abcam

Product datasheet

# Recombinant Human PRPS1 protein ab92935

1 图像

## 描述

| | |
|---|---|
| **产品名称** | 重组人PRPS1蛋白 |
| **纯度** | > 90 % SDS-PAGE.<br>ab92935 is purified using conventional chromatography techniques. |
| **表达系统** | Escherichia coli |
| **蛋白长度** | Full length protein |
| **无动物成分** | No |
| **性质** | Recombinant |
| **种属** | Human |
| **序列** | MGSSHHHHHH SSGLVPRGSH MPNIKIFSGS<br>SHQDLSQKIA DRLGLELGKV VTKKFSNQET<br>CVEIGESVRG EDVYIVQSGC GEINDNLMEL<br>LIMINACKIA SASRVTAVIP CFPYARQDKK<br>DKSRAPISAK LVANMLSVAG ADHIITMDLH<br>ASQIQGFFDI PVDNLYAEPA VLKWIRENIS<br>EWRNCTIVSP DAGGAKRVTS IADRLNVDFA<br>LIHKERKKAN EVDRMVLVGD VKDRVAILVD<br>DMADTCGTIC HAADKLLSAG ATRVYAILTH<br>GIFSGPAISR INNACFEAVV VTNTIPQEDK<br>MKHCSKIQVI DISMILAEAI RRTHNGESVS YLFSHVPL |

## 技术指标

Our **Abpromise guarantee** covers the use of **ab92935** 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 |
| **形式** | Liquid |

## 制备和贮存

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|---|---|
| **稳定性和存储** | Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.<br>pH: 8.00 |

Constituents: 0.0154% DTT, 0.316% Tris HCl, 20% Glycerol (glycerin, glycerine), 0.58% Sodium chloride

## 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.

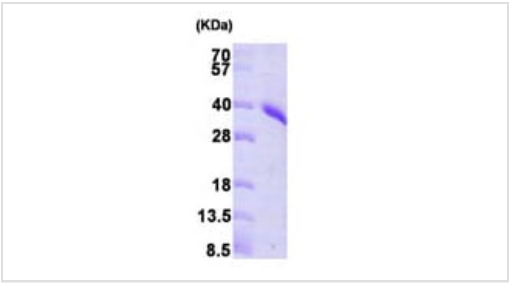
**疾病相关**

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.

## 图片




SDS-PAGE - Recombinant Human PRPS1 protein (ab92935)

15% SDS-PAGE showing ab92935 (3µg) at approximately 36.9kDa.

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