

Leukaemia Section

Short Communication

t(7;21)(p22;q22) RUNX1/USP42

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Abstract

Review on t(7;21)(p22;q22) RUNX1/USP42, with data on clinics, and the genes involved.

Clinics and pathology

Disease

Acute myeloid leukemia M0 (M0 AML)

Phenotype/cell stem origin

M0 AML

Epidemiology

Three cases to date; a 7-year old male, a 32-year old male, and a 52-year old female

Evolution

The 7-year old male patient relapsed; however, he is still alive, with a bone marrow graft, 10 years after diagnosis. The 32-year old male died of alternative causes shortly after diagnosis. The female patient did not relapse, and is still alive.

Disease

Acute myeloid leukemia M4 (M4-AML)

Phenotype/cell stem origin

M4-AML

Epidemiology

Three cases to date, with the possibility of a fourth. Excluding fourth: 2 males (ages 39 and 13), and 1 female (age 57). The fourth case was a 54 year old male with either M4 or M5 subtype.

Evolution

39-year old male is dead from alternative causes, no relapse; 57-year old female and 13-year old male are both alive, with no evidence of relapse.

Disease

Acute myeloid leukemia M5 (M5-AML)

Phenotype/cell stem origin

M5/M5a-AML

Epidemiology

Two cases with the possibility of a third (overlap with aforementioned M4 cases). First case is a 33-year old male; second case is a 68-year old female. The third case is, as mentioned, a 54-year old male.

Evolution

The 33-year old male had no relapse and is still alive. The 68-year old female is dead, 5 years after diagnosis. The 54-year old male is, as mentioned, dead 3 months after diagnosis.

Genes involved and proteins

USP42 (ubiquitin specific peptidase 42)

Location

7p22.1

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 (runt-related transcription factor 1 (acute myeloid leukemia 1; aml1 oncogene))

Location

21q22.12

Protein

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

Result of the chromosomal anomaly

Hybrid gene

Description

5' RUNX1- 3' UPS42

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