



PHKB (h): 293T Lysate: sc-115001

BACKGROUND

Phosphorylase kinase is a hexadecameric enzyme that is comprised of four copies of four subunits that are encoded by four separate genes: PHKA, PHKB, PHKG, and PHKD. This serine/threonine specific kinase converts glycogen phosphorylase b to glycogen phosphorylase a, resulting in the release of glucose-1-phosphate from glycogen. PHKB (phosphorylase β kinase regulatory subunit β) is a 1,093 amino acid subunit of phosphorylase kinase that, along with PHKA, has regulatory functions controlled by phosphorylation. Defects in the gene encoding PHKB are the cause of glycogen storage disease type 9B, which is also known as phosphorylase kinase deficiency of liver and muscle. This disease is characterized by a mild phenotype of hepatomegaly with only slightly elevated transaminase and plasma lipids, no clinical muscle involvement, and generally is correlated with a gradual improvement with increasing age. There are four isoforms of PHKB that are produced as a result of alternative splicing events.

REFERENCES

1. Wullrich-Schmoll, A. and Kilimann, M.W. 1996. Structure of the human gene encoding the phosphorylase kinase beta subunit (PHKB). *Eur. J. Biochem.* 238: 374-380.
2. van den Berg, I.E., van Beurden, E.A., de Klerk, J.B., van Diggelen, O.P., Malingre, H.E., Boer, M.M. and Berger, R. 1997. Autosomal recessive phosphorylase kinase deficiency in liver, caused by mutations in the gene encoding the β subunit (PHKB). *Am. J. Hum. Genet.* 61: 539-546.
3. Burwinkel, B., Moses, S.W. and Kilimann, M.W. 1997. Phosphorylase-kinase-deficient liver glycogenosis with an unusual biochemical phenotype in blood cells associated with a missense mutation in the β subunit gene (PHKB). *Hum. Genet.* 101: 170-174.
4. Burwinkel, B., Hu, B., Schroers, A., Clemens, P.R., Moses, S.W., Shin, Y.S., Pongratz, D., Vorgerd, M. and Kilimann, M.W. 2003. Muscle glycogenosis with low phosphorylase kinase activity: mutations in PHKA1, PHKG1 or six other candidate genes explain only a minority of cases. *Eur. J. Hum. Genet.* 11: 516-526.
5. Burwinkel, B., Rootwelt, T., Kvittingen, E.A., Chakraborty, P.K. and Kilimann, M.W. 2003. Severe phenotype of phosphorylase kinase-deficient liver glycogenosis with mutations in the PHKG2 gene. *Pediatr. Res.* 54: 834-839.
6. Beauchamp, N.J., Dalton, A., Ramaswami, U., Niinikoski, H., Mention, K., Kenny, P., Kolho, K.L., Raiman, J., Walter, J., Treacy, E., Tanner, S. and Sharrard, M. 2007. Glycogen storage disease type IX: High variability in clinical phenotype. *Mol. Genet. Metab.* 92: 88-99.
7. Daub, H., Olsen, J.V., Bairlein, M., Gnäd, F., Oppermann, F.S., Korner, R., Greff, Z., Keri, G., Stemmann, O. and Mann, M. 2008. Kinase-selective enrichment enables quantitative phosphoproteomics of the kinome across the cell cycle. *Mol. Cell* 31: 438-448.
8. Online Mendelian Inheritance in Man, OMIM™. 2009. Johns Hopkins University, Baltimore, MD. MIM Number: 172490. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

CHROMOSOMAL LOCATION

Genetic locus: PHKB (human) mapping to 16q12.1.

PRODUCT

PHKB (h): 293T Lysate represents a lysate of human PHKB transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

PHKB (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive PHKB antibodies. Recommended use: 10-20 μ l per lane.

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

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.

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.