SANTA CRUZ BIOTECHNOLOGY, INC.

GLCNE (m2): 293T Lysate: sc-125387

BACKGROUND

The bifunctional enzyme UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase (GNE/Mnk), or GLCNE, regulates and initiates biosynthesis of N-acetylneuraminic acid (NeuAc), a precursor of sialic acids. GLCNE is required for normal sialylation in hematopoietic cells. Sialylation is implicated in cell adhesion, signal transduction, tumorigenicity and metastatic behavior of malignant cells. It is upregulated after PKC-dependent phosphorylation and is most abundantly expressed in liver and placenta. It is also expressed, to a lesser extent, in heart, brain, lung, kidney, skeletal muscle and pancreas. Defects in GLCNE are the cause of sialuria, inclusion body myopathy 2 (IBM2) and Nonaka myopathy (NM) or distal myopathy with rimmed vacuoles (DMRV). Sialuria is an autosomal dominant disorder caused by a lack of feedback inhibition of GLCNE by CMP-NeuAc, resulting in overproduction of NeuAc. It is characterized by an accumulation of free sialic acid in the cytoplasm and large quantities of neuraminic acid in the urine. Both IBM2 and NM/DMRV are autosomal recessive neuromuscular disorders characterized by adult onset, distal and proximal muscle weakness (especially in the legs) and a typical muscle pathology including filamentous inclusions and rimmed vacuoles.

REFERENCES

1. Amouri, R., et al. 2005. Allelic heterogeneity of GNE gene mutation in two Tunisian families with autosomal recessive inclusion body myopathy. Neuromuscul. Disord. 15: 361-363.
2. Bork, K., et al. 2005. The intracellular concentration of sialic acid regulates the polysialylation of the neural cell adhesion molecule. FEBS Lett. 579: 5079-5083.
3. Krause, S., et al. 2005. Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. Exp. Cell Res. 304: 365-379.
4. Nonaka, I., et al. 2005. Distal myopathy with rimmed vacuoles and hereditary inclusion body myopathy. Curr. Neurol. Neurosci. Rep. 5: 61-65.
5. Ro, L.S., et al. 2005. Phenotypic variability in a Chinese family with rimmed vacuolar distal myopathy. J. Neurol. Neurosurg. Psychiatry 76: 752-755.
6. Salama, I., et al. 2005. No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. Biochem. Biophys. Res. Commun. 328: 221-226.
7. Sparks, S.E., et al. 2005. Use of a cell-free system to determine UDP-N-acetylglucosamine 2-epimerase and N-acetylmannosamine kinase activities in human hereditary inclusion body myopathy. Glycobiology 15: 1102-1110.
8. Suzuki, N., et al. 2005. Late-onset distal myopathy with rimmed vacuoles without mutation in the GNE or dysferlin genes. Muscle Nerve 32: 812-814.

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.

PROTOCOLS

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

CHROMOSOMAL LOCATION

Genetic locus: Gne (mouse) mapping to 4 B1.

PRODUCT

GLCNE (m2): 293T Lysate represents a lysate of mouse GLCNE transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

GLCNE (m2): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive GLCNE antibodies. Recommended use: 10-20 µl per lane.

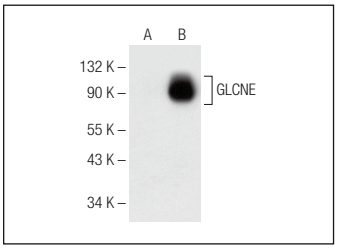
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

GLCNE (H-10): sc-376057 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse GLCNE expression in GLCNE 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



GLCNE (H-10): sc-376057. Western blot analysis of GLCNE expression in non-transfected: sc-117752 (**A**) and mouse GLCNE transfected: sc-125387 (**B**) 293T whole cell lysates.

RESEARCH USE

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