SANTA CRUZ BIOTECHNOLOGY, INC.

PAH (m): 293T Lysate: sc-122353

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 phenyl-ketonuria 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.
6. Fusetti, F., et al. 1998. Structure of tetrameric human phenylalanine hydroxylase and its implications for phenylketonuria. J. Biol. Chem. 273: 16962-16967.
7. Mallolas, J., et al. 2000. Clinical, biomedical, neurological and molecular study of 11 patients with new mutations in PAH gene. Rev. Neurol. 31: 907-910.

CHROMOSOMAL LOCATION

Genetic locus: Pah (mouse) mapping to 10 C1.

PRODUCT

PAH (m): 293T Lysate represents a lysate of mouse PAH transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

PAH (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive PAH antibodies Recommended use: 10-20 µl per lane.

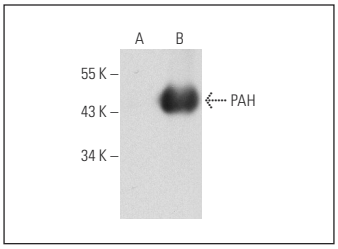
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

PAH (E-8): sc-271257 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse PAH expression in PAH transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

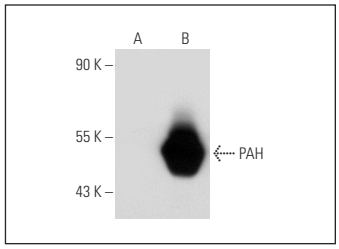
RECOMMENDED SUPPORT REAGENTS

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.

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.



PAH (PH8): sc-58398. Western blot analysis of PAH expression in non-transfected: sc-117752 (**A**) and mouse PAH transfected: sc-122353 (**B**) 293T whole cell lysates.

RESEARCH USE

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

PROTOCOLS

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