLL-N acts as a transcriptional repressor; MLL can be associated with more than 30 proteins, including the core components of the SWI/SNF chromatin remodeling complex and the transcription complex TFIID. MLL binds promoters of Hox genes through acetylation and methylation of histones. MLL is a major regulator of hematopoiesis and embryonic development, through regulation of Hox genes expression regulation (HoxA9 in particular).

Results of the chromosomal anomaly

\textbf{Hybrid gene}

\textbf{Description}

5' MLL-3' SMAP1

References


This article should be referenced as such: