t(6;7)(q23;q34)
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Identity

R-band analysis. Partial karyotype showing t(6;7)(q23;q34).

Clinics and pathology

Disease
T cell acute lymphoblastic leukemia (T-ALL).

Phenotype / cell stem origin
T cell precursor.

Epidemiology
Less than 5% among a series of non selected adult and pediatric T-ALLs (n = 3 out of 92). Six cases were described, all of them children, and 5 out of 6 being under 3 years old (1.1, 1.3, 1.8, 2.5, and 2.9 years old, respectively), which is very young for T-cell leukemia. The t(6;7) translocation could therefore be relatively common in this very low range of age.

Cytology
Lymphoblasts.

Prognosis
The prognosis is yet to be evaluated.

Cytogenetics

Cytogenetics morphological
t(6;7)(q23;q34) may be barely detectable by chromosome banding technics alone.
**Cytogenetics molecular**
Involvement of the TCRB locus and the MYB locus can be demonstrated using flanking FISH probes.

**Genes involved and Proteins**

**TRB**
- **Location:** 7q34
- **Protein**
  - T-cell receptor beta chain.

**C-MYB**
- **Location:** 6q23.3
- **DNA / RNA**
  - Spans over 38 kb, 15 exons (and additional alternative exons), mRNA 3.3 kb.
- **Protein**
  - v-myb myeloblastosis viral oncogene homolog.
  - Transcription factor: 640 amino acids.

**AHI-1**
- **Location:** 6q23.3
- **DNA / RNA**
  - Spans over 214 kb, 28 exons (and additional alternative exons), mRNA 5.5 kb.
- **Protein**
  - Jouberin (Abelson helper integration site 1 protein homolog) (AHI-1). 1196 amino acids including one SH3 domain and WD repeats.

**Results of the chromosomal anomaly**

**Hybrid gene**
- **Note:** No fusion gene
- The t(6;7)(q23.3;q34) translocation results in juxtaposition of TRB regulatory sequences to the MYB-AHI1 locus. It results in deregulated expression of C-MYB, as demonstrated by skewed allelic expression.

**Fusion protein**

**Oncogenesis**

C-MYB is a transcription factor involved in hematopoiesis. In T-cell differentiation, discrete threshold levels of MYB activity regulate transition through distinct stages, suggesting that a deregulated expression could disturb the maturation process and play a role in oncogenesis.

A potential role of AHI1 deregulation as a cofactor has to be evaluated.

Of note, the same locus at 6q23.3 is also involved in short tandem duplications of a about 230 kb genomic region which includes the C-MYB gene (about 10% T-ALL in children and adults). This somatic abnormality can be detected by array-CGH, genomic Q-PCR or fiber-FISH, but not or hardly by standard metaphasic or interphasic FISH.

**References**


*This article should be referenced as such:*