

MTHFR Rabbit pAb

Catalog # AP99264

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	P42898
Other Accession	P42898
Host	Rabbit
Clonality	Polyclonal
Calculated MW	74597
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human MTHFR
Epitope Specificity	165-300/656
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 methylenetetrahydrofolate reductase family.
SUBUNIT	Homodimer.
DISEASE	Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. Note=The disease is caused by mutations affecting the gene represented in this entry. Ischemic stroke (ISCHSTR) [MIM:601367]: A stroke is an acute neurologic event leading to death of neural tissue of the brain and resulting in loss of motor, sensory and/or cognitive function. Ischemic strokes, resulting from vascular occlusion, is considered to be a highly complex disease consisting of a group of heterogeneous disorders with multiple genetic and environmental risk factors. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Folate-sensitive neural tube defects (FS-NTD) [MIM:601634]: The most common NTDs are open spina bifida (myelomeningocele) and anencephaly. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	MTHFR catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a co-substrate for homocysteine remethylation to methionine. Genetic variation in this gene influences susceptibility to neural tube defects, occlusive vascular disease, colon cancer and acute leukemia. Mutations in this gene are associated with methylenetetrahydrofolate reductase deficiency.

Additional Information

Gene ID 4524

Other Names	MTHR_HUMAN; MTHFR; 1.5.1.53; MTHR_MOUSE; methylenetetrahydrofolate reductase; 5, 10-methylenetetrahydrofolate reductase (NADPH); methylenetetrahydrofolate reductase (NAD(P)H)
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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.

Protein Information

Name	MTHFR (HGNC:7436)
Function	Catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a cosubstrate for homocysteine remethylation to methionine (PubMed: 29891918). Represents a key regulatory connection between the folate and methionine cycles (Probable).

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