

Leukaemia Section

Short Communication

t(12;21)(q12;q22)

Jean-Loup Huret

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

Published in Atlas Database: January 2008

Online updated version: <http://AtlasGeneticsOncology.org/Anomalies/t1221q12q22ID1339.html>

DOI: 10.4267/2042/44406

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.
© 2009 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Clinics and pathology

Disease

Acute myeloid leukaemia (AML), M2 subtype.

Epidemiology

Only one case to date, a 76 year old male patient.

Prognosis

No data: the patient died, but no survival data was noted.

Cytogenetics

Additional anomalies

The t(12;21)(q12;q22) was the sole anomaly.

Genes involved and proteins

CPNE8

Location

12q12

Protein

CPNE8 is a member of the copines. Copines are highly conserved, widely expressed, calcium-dependent membrane binding proteins. They may have a role in membrane trafficking and mediate cellular processes by conferring calcium regulation to various signaling pathways. Copine 8 is strongly expressed in brain, heart, and prostate, and expressed at low level in most other tissues.

RUNX1

Location

21q22

Protein

Contains a RUNT binding domain and a trans-activation domain; forms heterodimers; nuclear localization; transcription factor; critical regulator of hematopoietic-cell development.

Result of the chromosomal anomaly

Hybrid gene

Description

5' RUNX1 (including the DNA binding domain) broken after exon 6 and fused to intron 2 of CPNE8. However, fusion is out of frame, and termination occurs after 2 amino acids from CPNE8, producing a truncated RUNX1.

Fusion protein

Description

The truncated RUNX1 includes the binding domain, but not the transactivation domain.

References

Maitra R, Grigoryev DN, Bera TK, Pastan IH, Lee B. Cloning, molecular characterization, and expression analysis of Copine 8. *Biochem Biophys Res Commun*. 2003 Apr 11;303(3):842-7

Ramsey H, Zhang DE, Richkind K, Burcoglu-O'Ral A, Hromas R. Fusion of AML1/Runx1 to copine VIII, a novel member of the copine family, in an aggressive acute myelogenous leukemia with t(12;21) translocation. *Leukemia*. 2003 Aug;17(8):1665-6

This article should be referenced as such:

Huret JL. t(12;21)(q12;q22). *Atlas Genet Cytogenet Oncol Haematol*. 2009; 13(2):132.