

GALC (h2): 293T Lysate: sc-170590

BACKGROUND

GALC (galactosylceramidase) is a lysosomal enzyme that hydrolyzes galactose ester bonds in various galactolipids, including galactosylceramide, galactosylsphingosine, lactosylceramide and monogalactosyldiglyceride. Galactolipids contain glucose and/or galactose and are found in the brain and other nerve tissue, especially the myelin sheath. Galactosylceramide is a major lipid in myelin, kidney and epithelial cells of the small intestine and colon. Mutations in the GALC gene that compromise protein function correlate to Krabbe disease (globoid cell leukodystrophy, GLD). GLD is an autosomal recessive condition that affects approximately 1 in 150,000 infants and results in progressive destruction of the nervous system. The "twitcher" mouse is a model system for GLD; the genotype is a premature stop codon (W339X) in the galactosylceramidase (GALC) gene that abolishes enzymatic activity. Alternate transcriptional splice variants, encoding different isoforms, have been characterized.

REFERENCES

1. Kondo, Y., et al. 2005. Galactocerebroside-deficient oligodendrocytes maintain stable central myelin by exogenous replacement of the missing enzyme in mice. *Proc. Natl. Acad. Sci. USA* 102: 18670-18675.
2. Rafi, M.A., et al. 2005. AAV-mediated expression of galactocerebroside symptoms and extended life span in murine models of globoid cell leukodystrophy. *Mol. Ther.* 11: 734-744.
3. Lin, D., et al. 2005. AAV2/5 vector expressing galactocerebroside ameliorates CNS disease in the murine model of globoid-cell leukodystrophy more efficiently than AAV2. *Mol. Ther.* 12: 422-430.
4. Meng, X.L., et al. 2005. GALC transduction leads to morphological improvement of the twitcher oligodendrocytes *in vivo*. *Mol. Genet. Metab.* 84: 332-343.
5. Luzi, P., et al. 2005. Biochemical and pathological evaluation of long-lived mice with globoid cell leukodystrophy after bone marrow transplantation. *Mol. Genet. Metab.* 86: 150-159.
6. Escolar, M.L., et al. 2005. Transplantation of umbilical-cord blood in babies with infantile Krabbe's disease. *N. Engl. J. Med.* 352: 2069-2081.
7. Luddi, A., et al. 2005. Galactosylceramidase deficiency causes sperm abnormalities in the mouse model of globoid cell leukodystrophy. *Exp. Cell Res.* 304: 59-68.
8. Beier, U.H. and Görögh, T. 2005. Implications of galactocerebroside and galactosylcerebroside metabolism in cancer cells. *Int. J. Cancer* 115: 6-10.
9. Pellegatta, S., et al. 2006. The therapeutic potential of neural stem/progenitor cells in murine globoid cell leukodystrophy is conditioned by macrophage/microglia activation. *Neurobiol. Dis.* 21: 314-323.

CHROMOSOMAL LOCATION

Genetic locus: GALC (human) mapping to 14q31.3.

PRODUCT

GALC (h2): 293T Lysate represents a lysate of human GALC transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

GALC (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive GALC 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.

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.