

Product name:	PAH Rabbit Polyclonal Antibody
Cat number:	ABN15696
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human PAH. AA range:351-400
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:20000-1:40000
Molecular Weight:	51kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

PAH encodes the enzyme phenylalanine hydroxylase that is the rate-limiting step in phenylalanine catabolism. Deficiency of this enzyme activity results in the autosomal recessive disorder phenylketonuria. [provided by RefSeq, Jul 2008],catalytic activity:L-phenylalanine + tetrahydrobiopterin + O(2) = L-tyrosine + 4a-hydroxytetrahydrobiopterin.,cofactor:Fe(2+) ion.,disease:Defects in PAH are the cause of hyperphenylalaninemia (HPA) [MIM:261600]. HPA is the mildest form of phenylalanine hydroxylase deficiency.,disease:Defects in PAH are the cause of non-phenylketonuria hyperphenylalaninemia (Non-PKU HPA) [MIM:261600]. Non-PKU HPA is a mild form of phenylalanine hydroxylase deficiency characterized by phenylalanine levels persistently below 600 mumol, which allows normal intellectual and behavioral development without treatment. Non-PKU HPA is usually caused by the combined effect of a mild hyperphenylalaninemia mutation and a severe one.,disease:Defects in PAH are the cause of phenylketonuria (PKU) [MIM:261600]. PKU is an autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor.,enzyme regulation:N-terminal region of PAH is thought to contain allosteric binding sites for phenylalanine and to constitute an "inhibitory" domain that regulates the activity of a catalytic domain in the C-terminal portion of the molecule.,online information:Phenylalanine hydroxylase entry,online information:Phenylalanine hydroxylase locus knowledgebase,pathway:Amino-acid degradation; L-phenylalanine degradation; acetoacetic acid and fumarate from L-phenylalanine: step 1/6.,polymorphism:The Glu-274 variant occurs on approximately 4% of African-American PAH alleles. The enzyme activity of the variant protein is indistinguishable from that of the wild-type form.,similarity:Belongs to the biopterin-dependent aromatic amino acid hydroxylase family.,similarity:Contains 1 ACT domain.,subunit:Homodimer.,