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Rabbit Anti-GNPTAB antibody

SL10460R

Product Name	GNPTAB
Chinese Name	溶酶体累积病相关蛋白/口吃相关蛋白抗体
Alias	N-acetylglucosamine-1-phosphotransferase subunit alpha; EC=2.7.8.17; GlcNAc-1-phosphotransferase subunits alpha/beta; GNPTA; GNPTA_HUMAN; Gnptab; KIAA1208; Stealth protein GNPTAB; UDP-N-acetylglucosamine-1-phosphotransferase subunits alpha/beta.
Research Area	Tumour Cell biology Signal transduction The new supersedes the old
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	(predicted: Human, Mouse, Rat, Pig, Cow, Horse, Rabbit,)
Applications	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000 （Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Theoretical molecular weight	105kDa
Cellular localization	cytoplasmic The cell membrane
Form	Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human N-acetylglucosamine-1-phosphotransferase subunit alpha: 1-100/1256
Lsotype	IgG
Purification	affinity purified by Protein A
Buffer Solution	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol.
Storage	Shipped at 4℃. Store at -20 °C for one year. Avoid repeated freeze/thaw cycles.
Attention	This product as supplied is intended for research use only, not for use in human, therapeutic or

diagnostic applications.

PubMed PubMed

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].

Product Detail

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.

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.

Subcellular Location:
N-acetylglucosamine-1-phosphotransferase subunit alpha: Golgi apparatus membrane; Single-pass type I membrane protein.
N-acetylglucosamine-1-phosphotransferase subunit beta: Golgi apparatus membrane; Single-pass type II membrane protein.

Tissue Specificity:
Expressed in the heart, whole brain, placenta, lung, liver, skeletal muscle, kidney and pancreas.

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 mucopolysaccharidurial. 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.

Similarity:
Belongs to the stealth family.
Contains 1 EF-hand domain.
Contains 2 LNR (Lin/Notch) repeats.

SWISS:
Q3T906

Gene ID:
79158

Database links:

Entrez Gene: 79158 Human

Omim: 607840 Human

SwissProt: Q3T906 Human

Unigene: 46850 Human