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Rabbit Anti-GNPTG antibody

SL13477R

Product Name:	GNPTG
Chinese Name:	溶酶体累积病相关蛋白/口吃相关蛋白抗体
Alias:	GlcNAc phosphotransferase gamma subunit; GlcNAc-1-phosphotransferase subunit gamma; GNPTAG; LP2537; N-acetylglucosamine-1-phosphate transferase gamma subunit; RJD9; UDP-N-acetylglucosamine-1-phosphotransferase subunit gamma; GNPTG_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	32kDa
Cellular localization:	cytoplasmicSecretory protein
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human GNPTG:41-140/305
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:	This gene encodes the gamma sunbunit of the N-acetylglucosamine-1-phosphotransferase complex. This hexameric complex, composed of alpha, beta and gamma subunits, catalyzes the first step in synthesis of a mannose 6-phosphate lysosomal recognition marker. This enzyme complex is necessary for targeting of

lysosomal hydrolases to the lysosome. Mutations in the gene encoding the gamma subunit have been associated with mucolipidosis IIIC, also known as mucolipidosis III gamma.[provided by RefSeq, Feb 2010].

Function:
May recognize the substrate of GlcNAc-1-phosphotransferase but also the lysosomal proteins with mannose-6-phosphate residues.

Subunit:
Hexamer of two alpha, two beta and two gamma subunit; 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:
Golgi Apparatus and Secreted.

Tissue Specificity:
Widely expressed.

DISEASE:
Defects in GNPTG are the cause of mucolipidosis type III complementation group C (MLIIIC) [MIM:252605]; also known as variant pseudo-Hurler polydystrophy. MLIIIC is an autosomal recessive disease of lysosomal hydrolase trafficking. Unlike the related diseases, mucolipidosis II and IIIA, the enzyme affected in mucolipidosis IIIC (GlcNAc-phosphotransferase) retains full transferase activity on synthetic substrates but lacks activity on lysosomal hydrolases. Typical clinical findings include stiffness of the hands and shoulders, claw-hand deformity, scoliosis, short stature, coarse facies, and mild mental retardation. Radiographically, severe dysostosis multiplex of the hip is characteristic and frequently disabling. The clinical diagnosis can be confirmed by finding elevated serum lysosomal enzyme levels and/or decreased lysosomal enzyme levels in cultured fibroblasts.

Similarity:
Contains 1 PRKCSH domain.

SWISS:
Q9UJJ9

Gene ID:
84572

Database links:

Entrez Gene: 84572Human

Omim: 607838Human

	SwissProt: Q9UJJ9Human Unigene: 241575Human **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.