

GCS1 Rabbit pAb

Catalog # AP97369

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q13724
Other Accession	Q13724
Predicted	Rat, Human, Mouse, Dog
Host	Rabbit
Clonality	Polyclonal
Calculated MW	91918
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human GCS1
Epitope Specificity	51-150/837
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the glycosyl hydrolase 63 family.
DISEASE	Defects in MOGS are the cause of type IIb congenital disorder of glycosylation (CDGIIb) [MIM:606056]; also known as glucosidase I deficiency. CDGIIb is characterized by marked generalized hypotonia and hypomotility of the neonate, dysmorphic features, including a prominent occiput, short palpebral fissures, retrognathia, high arched palate, generalized edema, and hypoplastic genitalia. Symptoms of the infant included hepatomegaly, hypoventilation, feeding problems and seizures. The clinical course was progressive and the infant did not survive more than a few months.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Glycosylation of asparagine residues in Asn-X-Ser/Thr motifs in proteins commonly occur in the lumen of the endoplasmic reticulum (ER). Glucosidase I catalyzes the first step in the N-linked oligosaccharide processing pathway. It specifically removes the distal alpha 1,2-linked glucose residue from the Glc3-Man9-GlcNAc2 oligosaccharide precursor. Glucosidase I contains a short cytosolic tail, a single pass transmembrane domain and a large C-terminal catalytic domain located on the luminal side of the ER. Mutations in the gene encoding Glucosidase I result in the congenital disorder glycosylation (CDG-IIb), which is characterized by generalized hypotonia, dysmorphic features, hepatomegaly, hypoventilation, feeding problems, seizures and death. Two point mutations in the Glucosidase I gene have been identified and result in amino acid substitutions, namely Arg486Thr and Phe652Leu, that affect polypeptide folding and active site formation.

Additional Information

Gene ID	7841
Other Names	CDG2B; CWH41; DER7; GCS1; MOGS_HUMAN; MOGS; Processing

A-glucosidase I; 3.2.1.106; mannosyl-oligosaccharide glucosidase; glucosidase I

Dilution WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000

Storage Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name MOGS ([HGNC:24862](#))

Function In the context of N-glycan degradation, cleaves the distal alpha 1,2-linked glucose residue from the Glc(3)Man(9)GlcNAc(2) oligosaccharide precursor in a highly specific manner.

Cellular Location Endoplasmic reticulum membrane; Single-pass type II membrane protein {ECO:0000250|UniProtKB:O88941}

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.