

APRT Rabbit pAb

Catalog # AP99746

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	P07741
Other Accession	P07741
Host	Rabbit
Clonality	Polyclonal
Calculated MW	19608
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human APRT
Epitope Specificity	101-180/180
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the purine/pyrimidine phosphoribosyltransferase family.
SUBUNIT	Homodimer.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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.

Additional Information

Gene ID	353
Other Names	APT_HUMAN; APRT; 2.4.2.7; APT_RAT; adenine phosphoribosyltransferase
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500

Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.
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Protein Information

Name	APRT (HGNC:626)
Function	Catalyzes a salvage reaction resulting in the formation of AMP, that is energetically less costly than de novo synthesis.
Cellular Location	Cytoplasm.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.