

## Product datasheet

# Recombinant Human Inosine triphosphate pyrophosphatase protein ab123470

### Overview

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<b>Product name</b>	Recombinant Human Inosine triphosphate pyrophosphatase protein
<b>Protein length</b>	Full length protein

### Description

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<b>Nature</b>	Recombinant
<b>Source</b>	Escherichia coli
<b>Amino Acid Sequence</b>	
<b>Accession</b>	<a href="#">Q9BY32</a>
<b>Species</b>	Human
<b>Sequence</b>	MGSSHHHHHH SSGLVPRGSH MMAASLVGKK IVFVTGNAKK LEEVVQILGD KFPCTLVAQK IDLPEYQGEP DEISIQKCQE AVRQVQGPVL VEDTCLCFNA LGGLPGPYIK WFLEKLPPEG LHQLLAGFED KSAYALCTFA LSTGDPSQPV RLFGRGRTSGR IVAPRGCQDF GWDPCFQPDG YEQTYAEMPK AEKNAVSHRF RALLELQEYF GSLAA
<b>Molecular weight</b>	24 kDa including tags
<b>Amino acids</b>	1 to 194
<b>Tags</b>	His tag N-Terminus

### Specifications

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Our [Abpromise guarantee](#) covers the use of **ab123470** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

<b>Applications</b>	SDS-PAGE
<b>Purity</b>	> 95 % SDS-PAGE. ab123470 was purified by proprietary chromatographic techniques and filter sterilized.
<b>Form</b>	Liquid
<b>Additional notes</b>	Although stable at 4°C for 1 week, ab123470 should be stored desiccated below -18°C. Please prevent freeze thaw cycles

## Preparation and Storage

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### Stability and Storage

Shipped at 4°C. Please see notes section.

pH: 8.00

Constituents: 0.24% Tris, 10% Glycerol

## General Info

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### Function

Hydrolyzes ITP and dITP to their respective monophosphate derivatives. Xanthosine 5'-triphosphate (XTP) is also a potential substrate. May be the major enzyme responsible for regulating ITP concentration in cells.

### Tissue specificity

Ubiquitous. Highly expressed in heart, liver, sex glands, thyroid and adrenal gland.

### Involvement in disease

Defects in ITPA are the cause of inosine triphosphate pyrophosphohydrolase deficiency (ITPA deficiency) [MIM:147520]. It is a common inherited trait characterized by the abnormal accumulation of inosine triphosphate (ITP) in erythrocytes and also leukocytes and fibroblasts. The pathological consequences of ITPA deficiency, if any, are unknown. However, it might have pharmacogenomic implications and be related to increased drug toxicity of purine analog drugs. Three different human populations have been reported with respect to their ITPase activity: high, mean (25% of high) and low activity. The variant Thr-32 is associated with complete loss of enzyme activity, may be by altering the local secondary structure of the protein. Heterozygotes for this polymorphism have 22.5% of the control activity: this is consistent with a dimeric structure of the enzyme.

### Sequence similarities

Belongs to the HAM1 NTPase family.

### Cellular localization

Cytoplasm.

**Please note:** All products are "FOR RESEARCH USE ONLY AND ARE NOT INTENDED FOR DIAGNOSTIC OR THERAPEUTIC USE"

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