



APRT (I-12): sc-107414

BACKGROUND

APRT (adenine phosphoribosyltransferase) 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 malformation 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.

REFERENCES

- Holden, J.A., Meredith, G.S. and Kelley, W.N. 1979. Human adenine phosphoribosyltransferase. Affinity purification, subunit structure, amino acid composition, and peptide mapping. *J. Biol. Chem.* 254: 6951-6955.
- Hidaka, Y., Tarle, S.A., O'Toole, T.E., Kelley, W.N. and Palella, T.D. 1987. Nucleotide sequence of the human APRT gene. *Nucleic Acids Res.* 15: 9086.
- Broderick, T.P., Schaff, D.A., Bertino, A.M., Dush, M.K., Tischfield, J.A. and Stambrook, P.J. 1987. Comparative anatomy of the human APRT gene and enzyme: nucleotide sequence divergence and conservation of a nonrandom CpG dinucleotide arrangement. *Proc. Natl. Acad. Sci. USA* 84: 3349-3353.
- Kamatani, N., Kuroshima, S., Terai, C., Hidaka, Y., Palella, T.D. and Nishioka, K. 1989. Detection of an amino acid substitution in the mutant enzyme for a special type of adenine phosphoribosyltransferase (APRT) deficiency by sequence-specific protein cleavage. *Am. J. Hum. Genet.* 45: 325-331.
- Chen, J., Sahota, A., Laxdal, T., Scrine, M., Bowman, S., Cui, C., Stambrook, P.J. and Tischfield, J.A. 1991. Identification of a single missense mutation in the adenine phosphoribosyltransferase (APRT) gene from five Icelandic patients and a British patient. *Am. J. Hum. Genet.* 49: 1306-1311.
- Menardi, C., Schneider, R., Neuschmid-Kaspar, F., Klocker, H., Hirsch-Kauffmann, M., Auer, B. and Schweiger, M. 1997. Human APRT deficiency: indication for multiple origins of the most common Caucasian mutation and detection of a novel type of mutation involving intrastrand-templated repair. *Hum. Mutat.* 10: 251-255.
- Taniguchi, A., Hakoda, M., Yamanaka, H., Terai, C., Hikiji, K., Kawaguchi, R., Konishi, N., Kashiwazaki, S. and Kamatani, N. 1998. A germline mutation abolishing the original stop codon of the human adenine phosphoribosyltransferase (APRT) gene leads to complete loss of the enzyme protein. *Hum. Genet.* 102: 197-202.

CHROMOSOMAL LOCATION

Genetic locus: Aprt (mouse) mapping to 8 E1.

SOURCE

APRT (I-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of APRT of mouse origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-107414 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

APRT (I-12) is recommended for detection of APRT of mouse and rat origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for APRT siRNA (m): sc-141179, APRT shRNA Plasmid (m): sc-141179-SH and APRT shRNA (m) Lentiviral Particles: sc-141179-V.

Molecular Weight of APRT: 20 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, ****DO NOT FREEZE****. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

See our web site at www.scbt.com or our catalog for detailed protocols and support products.