t(12;21)(p12;q22)

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Published in Atlas Database: August 1997

Online version is available at: http://AtlasGeneticsOncology.org/Anomalies/t1221.html
DOI: 10.4267/2042/32031

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Clinics and pathology

Disease
B cell ALL

Phenotype / cell stem origin
L1 and L2, CD10+.

Epidemiology
15 to 35% of paediatric B-lineage ALL: so far the most frequent translocation in this group; rare or absent in adults and in infants; age: children; no case > 20 yrs so far; male and female equally represented.

Clinics
Standard ALL.

Prognosis
CR in all cases; prognosis seems good.

Cytogenetics

Cytogenetics, morphological
t(12;21) often remained undetected.

Cytogenetics, molecular
Easily detected by chromosomes 12 and 21 painting or specific probes.

Additional anomalies
Frequent del(12)(p12) on the other chromosome; in rare cases duplication of der(21)t(12;21); looks like a +21.

Variants
t(6;12;21), t(3;12;21)

Genes involved and Proteins

ETV6
Location: 12p13
DNA / RNA
9 exons; alternate splicing.
Protein
Contains a HLH domain and a ETS-DNA binding domain; wide expression; nuclear localisation; ETS-related transcription factor.

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.

Results of the chromosomal anomaly

Hybrid gene
Description
TEL-AML1 chimaeric gene; 5' centromere to 3' telomere orientation.
Transcript
The fusion transcript on chromosome 21 TEL-AML1 is the crucial one; the AML1-TEL transcript is absent in some cases; the other TEL allele is often deleted.
Detection protocol
RT-PCR of the fusion transcript.

**Fusion protein**

**Description**
Helix loop helix of TEL fused to the nearly entire AML1 protein, comprising the Runt domain and the transactivation domain.

**References**


This article should be referenced as such: