

Leukaemia Section

Mini Review

t(11;20)(q23;q11)

Jen-Fen Fu, Lee-Yung Shih

Division of Hematology-Oncology, Department of Internal Medicine, Chang Gung Memorial Hospital, 199 Tung-Hwa North Road, Taipei 105, Taiwan

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Clinics and pathology

Disease

pro-B acute lymphoblastic leukemia (L2).

Epidemiology

One case described (42 years); male.

Clinics

The patient presented with fever and chills for 10 days. He had multiple 0.5 to 1.0 cm lymph nodes on bilateral necks, axillary and inguinal areas. He had no hepatosplenomegaly. His initial blood counts were HB 10.3 gm/dL, WBC 579.0 x 10⁹/L with 97% blasts, and platelet 37.0 x 10⁹/L.

Cytology

Bone marrow smear showed 99% blasts (FAB L2), which were negative for myeloperoxidase and exhibited diffuse fine granular patterns for PAS and acid phosphatase stains. Immunophenotypic analysis showed that the blasts were positive for CD19, CD33, CD34, HLA-DR and cyCD79; and negative for CD10, CD7, CD2, CD13, and cyMPO.

Treatment

The patient refused chemotherapy.

Cytogenetics

Cytogenetics morphological

Cytogenetic analysis was not performed.

Genes involved and Proteins

MLL

Location: 11q23

Protein

431 kDa; contains two DNA binding motifs (a AT hook, and Zinc fingers), a DNA methyltransferase motif, and a SET [Su(var)3-9, enhancer of Zeste, and trithorax] domain.

MAPRE1

Location: 20q11.2

Protein

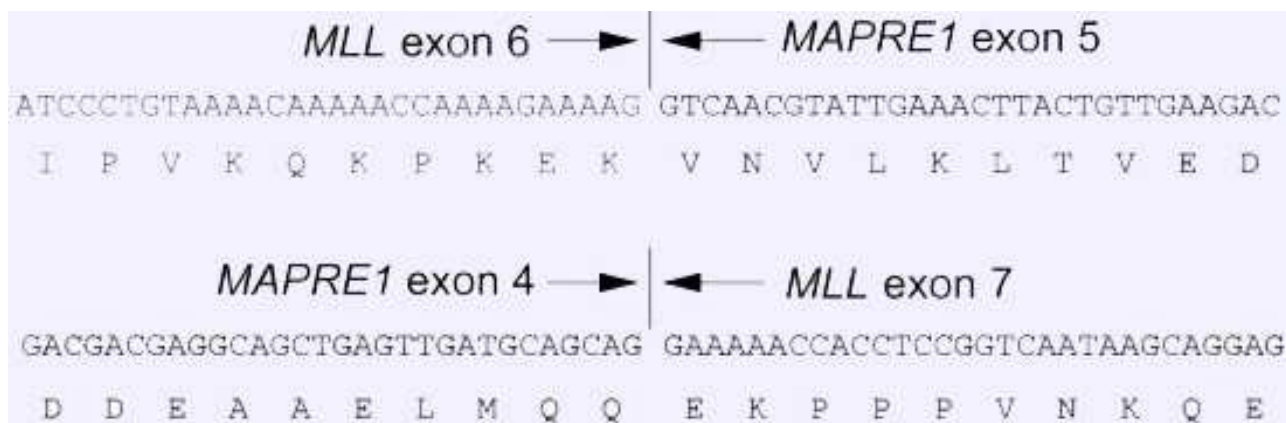
MAPRE1 encoding EB1 which contains a microtubule-binding domain, a dynactin-binding domain (DBD), and an APC-binding domain that is overlapped to DBD; localized at cytoplasmic microtubule tips, centrosomes, and spindle microtubules, and interacts with APC or dynein/dynactin complex to regulate microtubule dynamics, cell polarity, and chromosomal stability.

Results of the chromosomal anomaly

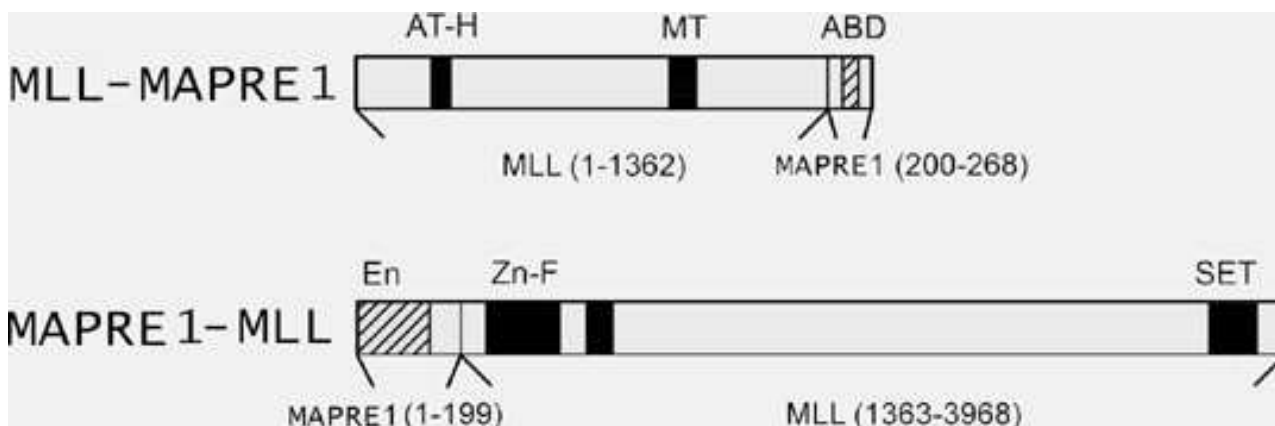
Hybrid gene

Description

5' MLL - 3' MAPRE1, with fusion of MLL exon 6 to MAPRE1 exon 5; the reciprocal in-frame MAPRE1-MLL is also transcribed.



Partial nucleotide and deduced amino acid sequence of the break junctions of MLL-MAPRE1 and MAPRE1-MLL fusion transcripts. Vertical lines indicate break junctions.



Schematic representation of MLL-EB1 and EB1-MLL fusion proteins. AT-H, AT hooks; MT, DNA methyltransferase motif; Zn-F, zinc finger domain; SET, [Su(var)3-9, enhancer of zeste, and trithorax] domain; En, microtubule-binding domain; ABD, APC-binding domain; numbers indicate amino acids of each protein.

Detection protocole

cDNA panhandle PCR.

Fusion protein

Description

NH2-AT hook and DNA methyltransferase motif from MLL fused to APC-binding domain of EB1.

References

Fu JF, Hsu HC, Shih LY. MLL is fused to EB1 (MAPRE1), which encodes a microtubule-associated protein, in a patient with acute lymphoblastic leukemia. *Genes Chromosomes Cancer* 2005;43:206-210.

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