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GNE Antibody Blocking Peptide

Catalog Number: bs-9882P
Activity: Not tested
Purification: HPLC
Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.
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.