SANTA CRUZ BIOTECHNOLOGY, INC.

PAH (E-8): sc-271257

BACKGROUND

The PAH gene encodes the enzyme phenylalanine hydroxylase (PAH), which converts phenylalanine to tyrosine and is the rate-limiting enzyme in phenylalanine catabolism. Mammalian PAH is a soluble, homotetrameric protein which is abundantly expressed in human liver. Deficiency of PAH activity results in the autosomal recessive disorder phenylketonuria (PKU), which is characterized by mental retardation unless a low phenylalanine diet is introduced early in life. The PAH gene, which maps to human chromosome 12q23.2, contains all the genetic information necessary to code for functional PAH, demonstrating that a single gene is involved in the classic disease phenotype. Numerous mutations can impair the PAH gene, which result in decreased enzyme activity and give rise to varying degrees of PKU. Multiple isozymes of PAH have been reported to exist, but these are most likely allelic variants of PAH that produce protein subunits with slightly different charge and electrophoretic migration.

REFERENCES

1. Hopkinson, D.A., et al. 1976. The distributions of subunit numbers and subunit sizes of enzymes: a study of the products of 100 human gene loci. Ann. Hum. Genet. 39: 383-411.
2. Ledley, F.D., et al. 1985. Gene transfer and expression of human phenylalanine hydroxylase. Science 228: 77-79.
3. Chestkov, V.V. and Laptev, A.V. 1988. Immunochemical detection and characteristics of the subunit composition of phenylalanine hydroxylase in the brain of man. Biull. Eksp. Biol. Med. 106: 30-34.
4. Wang, T., et al. 1991. Founder effect of a prevalent phenylketonuria mutation in the Oriental population. Proc. Natl. Acad. Sci. USA 88: 2146-2150.
5. Kappock, T.J., et al. 1995. Spectroscopic and kinetic properties of unphosphorylated rat hepatic phenylalanine hydroxylase expressed in *Escherichia coli*. Comparison of resting and activated states. J. Biol. Chem. 270: 30532-30544.

CHROMOSOMAL LOCATION

Genetic locus: PAH (human) mapping to 12q23.2; Pah (mouse) mapping to 10 C1.

SOURCE

PAH (E-8) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 404-432 near the C-terminus of PAH of human origin.

PRODUCT

Each vial contains 200 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-271257 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

APPLICATIONS

PAH (E-8) is recommended for detection of PAH of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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).

PAH (E-8) is also recommended for detection of PAH in additional species, including equine, canine and porcine.

Suitable for use as control antibody for PAH siRNA (h): sc-41528, PAH siRNA (m): sc-41529, PAH shRNA Plasmid (h): sc-41528-SH, PAH shRNA Plasmid (m): sc-41529-SH, PAH shRNA (h) Lentiviral Particles: sc-41528-V and PAH shRNA (m) Lentiviral Particles: sc-41529-V.

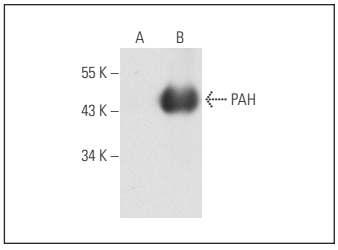
Molecular Weight of PAH: 51 kDa.

Positive Controls: PAH (m): 293T Lysate: sc-122353.

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



PAH (E-8): sc-271257. Western blot analysis of PAH expression in non-transfected: sc-117752 (**A**) and mouse PAH transfected: sc-122353 (**B**) 293T whole cell lysates.

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.