

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

# 腺嘌呤磷酸核糖转移酶抗体

产品货号	:	mlR23830
英文名称	:	APRT
中文名称	:	腺嘌呤磷酸核糖转移酶抗体
别 名 APT_HUM <i>A</i>		Adenine phosphoribosyltransferase; AMP; AMP diphosphorylase; AMP pyrophosphorylase; APRT; DKFZp686D13177; MGC125856; MGC125857; MGC129961; Transphosphoribosidase.
研究领域	:	肿瘤 细胞生物 免疫学 神经生物学 新陈代谢
抗体来源	:	Rabbit
克隆类型	:	Polyclonal
交叉反应	:	Human, Mouse, Rat, Pig,
		WB=1:500-2000 ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 所依抗原修复)



カ I 里 · ISKDa	分	子	量	:	19kDa
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细胞定位: 细胞浆

性 状: Lyophilized or Liquid

浓 度: 1mg/1ml

免疫原: KLH conjugated synthetic peptide derived from human APRT:171-270/574 <Extracellular>

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  $^{\circ}$  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 $^{\circ}$  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  $^{\circ}$  C.

PubMed: PubMed

产品介绍: APRT is a 180 amino acid protein that localizes to the cytoplasm and belongs to the purine/pyrimidine phosphoribosyltransferase family. Existing as a homodimer, APRT functions to catalyze the



formation of inorganic pyrophosphate and AMP from adenine and 5-phosphoribosyl-1-pyrophosphate (PRPP), a reaction that is essential for both purine metabolism and AMP biosynthesis. Defects in the gene encoding APRT are the cause of APRT deficiency, also known as 2,8-dihydroxyadenine urolithiasis, which is an autosomal recessive disease that results in renal failure. The gene encoding APRT maps to human chromosome 16, which encodes over 900 genes and comprises nearly 3% of the human genome. The GAN gene is located on chromosome 16 and, with mutation, may lead to giant axonal neuropathy, a nervous system disorder characterized by increasing malfunction with growth. The rare disorder Rubinstein-Taybi syndrome is also associated with chromosome 16, as is Crohn's disease, which is a gastrointestinal inflammatory condition.

#### **Function:**

Catalyzes a salvage reaction resulting in the formation of AMP, that is energically less costly than de novo synthesis.

#### **Subunit:**

Homodimer.

## Subcellular Location:

Cytoplasm.

### DISEASE:

Defects in APRT are the cause of adenine phosphoribosyltransferase deficiency (APRTD) [MIM:102600]; also known as 2,8-dihydroxyadenine urolithiasis. An enzymatic deficiency that can lead to urolithiasis and renal failure. Patients have 2,8-dihydroxyadenine (DHA) urinary stones.

#### Similarity:

Belongs to the purine/pyrimidine phosphoribosyltransferase family.



c١	A	IC	c.

P07741

## Gene ID:

353

## Important Note:

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

# 产品图片

