

Leukaemia Section

Short Communication

t(8;21)(q24;q22)

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Clinics and pathology

Disease

T-cell acute lymphoblastic leukemia (ALL) and acute non lymphocytic leukemia (ANLL).

Phenotype/cell stem origin

1 case of T-cell ALL and 2 cases of ANLL, one of which was a M4.

Epidemiology

2 documented cases, male patients aged 5 yrs (ALL case) and 42 yrs (ANLL case).

Cytogenetics

Cytogenetics morphological

+21 (ALL case); complex karyotype (ANLL case).

Genes involved and proteins

Note

This translocation may be heterogeneous at the molecular level, as it is concerning the phenotype.

TRPS1

Location

8q24

Protein

Transcriptional repressor.

Germinal mutations

Involved in tricho-rhino-phalangeal syndrome.

Somatic mutations

Involved with AML1 in the M4-ANLL case.

AML1

Location

21q22

DNA/RNA

Transcription is from telomere to centromere

Protein

Contains a Runt domain and, in the C-term, a transactivation domain; forms heterodimers; widely expressed; nuclear localisation; transcription factor (activator) for various hematopoietic-specific genes.

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