Leukaemia Section
Short Communication

t(7;21)(p22;q22)
Jean-Loup Huret

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

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Clinics and pathology

Disease
Acute myeloid leukemia (AML).

Phenotype / cell stem origin
M0 AML.

Epidemiology
Only one case to date, a 7-year-old boy.

Evolution
The patient relapsed; however, he is still alive, with a bone marrow graft, 10 years after diagnosis.

Genes involved and Proteins

UPS42

Protein
USP42 (ubiquitin specific protease 42), belongs to the ubiquitin specific protease family. Ubiquitins are highly conserved proteins. Ubiquitins target proteins for degradation in the proteasome. Some USPs, however, act in the opposite reaction. These ubiquitin specific proteases (cysteine proteases) are also called deubiquitinating enzymes. They cleave ubiquitin from ubiquitin-conjugated target proteins and may lead to protein stabilization. Usp42 can cleave ubiquitin from ubiquitinated substrates. Usp42 seems to be a deubiquitinating enzyme. It may play an important role in mouse embryogenesis.

RUNX1

Location: 21q22

Protein
Transcription factor (activator) for various hematopoietic-specific genes, which expression is limited to hematopoetic stem cells, and endothelial cells and mesenchymal cells in the embryo; core binding factor family member which forms heterodimers with CBFB; binds to the core site 5’ PyGPyGGTPy 3’ of promotors and enhancers.

Results of the chromosomal anomaly

Hybrid gene

Description
5’ RUNX1- 3’ UPS42

References


This article should be referenced as such: