

GNPTAB (D-13): sc-107561

BACKGROUND

GlcNAc-1-phosphotransferase subunits α/β (GNPTAB), also known as N-acetylglucosamine-1-phosphotransferase subunits α/β or UDP-N-acetylglucosamine-1-phosphotransferase subunits α/β , is a 1,256 amino acid member of the stealth family of proteins. Localized to the Golgi apparatus membrane, GNPTAB is expressed in heart, brain, placenta, lung, liver, kidney, pancreas and skeletal muscle. GNPTAB catalyzes the formation of mannose 6-phosphate (M6P) markers on high mannose type oligosaccharides in the Golgi apparatus. M6Ps bind to the M6P receptors (MPR), after which MPRs can mediate the vesicular transport of lysosomal enzymes to the endosomal/prelysosomal compartment. Defects in the gene encoding GNPTAB lead to mucopolipidosis type II (MLII), also known as inclusion cell disease (ICD), and mucopolipidosis type III complementation group A (MLIIIA), also known as variant pseudo-Hurler polydystrophy. Two isoforms of GNPTAB exist as a result of alternative splicing events.

REFERENCES

- Dittmer, F. and von Figura, K. 1999. Phosphorylation of arylsulphatase A occurs through multiple interactions with the UDP-N-acetylglucosamine-1-phosphotransferase proximal and distal to its retrieval site by the KDEL receptor. *Biochem. J.* 340: 729-736.
- Tiede, S., et al. 2004. A novel mutation in UDP-N-acetylglucosamine-1-phosphotransferase γ subunit (GNPTAG) in two siblings with mucopolipidosis type III alters a used glycosylation site. *Hum. Mutat.* 24: 535.
- Raas-Rothschild, A., et al. 2004. Genomic organisation of the UDP-N-acetylglucosamine-1-phosphotransferase γ subunit (GNPTAG) and its mutations in mucopolipidosis III. *J. Med. Genet.* 41: e52.
- Tiede, S., et al. 2005. Missense mutations in N-acetylglucosamine-1-phosphotransferase α/β subunit gene in a patient with mucopolipidosis III and a mild clinical phenotype. *Am. J. Med. Genet. A.* 137A: 235-240.
- Tiede, S., et al. 2005. Mucopolipidosis II is caused by mutations in GNPTA encoding the α/β GlcNAc-1-phosphotransferase. *Nat. Med.* 11: 1109-1112.
- Sperisen, P., et al. 2005. Stealth proteins: in silico identification of a novel protein family rendering bacterial pathogens invisible to host immune defense. *PLoS Comput. Biol.* 1: e63.
- Kudo, M., et al. 2006. Mucopolipidosis II (I-cell disease) and mucopolipidosis IIIA (classical pseudo-hurler polydystrophy) are caused by mutations in the GlcNAc-phosphotransferase α/β -subunits precursor gene. *Am. J. Hum. Genet.* 78: 451-463.
- Tiede, S., et al. 2006. Missense mutation in the N-acetylglucosamine-1-phosphotransferase gene (GNPTA) in a patient with mucopolipidosis II induces changes in the size and cellular distribution of GNPTG. *Hum. Mutat.* 27: 830-831.

CHROMOSOMAL LOCATION

Genetic locus: GNPTAB (human) mapping to 12q23.2; Gnptab (mouse) mapping to 10 C1.

SOURCE

GNPTAB (D-13) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of GNPTAB of human origin.

PRODUCT

Each vial contains 200 μ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-107561 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

GNPTAB (D-13) is recommended for detection of GNPTAB of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 μ g per 100-500 μ g of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

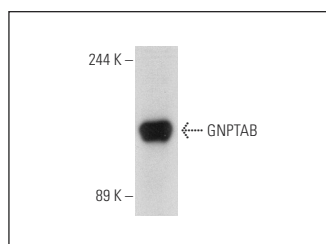
GNPTAB (D-13) is also recommended for detection of GNPTAB in additional species, including bovine.

Suitable for use as control antibody for GNPTAB siRNA (h): sc-95992, GNPTAB siRNA (m): sc-145658, GNPTAB shRNA Plasmid (h): sc-95992-SH, GNPTAB shRNA Plasmid (m): sc-145658-SH, GNPTAB shRNA (h) Lentiviral Particles: sc-95992-V and GNPTAB shRNA (m) Lentiviral Particles: sc-145658-V.

Molecular Weight of GNPTAB: 144 kDa.

Positive Controls: mouse brain extract: sc-2253.

DATA



GNPTAB (D-13): sc-107561. Western blot analysis of GNPTAB expression in mouse brain tissue extract.

STORAGE

Store at 4° C, ****DO NOT FREEZE****. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

See our web site at www.scbt.com or our catalog for detailed protocols and support products.