

PBGD (B-6): sc-166788

BACKGROUND

PBGD (porphobilinogen deaminase), also designated Hydroxymethylbilane synthase, is a cytoplasmic enzyme found in the heme synthesis pathway. PBGD belongs to the HMBS (hydroxymethylbilane synthase) family. Deficiency of PBGD causes errors in pyrrole metabolism which in turn leads to an inherited autosomal disorder called acute intermittent porphyria (AIP) which is characterized by acute attacks of neurological dysfunctions with hypertension, tachycardia, peripheral neurologic disturbances, abdominal pain and excessive amounts of aminolevulinic acid and porphobilinogen in the urine.

REFERENCES

1. Grandchamp, B., et al. 1987. Tissue-specific expression of porphobilinogen deaminase. Two isoenzymes from a single gene. *Eur. J. Biochem.* 162: 105-110.
2. Mustajoki, S., et al. 2000. Acute intermittent porphyria: expression of mutant and wildtype porphobilinogen deaminase in COS-1 cells. *Mol. Med.* 6: 670-679.
3. Schneider-Yin, X., et al. 2004. Mutation hotspots in the human porphobilinogen deaminase gene: recurrent mutations G111R and R173Q occurring at CpG motifs. *J. Inherit. Metab. Dis.* 27: 625-631.
4. Neuvians, T.P., et al. 2005. Standardization strategy for quantitative PCR in human seminoma and normal testis. *J. Biotechnol.* 117: 163-171.
5. von und zu Fraunberg, M., et al. 2005. Clinical and biochemical characteristics and genotype-phenotype correlation in 143 Finnish and Russian patients with acute intermittent porphyria. *Medicine* 84: 35-47.
6. Sheppard, L. and Dorman, T. 2005. Anesthesia in a child with homozygous porphobilinogen deaminase deficiency: a severe form of acute intermittent porphyria. *Paediatr. Anaesth.* 15: 426-428.
7. SWISS-PROT/TrEMBL (P08397). World Wide Web URL: <http://www.expasy.ch/sprot/sprot-top.html>

CHROMOSOMAL LOCATION

Genetic locus: HMBS (human) mapping to 11q23.3.

SOURCE

PBGD (B-6) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 315-340 near the C-terminus of PBGD of human origin.

PRODUCT

Each vial contains 200 µg IgG₁ 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-166788 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

STORAGE

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

APPLICATIONS

PBGD (B-6) is recommended for detection of PBGD of 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), immunohistochemistry (including paraffin-embedded sections) (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 PBGD siRNA (h): sc-45702, PBGD shRNA Plasmid (h): sc-45702-SH and PBGD shRNA (h) Lentiviral Particles: sc-45702-V.

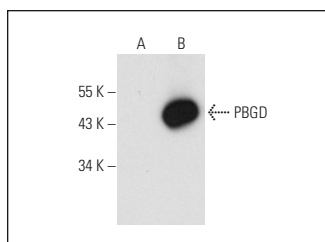
Molecular Weight of PBGD: 42-44 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227, U-937 cell lysate: sc-2239 or PBGD (h): 293 Lysate: sc-110913.

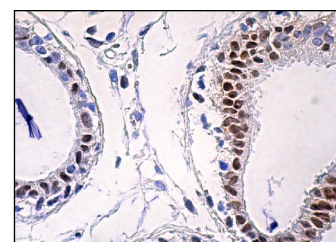
RECOMMENDED SUPPORT REAGENTS

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. 4) Immunohistochemistry: use m-IgGκ BP-HRP: sc-516102 with DAB, 50X: sc-24982 and Immunohistomount: sc-45086, or Organo/Limonene Mount: sc-45087.

DATA



PBGD (B-6): sc-166788. Western blot analysis of PBGD expression in non-transfected: sc-110760 (A) and human PBGD transfected: sc-110913 (B) 293 whole cell lysates.



PBGD (B-6): sc-166788. Immunoperoxidase staining of formalin fixed, paraffin-embedded human breast tissue showing nuclear staining of glandular cells.

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.