Leukaemia Section
Short Communication

t(1;17)(p36;q21) WNT3 or NSF/PRDM16

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Abstract
Review on t(1;17)(p36;q21) translocation, with data on clinics, and the genes involved.

Keywords
chromosome 1; chromosome 17; t(1;17)(p36;q21); PRDM16; WNT3; NSF

Clinics and pathology

Disease
Acute myeloid leukaemia.

Clinics
Only one case to date, a 5-year-old girl who presented with acute myeloid leukaemia. She died 6 months after diagnosis (Duhoux et al., 2012).

Cytogenetics

Cytogenetics morphological
The t(1;17)(p36;q21) was the sole anomaly.

Genes involved and proteins

PRDM16, on chromosome 1, was implicated in the translocation. However, which gene on chromosome 17 is involved in the malignant process is unknown: two genes are located in the vicinity of the 17q21 breakpoint: WNT3 (44839872 - 44896126 bp from pter) and NSF (44668035 - 44834828 bp from pter). WNT3 (17q21.31), 355 amino acids, is one of the 19 known Wnt proteins. They bind a frizzled (Fz)/low density lipoprotein receptor related protein (LRP) complex, activating the cytoplasmic protein dishevelled (DVL1, DVL2 or DVL3), and the b-catenin Wnt/beta catenin signaling pathway is activated (Thorstensen and Lothe, 2003). NSF (17q21.31), 744 amino acids, is a hexameric ATPase. NSF catalyzes the fusion of transport vesicles. Vesicle fusion is driven by specific associations of complementary SNARE proteins (soluble NSF attachment protein receptor) residing on the vesicle (v-SNAREs) and target (t-SNAREs) membranes. This mechanism involves NSF and its adaptor protein, NAPA. This mechanism of vesicle fusion include endoplasmic reticulum-Golgi transport, intra-Golgi vesicle fusion, trafficking from the trans-Golgi network to the plasma membrane, neuromediator exocytosis, and synaptic vesicle fusion (Naydenov and Ivanov, 2013).

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).

**Result of the chromosomal anomaly**

**Fusion protein**

**Oncogenesis**

PRDM16 was not overexpressed (Duhoux et al., 2012)

**References**


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