

# GALK1 (C-1): sc-393356

## BACKGROUND

Galactose kinase (GALK1), also often designated galactokinase, is important in the first step of the galactose metabolism pathway. GALK1, which belongs to the GHMP kinase family of proteins, is a crucial enzyme for galactose metabolism. Defects in the gene encoding for galactose kinase, GALK1, can cause galactosemia II, an autosomal recessive disorder characterized by congenital cataracts during infancy, often within the first two weeks of life. In the adult population it can cause presenile cataracts that are secondary to accumulation of galactitol in the lens of the eye.

## REFERENCES

1. Hunter, M., et al. 2001. Novel mutations in the GALK1 gene in patients with galactokinase deficiency. *Hum. Mutat.* 17: 77-78.
2. Okano, Y., et al. 2001. A genetic factor for age-related cataract: identification and characterization of a novel galactokinase variant, "Osaka", in Asians. *Am. J. Hum. Genet.* 68: 1036-1042.
3. Timson, D.J., et al. 2003. Functional analysis of disease-causing mutations in human galactokinase. *Eur. J. Biochem.* 270: 1767-1774.
4. Holden, H.M., et al. 2004. Galactokinase: structure, function and role in type II galactosemia. *Cell. Mol. Life Sci.* 61: 2471-2484.
5. Ross, K.L., et al. 2004. Differential roles of the Leloir pathway enzymes and metabolites in defining galactose sensitivity in yeast. *Mol. Genet. Metab.* 83: 103-116.
6. Sangiuolo, F., et al. 2004. Biochemical characterization of two GALK1 mutations in patients with galactokinase deficiency. *Hum. Mutat.* 23: 396.
7. Thoden, J.B., et al. 2005. Molecular structure of human galactokinase: implications for type II galactosemia. *J. Biol. Chem.* 280:9662-9670.

## CHROMOSOMAL LOCATION

Genetic locus: GALK1 (human) mapping to 17q25.1; Galk1 (mouse) mapping to 11 E2.

## SOURCE

GALK1 (C-1) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 18-37 at the N-terminus of GALK1 of human origin.

## PRODUCT

Each vial contains 200 µg IgG<sub>2b</sub> 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-393356 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

GALK1 (C-1) is recommended for detection of GALK1 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).

Suitable for use as control antibody for GALK1 siRNA (h): sc-60671, GALK1 siRNA (m): sc-60672, GALK1 shRNA Plasmid (h): sc-60671-SH, GALK1 shRNA Plasmid (m): sc-60672-SH, GALK1 shRNA (h) Lentiviral Particles: sc-60671-V and GALK1 shRNA (m) Lentiviral Particles: sc-60672-V.

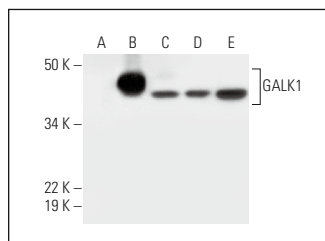
Molecular Weight of GALK1: 42 kDa.

Positive Controls: GALK1 (m): 293T Lysate: sc-125370, Hep G2 cell lysate: sc-2227 or Y79 cell lysate: sc-2240.

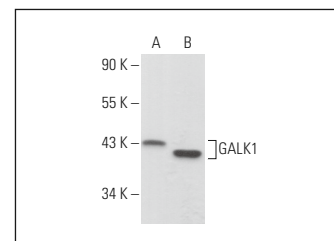
## RECOMMENDED SUPPORT REAGENTS

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



GALK1 (C-1): sc-393356. Western blot analysis of GALK1 expression in non-transfected 293T: sc-117752 (A), mouse GALK1 transfected 293T: sc-125370 (B), Hep G2 (C) and Y79 (D) whole cell lysates and human liver tissue extract (E).



GALK1 (C-1): sc-393356. Western blot analysis of GALK1 expression in Hep G2 whole cell lysate (A) and rat eye tissue extract (B).

## RESEARCH USE

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

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.