t(1;12)(p36;p13) ETV6/PRDM16

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Abstract
Review on t(1;12)(p36;p13) translocations, with data on clinics, and the genes involved.

Keywords
chromosome 1; chromosome 12; t(1;12)(p36;p13); PRDM16; ETV6

Clinics and pathology

Disease
Acute myeloid leukaemia (AML).

Phenotype/cell stem origin
AML M4 according to immunophenotyping vs M1 according to cytology.

Epidemiology
Found in one case of AML in a 46 years old female Duhoux et al., 2012).

Cytology
Leukemic infiltrate of myeloid origin (blasts I and II); high proportion of myeloid blast cells and restricted percentage of monocytes.

Prognosis
The patient died 12 months after diagnosis.

Cytogenetics

Cytogenetics morphological
The t(1;12)(p36;p13.2) was the sole anomaly.

Genes involved and proteins

PRDM16 (PR domain containing 16)
Location
1p36.32
DNA/RNA
11 splice variants
Protein
1276 amino acids and smaller proteins. Contains a N-term PR domain; 7 Zinc fingers, a proline-rich domain, and 3 Zinc fingers in the C-term. Binds DNA. Transcription activator; PRDM16 has an intrinsic histone methyltransferase activity. PRDM16 forms a transcriptional complex with CEBPB. PRDM16 plays a downstream regulatory role in mediating TGFβ signaling (Bjork et al., 2010). PRDM16 induces brown fat determination and differentiation. PRDM16 is expressed selectively in the earliest stem and progenitor hematopoietic cells, and is required for the maintenance of the hematopoietic stem cell pool during development.
PRDM16 is also required for survival, cell-cycle regulation and self-renewal in neural stem cells (Chuikov et al., 2010; Kajimura et al., 2010; Aguilo et al., 2011; Chi and Cohen, 2016).

**ETV6 (ets variant 6)**

**Location**
12p13.2

**Protein**
452 amino acids. The ETV6 protein contains from N-term to C-Term a HLH (helix-loop-helix, aa 40-124) domain (also referred to as the pointed or sterile alpha motif domain), responsible for hetero- and homodimerization, an internal domain, involved in the recruitment of a repression complex including NCOR1 (17p12), NCOR2 (12q24.31), SIN3A (15q24.2), and SIN3B (19p13.11), and an ETS domain (aa 339-420), responsible for sequence specific DNA-binding and protein-protein interaction (De Braekeleer et al., 2014).

**Result of the chromosomal anomaly**

**Hybrid gene**

**Description**
ETV6 exon 4 is fused to PRDM16 exon 2.

**Detection**
PCR, FISH.
References


This article should be referenced as such: