Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Short Communication

t(11;19)(q23;p13.3) MLL/ACER1

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

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Clinics and pathology

Disease

Acute lymphocytic leukemia (ALL)

Epidemiology

Only one case to date, a case of congenital leukemia (Lo Nigro et al., 2002).

Genes involved and proteins

MLL

Location
11q23

DNA/RNA
36 exons, multiple transcripts 13-15 kb.

Protein
3969 amino acids; 431 kDa; contains two DNA binding motifs (a AT hook and a CXXC domain), a DNA methyl transferase motif, a bromodomain. MLL is cleaved by taspase 1 into 2 proteins before entering the nucleus, called MLL-N and MLL-C. The FYRN and FRYC domains of native MLL associate MLL-N and MLL-C in a stable complex; they form a multiprotein complex with transcription factor TFIID. MLL is a transcriptional regulatory factor. 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 pro-motors of HOX genes through acetylation and methylation of histones. MLL is a major regulator of hematopoesis and embryonic development.

ACER1

Location
19p13.3

Protein
ACER1 is the alkaline ceramidase 1. Ceramidases catalyze hydrolysis of ceramide to generate sphingosine (SPH), which is phosphorylated to form sphingosine-1-phosphate (S1P). Ceramide, SPH, and S1P are bioactive lipids that mediate cell proliferation, differentiation, apoptosis, adhesion and migration (Mao and Obeid, 2008).

Result of the chromosomal anomaly

Hybrid gene

Description
5' MLL - 3' ACER1; fusion of MLL intron 8 to ACER1.

References

Lo Nigro L, Slater DJ, Rappaport EF, Biondi A, Maude S, Megnigal MD, Bungaro S, Schiliro G, Felix CA.. Two partner genes of MLL and additional heterogeneity in t(11;19)(q23;p13) translocations. Blood 2002; 2080 p531a.

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