

# Leukaemia Section

## Short Communication

### t(11;21)(q13;q22)

Jean-Loup Huret, Yongquan Xue

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH); Jiangsu Institute of Hematology, the First Affiliated Hospital of Soochow University, Suzhou, PR China (YX)

Published in Atlas Database: December 2009

Online updated version : <http://AtlasGeneticsOncology.org/Anomalies/t1121q13q22ID1505.html>  
DOI: 10.4267/2042/44876

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.  
© 2010 Atlas of Genetics and Cytogenetics in Oncology and Haematology

## Clinics and pathology

### Disease

Myeloid malignancies

### Epidemiology

Three cases of t(11;21)(q13;q22) in myeloid malignancies are available to date: a 63-year-old female patient with AML evolving from myelodysplastic syndrome (MDS) (Imagama et al., 2007), a 65-year-old male patient with a M2 acute myeloid leukemia (M2-AML) (Dai et al., 2007), and a 70-year-old male patient with a chronic myelogenous leukemia (CML). The t(11;21) was found during transformation into blastic crisis (BC-CML) and in the subsequent reversion to the chronic phase (Wang et al., 1988).

### Prognosis

The MDS-to-AML case relapsed two years after remission. The M2-AML case died 10 months after diagnosis. The CML case was remaining in remission of blast crisis for 4 months at the time of the report.

## Cytogenetics

### Cytogenetics morphological

The t(11;21) was part of a complex karyotype in the MDS-to-AML case, found with an additional anomaly in the M2-AML case, and accompanying the classical t(9;22)(q34;q11) in the CML case.

## Genes involved and proteins

### Note

The involvement of RUNX1 was proved in the two AML cases; the involvement of MACROD1/LRP16 as the partner of RUNX1 was established in the case described by Imagama et al.

### MACROD1

#### Location

11q13.1

#### Note

MACROD1 is also known as LRP16.

#### Protein

MACROD1/LRP16 gene has been characterized as an estrogen-responsive gene. LRP16 is required for ERalpha signaling transduction by functioning as an ERalpha coactivator (Han et al., 2007; Han et al., 2008). MACROD1/LRP16-overexpression promotes the cell cycle, and cell proliferation (Yang et al., 2009).

### RUNX1

#### Location

21q22

#### DNA/RNA

Transcription from telomere to centromere.

#### Protein

Contains the RUNT binding domain at 5' portion and the transactivation domain at 3' portion.  
Forms heterodimers; widely expressed; nuclear localization; a transcription factor and critical regulator of hematopoietic-cell development.

## Result of the chromosomal anomaly

### Hybrid gene

#### Description

In the case described by Imagama et al. 2007, the translocation fuses RUNX1 exon 5 or exon 6 to MACROD1 exon 2, suggesting that the RUNX1 breakpoint lies in intron 6 and that alternative fusion splice variants are generated. The reciprocal MACROD1-RUNX1 fusion was also detected.

## References

Wang TY, Raza A, Sait SN, Stein A, Schumer J, Sandberg AA. A t(11;21)(13;q22) in Ph-positive chronic myelogenous leukemia. *Cancer Genet Cytogenet.* 1988 Apr;31(2):187-91

Dai H, Xue Y, Pan J, Wu Y, Wang Y, Shen J, Zhang J. Two novel translocations disrupt the RUNX1 gene in acute myeloid leukemia. *Cancer Genet Cytogenet.* 2007 Sep;177(2):120-4

Han WD, Zhao YL, Meng YG, Zang L, Wu ZQ, Li Q, Si YL, Huang K, Ba JM, Morinaga H, Nomura M, Mu YM. Estrogenically regulated LRP16 interacts with estrogen receptor alpha and enhances the receptor's transcriptional activity. *Endocr Relat Cancer.* 2007 Sep;14(3):741-53

Imagama S, Abe A, Suzuki M, Hayakawa F, Katsumi A, Emi N, Kiyoi H, Naoe T. LRP16 is fused to RUNX1 in monocytic leukemia cell line with t(11;21)(q13;q22). *Eur J Haematol.* 2007 Jul;79(1):25-31

Han WD, Si YL, Zhao YL, Li Q, Wu ZQ, Hao HJ, Song HJ. GC-rich promoter elements maximally confers estrogen-induced transactivation of LRP16 gene through ERalpha/Sp1 interaction in MCF-7 cells. *J Steroid Biochem Mol Biol.* 2008 Mar;109(1-2):47-56

Yang B, Lu XC, Chi XH, Han WD, Yu L, Lou FD. [LRP16 gene function based on bioinformatic analysis]. *Ai Zheng.* 2009 Dec;28(12):1283-90

---

*This article should be referenced as such:*

Huret JL, Xue Y. t(11;21)(q13;q22). *Atlas Genet Cytogenet Oncol Haematol.* 2010; 14(10):990-991.

---