

## Gene Section

### Mini Review

# PTPN7 (protein tyrosine phosphatase, non-receptor type 7)

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## Identity

**Other names:** BPTP-4; HEPTP; LC-PTP; LPTP; PTPNI

**HGNC (Hugo):** PTPN7

**Location:** 1q32.1

## DNA/RNA

### Description

The premessenger has 10 exons and covers 14.59 kb on the genome.

### Transcription

The complete mRNA is 3784 bp long. 2 alternatively spliced transcript variants encoding different isoforms have been found, but it has also been reported that transcription produces 16 different mRNAs, 15 alternatively spliced variants and 1 unspliced form. Of the 2 described variants, variant 1 (2,805 bp linear mRNA) contains a different 5' region, which includes a part of the coding sequence when compared to variant 2. Variant 2 (3,263 bp linear mRNA) contains an alternate 5' region, which includes an additional in-frame translation start codon, as compared to variant 1. It thus encodes a protein that is 39 aa longer at the N-terminus.

### Pseudogene

No pseudogenes have been found.

## Protein

### Description

The hematopoietic protein tyrosine phosphatase (HePTP) protein is a 40,5 kDa protein of 360 amino acids. It is a class I non-receptor PTP that is strongly expressed in T cells. It is composed of a C-terminal classical PTP domain (residues 44-339) and a short N-terminal extension (residues 1-43) that functions to direct HePTP to its physiological substrates.

### Expression

Thymus, spleen, leukocytes.

### Localisation

Cytoplasmic.

### Function

Protein tyrosine phosphatase activity, hydrolase activity, phosphoric monoester hydrolase activity, receptor activity- participation in MAPK signaling pathways, T cell receptor signaling pathway and protein amino acid dephosphorylation.

The protein can interact with tyrosine-phosphorylated MAPK1, MAPK3 and several other MAP kinases and suppress the MAP kinase activities. Plays a role in the regulation of T and B-lymphocyte development and signal transduction.

### Homology

HePTP has high homologies with striatal-enriched

phosphatase (STEP) and PCPTP (PC12 protein Tyr phosphatase).

## Mutations

### **Germinal**

No germline mutations are described.

### **Somatic**

Mutations have not been observed.

## Implicated in

### **Acute leukemia**

#### **Disease**

Myelodysplastic syndrome and myelogenous leukemia; HePTP often is dysregulated in the preleukemic disorder myelodysplastic syndrome and myelogenous leukemia (elevated expression of HePTP). The first indication of a role of HePTP in cell proliferation or differentiation came from the finding that the HePTP gene is located on the long arm of chromosome 1, which is often found in extra copies (trisomy) in bone marrow cells from patients with myelodysplastic syndrome, which is characterized by reduced hematopoiesis and increased risk of acute leukemia.

### **Non-Hodgkin Lymphoma**

#### **Disease**

Pediatric lymphoma; HePTP is down-regulated in pediatric lymphoma compared to control lymphoid cells. Loss of HePTP might indicate increased cell proliferation and/or survival of lymphoma cells.

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