abcam

Product datasheet

Recombinant Human PRPS1 protein ab88987

概述

产品名称	重组人PRPS1蛋白
蛋白长度	Full length protein

描述

性质	Recombinant
来源	Saccharomyces cerevisiae

氨基酸序列

种属	Human
序列	MPNIKIFSGSSHQDLSQKIADRLGLELGKVVTKKFSNQETCVEIGESVRG EDVYIVQSGCGEINDNLM ELLIMINACKIASASRVTAVIPCFPYARQD KKDKSRAPISAKLVANMLSVAGADHIITMDLHASQIQG FFDIPVDNLY AEPAVLKWIRENISEWRNCTIVSPDAGGAKRVTSIADRLNVDFALIHKER KKANEVDR MVLVGDVKDRVAILVDDMADTCGTICHAADKLLSAGATRV YAILTHGIFSGPAISRINNACFEAVVVT NTIPQEDKMKHCSKIQVIDI SMILAEAIRRTHNGESVSYLFSHVPL

技术指标

Our Abpromise guarantee covers the use of **ab88987** in the following tested applications.

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

应用	SDS-PAGE
纯度	> 90 % SDS-PAGE. ab88987 was purified using affinity chromatography
形式	Liquid

制备和贮存

稳定性和存储	Shipped at 4°C. Store at -20°C. Stable for 12 months at -20°C.

Preservative: None
Constituents: 30% Glycerol, 0.5% Triton-X-100, 50mM HEPES, 30mM Glutathione, 100mM Sodium chloride, 1mM DTT, pH 7.5

常规信息

功能

Catalyzes the synthesis of phosphoribosylpyrophosphate (PRPP) that is essential for nucleotide synthesis.

通路

Metabolic intermediate biosynthesis; 5-phospho-alpha-D-ribose 1-diphosphate biosynthesis; 5-phospho-alpha-D-ribose 1-diphosphate from D-ribose 5-phosphate (route I): step 1/1.

疾病相关

Defects in PRPS1 are the cause of phosphoribosylpyrophosphate synthetase superactivity (PRPS1 superactivity) [MIM:300661]; also known as PRPS-related gout. It is a familial disorder characterized by excessive purine production, gout and uric acid urolithiasis.
Defects in PRPS1 are the cause of Charcot-Marie-Tooth disease X-linked recessive type 5 (CMTX5) [MIM:311070]; also known as optic atrophy-polyneuropathy-deafness or Rosenberg-Chutorian syndrome. CMTX5 is a form of Charcot-Marie-Tooth disease, the most common inherited disorder of the peripheral nervous system. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies characterized by severely reduced motor nerve conduction velocities (NCVs) (less than 38m/s) and segmental demyelination and remyelination, and primary peripheral axonal neuropathies characterized by normal or mildly reduced NCVs and chronic axonal degeneration and regeneration on nerve biopsy.
Defects in PRPS1 are the cause of ARTS syndrome (ARTS) [MIM:301835]; also known as fatal ataxia X-linked with deafness and loss of vision. ARTS is a disorder characterized by mental retardation, early-onset hypotonia, ataxia, delayed motor development, hearing impairment, and optic atrophy. Susceptibility to infections, especially of the upper respiratory tract, can result in early death.
Defects in PRPS1 are the cause of deafness X-linked type 1 (DFNX1) [MIM:304500]; also known as congenital sensorineural deafness X-linked 2 (DFN2). It is a form of deafness characterized by progressive, severe-to-profound sensorineural hearing loss in males. Females manifest mild to moderate hearing loss.

序列相似性

Belongs to the ribose-phosphate pyrophosphokinase family.

Terms and conditions

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