磷酸核糖焦磷酸合成酶 1 抗体

产品货号： mlR19407

英文名称： PRPS1

中文名称： 磷酸核糖焦磷酸合成酶 1 抗体

别　　名： ARTS; CMTX5; Deafness 2 perceptive congenital; Deafness X linked 2 perceptive congenital; DFN2; DFNX1; EC 2.7.6.1; KIAA0967; Phosphoribosyl pyrophosphate synthase I; Phosphoribosyl pyrophosphate synthetase I; PPRibP; Prps1; PRPS1_HUMAN; PRS I; PRS-I; PRSI; Ribose phosphate pyrophosphokinase I; Ribose-phosphate pyrophosphokinase 1.

研究领域： 肿瘤　细胞生物　神经生物学　转录调节因子　表观遗传学

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Cow, Xenopus laevis, Xenopus tropicalis

产品应用： IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）
not yet tested in other applications.
optimal dilutions/concentrations should be determined by the end user.

分 子 量： 34kDa

细胞定位： 细胞浆

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human PRPS1:101-200/318

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 ° C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20° C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 ° C.

PubMed： PubMed

产品介绍： This gene encodes an enzyme that catalyzes the phosphoribosylation of ribose 5-phosphate to 5-phosphoribosyl-1-pyrophosphate, which is necessary for purine metabolism and nucleotide biosynthesis. Defects in this gene are a cause of phosphoribosylpyrophosphate synthetase superactivity, Charcot-Marie-Tooth disease X-linked recessive type 5 and Arts Syndrome. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Feb 2011]

Function:

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

DISEASE:

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.

Similarity:

Belongs to the ribose-phosphate pyrophosphokinase family.

SWISS:

P60891

Gene ID:

5631

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.