abcam

Product datasheet

Recombinant Human Inosine triphosphate pyrophosphatase protein ab123470

1 References

Overview

Product name	Recombinant Human Inosine triphosphate pyrophosphatase protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Escherichia coli

Amino Acid Sequence

Accession	Q9BY32
Species	Human
Sequence	MGSSHHHHHH SSGLVPRGSH MMAASLVGKK IVFVTGNAKK LEEVVQILGD KFPCTLVAQK IDLPEYQGEP DEISIQKCQE AVRQVQGPVL VEDTCLCFNA LGGLPGPYIK WFLEKLKPEG LHQLLAGFED KSAYALCTFA LSTGDPSQPV RLFRGRTSGR IVAPRGCQDF GWDPCFQPDG YEQTYAEMPK AEKNAVSHRF RALLELQEYF GSLAA
Molecular weight	24 kDa including tags
Amino acids	1 to 194
Tags	His tag N-Terminus

Specifications

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.

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

prevent freeze thaw cycles

Preparation and Storage

Stability and Storage

Shipped at 4°C. Please see notes section.

pH: 8.00
Constituents: 0.24% Tris, 10% Glycerol

General Info

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.

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