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GNPTAB Rabbit pAb

Catalog # AP96572

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q3T906
Other Accession	Q3T906
Predicted	Rat, Pig, Human, Mouse, Dog, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	143622
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human N-acetylglucosamine-1-phosphotransferase subunit beta
Epitope Specificity	901-1000/1256
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 stealth family. Contains 1 EF-hand domain. Contains 2 LNR (Lin/Notch) repeats.
SUBUNIT	Hexamer of two alpha, two beta and two gamma subunits; disulfide-linked. It is believed that the alpha and/or the beta subunit of the enzyme contain the catalytic portion and that the gamma subunit functions in recognition of the lysosomal enzymes.
Post-translational modifications	The alpha- and beta-subunits appear to be generated by a proteolytic cleavage at the Lys-928-Asp-929 bond.
DISEASE	Defects in GNPTAB are the cause of mucolipidosis type II (MLII) [MIM:252500]; also known as inclusion cell disease or I-cell disease (ICD). MLII is a fatal, autosomal recessive, lysosomal storage disorder characterized by severe clinical and radiologic features, peculiar fibroblast inclusions, and no excessive mucopolysaccharidurie. Congenital dislocation of the hip, thoracic deformities, hernia, and hyperplastic gums are evident soon after birth. Defects in GNPTAB are the cause of mucolipidosis type III complementation group A (MLIIIA) [MIM:252600]; also known as variant pseudo-Hurler polydystrophy. MLIIIA is an autosomal recessive disease of lysosomal enzyme targeting. Clinically MLIII is characterized by restricted joint mobility, skeletal dysplasia, and short stature. Mildly coarsened facial features and thickening of the skin have been described. Cardiac valvular disease and corneal clouding may also occur. Half of the reported patients show learning disabilities or mental retardation.
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 two of three subunit types of the membrane-bound enzyme N-acetylglucosamine-1-phosphotransferase, a heterohexameric complex composed of two alpha, two beta, and two gamma subunits. The encoded protein is proteolytically cleaved at the Lys928-Asp929 bond to yield mature alpha and beta polypeptides while the gamma subunits are the product of a distinct gene (GeneID 84572). In the Golgi apparatus, the heterohexameric complex catalyzes the first step in the synthesis of mannose

6-phosphate recognition markers on certain oligosaccharides of newly synthesized lysosomal enzymes. These recognition markers are essential for appropriate trafficking of lysosomal enzymes. Mutations in this gene have been associated with both mucolipidosis II and mucolipidosis IIIA.[provided by RefSeq, May 2010].

Additional Information

Gene ID	79158
Other Names	GNPTA; ICD; EG432486; RGD1564821; GNPTA_HUMAN; GNPTAB; GlcNAc-1-phosphotransferase subunits alpha/beta; Stealth protein GNPTAB; UDP-N-acetylglucosamine-1-phosphotransferase subunits alpha/beta; 2.7.8.17; KIAA1208; GNPTA_MOUSE; N-acetylglucosamine-1-phosphate transferase subunits alpha and beta; N-acetylglucosamine-1-phosphate transferase, alpha and beta subunits
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	GNPTAB
Synonyms	GNPTA, KIAA1208
Function	Catalyzes the formation of mannose 6-phosphate (M6P) markers on high mannose type oligosaccharides in the Golgi apparatus. M6P residues are required to bind to the M6P receptors (MPR), which mediate the vesicular transport of lysosomal enzymes to the endosomal/prelysosomal compartment.
Cellular Location	[N-acetylglucosamine-1-phosphotransferase subunit alpha]: Golgi apparatus membrane; Single-pass type I membrane protein
Tissue Location	Expressed in the heart, whole brain, placenta, lung, liver, skeletal muscle, kidney and pancreas

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