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Leukaemia Section
Short Communication

t(6;11)(q13;q23)

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Clinics and pathology

Disease

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

Phenotype / cell stem origin

One case was a M4 acute myeloid leukaemia.

Epidemiology

The patient was a 14-year-old girl.

Prognosis

Survival was 18 months in the only documented case.

Cytogenetics

Additional anomalies

Sole anomaly.

Genes involved and Proteins

SMAP1

Location: 6q13

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.

MLL

Location: 11q23

Protein

Transcriptional regulatory factor; MLL may have yin-yang functions though actions of MLL-N and MLL-C (e.g. desacetylation/acetylation); 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 promotors of HOX genes through acetylation and methylation of histones. MLL is a major regulator of hematopoesis and embryonic development, through regulation of HOX genes expression regulation (HOXA9 in particular).

Results of the chromosomal anomaly

Hybrid gene

Description

5' MLL-3' SMAP1

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