

# **Gene Section**

## **Short Communication**

# TFPT (TCF3/E2A fusion partner)

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## **Identity**

Other names: FB1 HGNC (Hugo): TFPT Location: 19q13.4



Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

## DNA/RNA

#### Description

Genomic structure of six exons and five introns spanning about 10 kb.

#### **Transcription**

Two transcripts of 1.1 kb and 1.2 kb, expressed mainly in brain and in hemopoietic cell lines. The rat ortholog, Amida, was found highly expressed also in rat testis. TATA less promoter, that can be transactivated "in vitro" by PU.1 and Ikaros 2. Orientation, minus strand.

### **Protein**

#### Description

Conserved protein of 253 amino acids (in man) with two nuclear localization signals (NLS) (n.68-75 and n.190-194) and a DNA binding domain located between the two NLSs.

#### **Expression**

Constitutive.

#### Localisation

Nuclear.

#### **Function**

Overexpression of TFPT/Amida in cultured cells induces arrest in G2-M and apoptosis. Biochemical studies indicate that TFPT/Amida interacts with Cdc2/CDK1 in mitosis and its overexpression results in a decrease of Cdc2/CDK1 activity (5).It is also suggested that the TFPT/Amida DNA binding activity is necessary for cell cycle inhibition and that Amida phophorylation by Cdc2/CDK1, detected "in vitro", might decrease this DNA binding activity.

#### Homology

Very high homology with mouse and rat orthologs.

### **Mutations**

#### Somatic

Involved in chromosome rearrangement in leukaemia.

# Implicated in

#### Childhood pre-B ALL

#### Note

We detected 8 cases out of 200: incidence about 4% of childhood pre-B ALL.

#### Cytogenetics

Following the position of the two involved genes, E2A on 19p13 and FB1 on 19q13 an inv(19)(p13q13) appears more likely but a translocation t(19;19)(p13;q13) cannot be yet ruled out. Still pending.

#### Hybrid/Mutated gene

E2A-FB1.

#### **Abnormal protein**

Since the chimeric transcripts so far analyzed contain

the FB1 sequence fused out of frame to E2A and no truncated E2A protein was detected by Western blot, we suggest that no fusion protein is produced.

## **Breakpoints**

#### Note

We detected different joining points in the transcripts of the different analyzed cases indicating different breakpoints in a genomic region spanning exons 15-17 on TCF3 and exons 5-6 on TFPT.

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