

GNPTAB siRNA (h): sc-95992

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

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2. 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.
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4. 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.
5. 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.
6. 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.
7. 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.
8. 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.
9. Gelfman, C.M., et al. 2007. Mice lacking α/β subunits of GlcNAc-1-phosphotransferase exhibit growth retardation, retinal degeneration, and secretory cell lesions. *Invest. Ophthalmol. Vis. Sci.* 48: 5221-5228.

CHROMOSOMAL LOCATION

Genetic locus: GNPTAB (human) mapping to 12q23.2.

PRODUCT

GNPTAB siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see GNPTAB shRNA Plasmid (h): sc-95992-SH and GNPTAB shRNA (h) Lentiviral Particles: sc-95992-V as alternate gene silencing products.

For independent verification of GNPTAB (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-95992A, sc-95992B and sc-95992C.

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNAses and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 μ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330 μ l of RNase-free water makes a 10 μ M solution in a 10 μ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

GNPTAB siRNA (h) is recommended for the inhibition of GNPTAB expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 μ M in 66 μ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor GNPTAB gene expression knockdown using RT-PCR Primer: GNPTAB (h)-PR: sc-95992-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

SELECT PRODUCT CITATIONS

1. Tiede, S., et al. 2021. UDP-GlcNAc-1-phosphotransferase is a clinically important regulator of human and mouse hair pigmentation. *J. Invest. Dermatol.* 141: 2957-2965.e5.

RESEARCH USE

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