

Leukaemia Section

Short Communication

t(11;21)(p14;q22) RUNX1/KIAA1549L

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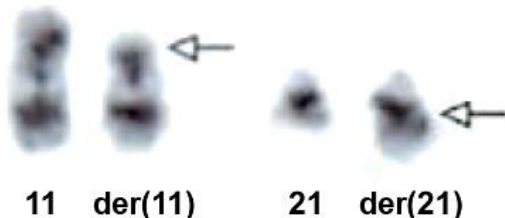
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Abstract

Review on t(11;21)(p14;q22) RUNX1/KIAA1549L BL, with data on clinics, and the genes implicated.

Identity



G-banded partial karyotype of a patient with a t(11;21)(p14;q22). Arrows indicate derivative chromosomes. Clinics and pathology

Disease

Acute myeloid leukemia (AML), AML-M1 by FAB subtype

Phenotype/cell stem origin

CD13, CD19, CD33, CD34, and HLA-DR were positive.

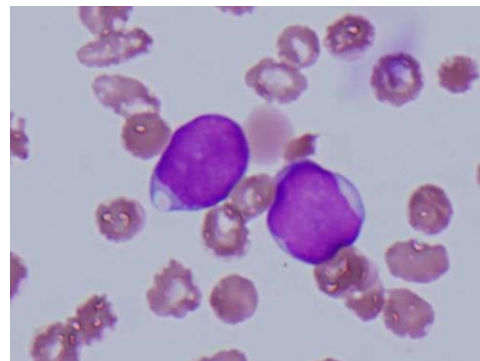
Epidemiology

This is a rare chromosomal rearrangement. A case of MDS with t(11;21)(p14;q22) involving the RUNX1 locus with RUNX1 gene amplification (Moosavi et al., 2009) and a case of AML-M4 with t(11;21)(p13;q22) (Arber et al., 2002) were previously reported. This case is only one AML

patient characterized at molecular level to date (Abe et al., 2012).

Clinics

A 78-year-old man suffering from bleeding tendency and fatigue with dyspnea for one month was diagnosed as AML.



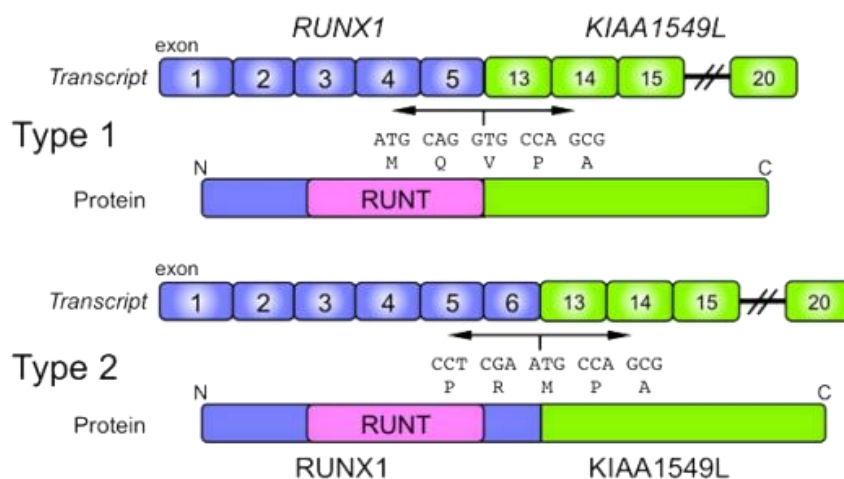
Leukemic cells had large nuclei and little cytoplasm without azure granules.

Cytology

Blast morphology showed minimal differentiation implicating AML M1. Leukemia cells were weakly positive for myeloperoxidase and negative for esterase.

Pathology

Bone marrow examination at diagnosis showed hypocellular marrow with 57% leukemic blasts.



Treatment

The patient received two courses of remission induction chemotherapy with daunorubicin and cytarabine, however, a complete remission was not achieved. The leukemia cells were slow-growing in the early period after diagnosis, so that he received 11 cycles of low dose cytarabine after induction failure and lived for 2 years. He was died from progression of leukemia possible with intracranial hemorrhage.

Genes involved and proteins

KIAA1549L

Location

11p13-14

Note

KIAA1549L is also known as C11ORF41 or C11ORF69. The function of KIAA1549L is not known.

Northern blot analysis of several human tissues detected two transcripts of 11 and 7.9 kb in brain (Gawin et al., 1999). KIAA1549L indicates a KIAA1549-like ortholog. KIAA1549 is known as a fusion partner of BRAF in pilocytic astrocytomas (Jones et al., 2008).

DNA/RNA

The KIAA1549L gene contains 20 exons spanning 132 kb of genomic DNA. Four transcripts are known. Transcription orientation: telomere to centromere.

Protein

The predicted KIAA1549L proteins contain 1849 amino acids, 199 kDa.

RUNX1

Location

21q22

DNA/RNA

Transcription orientation: telomere to centromere.

Protein

The predicted RUNX1 proteins contain 250, 453 and 480 amino acids designated as RUNX1a, RUNX1b and RUNX1c, respectively.

All 3 proteins contain the 128-amino acid Runt domain, but RUNX1a does not contain a transcriptional activation domain of C-terminal region.

Result of the chromosomal anomaly

Hybrid gene

Description

5' RUNX1-KIAA1549L 3'.

Transcript

Two types of in-frame RUNX1-KIAA1549L fusion transcripts were detected. One was a fusion between exon 5 of RUNX1 and exon 13 of KIAA1549L (Type 1) and the other was between exon 6 of RUNX1 and exon 13 of KIAA1549L (Type 2). A reciprocal KIAA1549L-RUNX1 fusion was not detected. Both fusion transcripts include the region encoding Runt homology domain of RUNX1.

References

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