10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

HGSNAT Rabbit pAb

HGSNAT Rabbit pAb
Catalog # AP56292

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q68CP4
Predicted	Dog, Human, Mouse, Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	73293
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human HGSNAT
Epitope Specificity	301-400/663
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Lysosome membrane. Colocalizes with the lysosomal marker LAMP2. The signal peptide is not cleaved upon translocation into the endoplasmic reticulum; the precursor is probably targeted to the lysosomes via the adapter protein complex-mediated pathway that involves tyrosine- and/or dileucine-based conserved amino acid motifs in the last C-terminus 16-amino acid domain.
DISEASE	Defects in HGSNAT are the cause of mucopolysaccharidosis type 3C (MPS3C) [MIM:252930]; also known as Sanfilippo C syndrome. MPS3C is a form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a lysosomal acetyltransferase, which is one of several enzymes involved in the lysosomal degradation of heparin sulfate. Mutations in this gene are associated with Sanfilippo syndrome C, one type of the lysosomal storage disease mucopolysaccaridosis III, which results from impaired degradation of heparan sulfate. [provided by RefSeq, Jan 2009]

Additional Information

Gene ID	138050
Other Names	Heparan-alpha-glucosaminide N-acetyltransferase, 2.3.1.78, Transmembrane protein 76, HGSNAT, TMEM76

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	HGSNAT
Synonyms	TMEM76
Function	Lysosomal acetyltransferase that acetylates the non-reducing terminal alpha-glucosamine residue of intralysosomal heparin or heparan sulfate, converting it into a substrate for luminal alpha-N-acetyl glucosaminidase.
Cellular Location	Lysosome membrane; Multi-pass membrane protein Note=Colocalizes with the lysosomal marker LAMP2. The signal peptide is not cleaved upon translocation into the endoplasmic reticulum; the precursor is probably targeted to the lysosomes via the adapter protein complex-mediated pathway that involves tyrosine- and/or dileucine-based conserved amino acid motifs in the last C-terminus 16-amino acid domain
Tissue Location	Widely expressed, with highest level in leukocytes, heart, liver, skeletal muscle, lung, placenta and liver

Background

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