

# Atlas of Genetics and Cytogenetics in Oncology and Haematology

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[Home](#) [Genes](#) [Leukemias](#) [Tumors](#) [Cancer prone](#) [Deep Insight](#) [Case Reports](#) [Portal](#)  
[Journals](#) [Teaching](#)

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[X](#) [Y](#) [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [NA](#)

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## Gene Form for submission

Select all, copy and past this form in a ClarisWorks or Word file, save as a model, ... ready for use

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DATE 2010-01-22

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UPDATE

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### Identity ----->

GENE NAME PTPN7

ALIAS Other names: BPTP-4, HEPTP, LC-PTP, LPTP, PTPN1

HUGO NAME protein tyrosine phosphatase, non-receptor type 7

LOCATION 1q32.1

LOCAL ORDER

## NOTE

To which CATEGORY this gene/protein belongs? Please, tick the box(es)

- Cell cycle	<input type="checkbox"/>	- Extracellular matrix	<input type="checkbox"/>
- Apoptosis (extrinsic)	<input type="checkbox"/>	- Cytoskeleton / scaffold	<input type="checkbox"/>
- Apoptosis (intrinsic)	<input type="checkbox"/>	- Cell junctions	<input type="checkbox"/>
		- Channels and gap junctions	<input type="checkbox"/>
- DNA replication and recombination	<input type="checkbox"/>	- Exocytosis-endocytosis, vesicle traffic	<input type="checkbox"/>
- DNA repair	<input type="checkbox"/>		
- Nucleole machinery	<input type="checkbox"/>	- Signal: Nuclear receptors	<input type="checkbox"/>
- Transcription-translation (transcription)	<input type="checkbox"/>	- Signal: Secreted factors	<input type="checkbox"/>
- Transcription-translation (spliceosome)	<input type="checkbox"/>	- Signal: Membrane receptors	<input type="checkbox"/>
- Transcription-translation (translation)	<input type="checkbox"/>	- Signal: Intracellular transduction: AMPc signaling	
- Micro RNA	<input type="checkbox"/>	- Signal: Intracellular transduction: PLCG signaling	<input type="checkbox"/>
- Chromatin	<input type="checkbox"/>	- Signal: Intracellular transduction: RAS / RAF / MAPK signaling	x
- Nuclear membrane trafficking	<input type="checkbox"/>	- Signal: Intracellular transduction: PI3K / AKT / mTOR signaling	<input type="checkbox"/>
- Mitosis / centrosome-microtubules-kinetochore	<input type="checkbox"/>	- Signal: Intracellular transduction: JAK / STAT signaling	<input type="checkbox"/>
		- Signal: Intracellular transduction: TGFb signaling	<input type="checkbox"/>
- Mitochondria (oxydative phosphorylation)	<input type="checkbox"/>	- Signal: Intracellular transduction: WNT signaling	<input type="checkbox"/>
- Mitochondria (TOM and TIM)	<input type="checkbox"/>	- Signal: Intracellular transduction: Hedgehog signaling	<input type="checkbox"/>
- Proteasome - ubiquitination	<input type="checkbox"/>	- Signal: Intracellular transduction: NF-kB signaling	<input type="checkbox"/>
		- Signal: Transcription factors	x
- Immunity	x		
- Angiogenesis	<input type="checkbox"/>		
- OTHER:			

**DNA ----->**

NOTE

DIAGRAM (see below)

LEGEND DIAGRAM

**DNA 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 psuedogenes have been found

**Protein ----->**

NOTE

DIAGRAM

LEGEND DIAGRAM

**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 ----->

NOTE

GERMINAL No germline mutations are described.

SOMATIC Mutations have not been observed.

DIAGRAM

LEGEND DIAGRAM

## Implicated in ----->

NAME Acute leukemia

NOTE

DISEASES 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.

CYTOGENETICS

HYBRID GENE

DIAGRAM

LEGEND DIAGRAM

FUSION PROTEIN

DIAGRAM

LEGEND DIAGRAM

ONCOGENESIS

... again if necessary :

NAME Non-Hodgkin Lymphoma

NOTE

DISEASES 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.

PROGNOSIS

CYTOGENETICS

HYBRID GENE

DIAGRAM

LEGEND DIAGRAM

FUSION PROTEIN

DIAGRAM

LEGEND DIAGRAM

ONCOGENESIS

### **Breakpoints** (if variables) ----->

DIAGRAM

LEGEND DIAGRAM

NOTE

### **To be noted** (specific items) ----->

TO BE NOTED

**External links** (addresses are in the Database section of the Atlas; find the Hugo name (or, else, GDB), the LocusLink name and number, and SwissProt ID; we will automatically find other links; please, add specific databases) ----->

HUGO PTPN7 9659

LOCUSLINK Entrez Gene (NCBI) PTPN7 5778  
protein tyrosine phosphatase, non-receptor type 7

OTHER DATABASES SwissProt ID P35236

NOTE

## References (PLEASE, see below and conform to the style) ----->

TITLE Cloning and expression of an inducible lymphoid-specific, protein tyrosine phosphatase (HePTPase)

AUTHORS Zanke B, Suzuki H, Kishihara K, Mizzen L, Minden M, Pawson A, Mak TW.

REFERENCE Eur. J. Immunol. 1992. 22(1):235-9.

PUBMED PMID: 1530918

TITLE Structure of the human LC-PTP (HePTP) gene: similarity in genomic organization within protein-tyrosine phosphatase genes.

AUTHORS Adachi M, Miyachi T, Sekiya M, Hinoda Y, Yachi A, Imai K.

REFERENCE Oncogene. 1994. 9(10):3031-5.

PUBMED PMID: 8084610

TITLE A hematopoietic protein tyrosine phosphatase (HePTP) gene that is amplified and overexpressed in myeloid malignancies maps to chromosome 1q32.1

AUTHORS Zanke B, Squire J, Griesser H, Henry M, Suzuki H, Patterson B, Minden M, Mak TW.

REFERENCE Leukemia. 1994. 8(2):236-44.

PUBMED PMID: 8309248

TITLE Cloning and expression of PCPTP1 encoding protein tyrosine phosphatase.

AUTHORS Shiozuka K, Watanabe Y, Ikeda T, Hashimoto S, Kawashima H.

REFERENCE Gene. 1995. 11(162):279-84.

PUBMED PMID: 7557444

TITLE Negative regulation of T cell antigen receptor signal transduction by hematopoietic tyrosine phosphatase (HePTP).

AUTHORS Saxena M, Williams S, Gilman J, Mustelin T.

REFERENCE J. Biol. Chem. 1998. 19(273):15340-4.

PUBMED PMID: 9624114

TITLE The next wave: Protein tyrosine phosphatases enter T cell antigen receptor signalling.

AUTHORS Mustelin T, Brockdorff J, Rudbeck L, Gjørloff Wingren A, Han S, Wang, X, Taylor P, Saxena M.

REFERENCE Cell. Signal. 1999. 11(9):637-50. REVIEW.

PUBMED PMID: 10530872

TITLE Direct suppression of TCR-mediated activation of extracellular signal-regulated kinase by leukocyte protein tyrosine phosphatase, a tyrosine specific phosphatase.

AUTHORS Oh-Hora M, Ogata M, Mori Y, Adachi M, Imai K, Kosugi A, Hamaoka T.

REFERENCE J. Immunol. 1999. 163(3):1282-8.

PUBMED PMID: 10415025

TITLE Inhibition of T cell signaling by mitogen-activated protein kinase-targeted hematopoietic tyrosine phosphatase (HePTP).

AUTHORS Saxena M, Williams S, Brockdorff J, Gilman J, Mustelin T.

REFERENCE J. Biol. Chem. 1999. 23(274):11693-700.

PUBMED PMID: 10206983

TITLE Subcellular localization of intracellular protein tyrosine phosphatases in T cells.

AUTHORS Gjørloff Wingren A, Saxena M, Han S, Wang X, Alonso A, Renedo M, Oh P, Williams S, Schnitzer J, Mustelin T.

REFERENCE Eur. J. Immunol. 2000. 30(8):2412-21.

PUBMED PMID: 10940933

TITLE The MAP-kinase ERK2 is a specific substrate of the protein tyrosine phosphatase HePTP.

AUTHORS Pettiford SM, Herbst R.

REFERENCE Oncogene 2000 19(7):858-69.

PUBMED PMID: 10702794

TITLE

Structure of the hematopoietic tyrosine phosphatase (HePTP)catalytic domain: structure of a KIM phosphatase with phosphate bound at the active site.

AUTHORS Mustelin T, Tautz L, Page R.

REFERENCE J. Mol. Biol. 2005. 18;354(1):150-63.

PUBMED PMID: 16226275

TITLE Immunohistochemical analyses of phosphatases in childhood B-cell lymphoma: lower expression of PTEN and HePTP and higher number of positive cells for nuclear SHP2 in B-cell lymphoma cases compared to controls.

AUTHORS Fridberg M, Kjellström S, Anagnostaki L, Skogvall I, Mustelin T, Wiebe T, Persson JL, Dictor M, Gjörlöf Wingren A.

REFERENCE Pediatric Hematol. And Oncol. 2008, 25:528-40.

PUBMED PMID: 18728972

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- Style to be used for References:

\* classified by years, from the oldest to the most recent, and by alphabetical order within a year

\* style:

Title.

Author1 AB, Author2 CD, Author3 E.

Journal. Year date;Vol(Number):first page-last page. (add: REVIEW if it is)

PubMed number

Example:

All teddy bears are gentle persons.

Paddington B, The Pooh W.

Int J Teddy Bears. 2003 Dec 25;12(12):501-9. (REVIEW)

PMID 1642873591

- For Images, please:

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\* Please use preferably Arial 9 point for lettering.

\* Images should be sent by e-mail, preferably in PNG, JPG or PSD (for photographs: chromosomes, histo-pathology, radiographs, ...); PPT or PNG, PSD, SVG, EPS, WMF, JPG, GIF (for drawings); the desk may have problems with other formats.

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