SunLong Biotech Co.,LTD
Tel: 0086-571- 56623320 Fax:0086-571- 56623318
E-mail:sales@sunlongbiotech.com
www.sunlongbiotech.com

Rabbit Anti-GLCNE antibody

SL9882R

Product Name:	GLCNE
Chinese Name:	GLCNE蛋白抗体
Alias:	IBM2; Uae1; Bifunctional UDP N acetylglucosamine 2 epimerase/N acetylmannosamine kinase; DMRV; ManAc kinase; N acylmannosamine kinase; NM; RP23-209M8.6; UDP GlcNAc 2 epimerase; UDP GlcNAc 2 epimerase/ManAc kinase; Uridine diphosphate N acetylglucosamine 2 epimerase; GLCNE_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Pig,Cow,Horse,Rabbit,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:50-200（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	79kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human GLCNE:131-230/722
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	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.

Function:
Regulates and initiates biosynthesis of N-acetylneuraminic acid (NeuAc), a precursor of sialic acids. Plays an essential role in early development (By similarity). Required for normal sialylation in hematopoietic cells. Sialylation is implicated in cell adhesion, signal transduction, tumorigenicity and metastatic behavior of malignant cells.

Subunit:
Homodimer and homohexamer.

Subcellular Location:
Cytoplasmic

Tissue Specificity:
Highest expression in liver and placenta. Also found in heart, brain, lung, kidney, skeletal muscle and pancreas. Isoform 1 is expressed in heart, brain, kidney, liver, placenta, lung, spleen, pancreas, skeletal muscle and colon. Isoform 2 is expressed mainly in placenta, but also in brain, kidney, liver, lung, pancreas and colon. Isoform 3 is expressed at low level in kidney, liver, placenta and colon.

Post-translational modifications:
Phosphorylated by PKC (By similarity).

DISEASE:
Defects in GNE are a cause of sialuria (SIALURIA) [MIM:269921]; also known as sialuria French type. In sialuria, free sialic acid accumulates in the cytoplasm and gram quantities of neuraminic acid are secreted in the urine. The metabolic defect involves lack of feedback inhibition of UDP-GlcNAc 2-epimerase by CMP-Neu5Ac, resulting in constitutive overproduction of free Neu5Ac. Clinical features include variable degrees of developmental delay, coarse facial features and hepatomegaly. Sialuria inheritance is autosomal dominant.
Defects in GNE are the cause of inclusion body myopathy type 2 (IBM2) [MIM:600737]. Hereditary inclusion body myopathies are a group of neuromuscular disorders characterized by adult onset, slowly progressive distal and proximal weakness

and a typical muscle pathology including rimmed vacuoles and filamentous inclusions. IBM2 is an autosomal recessive disorder affecting mainly leg muscles, but with an unusual distribution that spares the quadriceps as also observed in Nonaka myopathy. Defects in GNE are the cause of Nonaka myopathy (NM) [MIM:605820]; also known as distal myopathy with rimmed vacuoles (DMRV). NM is an autosomal recessive muscular disorder, allelic to inclusion body myopathy 2. It is characterized by weakness of the anterior compartment of the lower limbs with onset in early adulthood, and sparing of the quadriceps muscles. As the inclusion body myopathy, NM is histologically characterized by the presence of numerous rimmed vacuoles without inflammatory changes in muscle specimens.

Similarity:
In the N-terminal section; belongs to the UDP-N-acetylglucosamine 2-epimerase family.
In the C-terminal section; belongs to the ROK (NagC/XylR) family.

SWISS:
Q9Y223

Gene ID:
10020

Database links:

Entrez Gene: 10020Human

Entrez Gene: 50798Mouse

Entrez Gene: 114711Rat

Omim: 603824Human

SwissProt: Q9Y223Human

SwissProt: Q91WG8Mouse

SwissProt: O35826Rat

Unigene: 5920Human

Unigene: 256718Mouse

Unigene: 18753Rat

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.