

COH1 siRNA (h): sc-105225

BACKGROUND

COH1 (Cohen syndrome protein 1), also known as VPS13B (vacuolar protein sorting-associated protein 13B) or CHS1, is a 4,022 amino acid protein that belongs to the VPS13 family. COH1 is widely expressed and may be involved in protein sorting in post Golgi membrane traffic. Defects in the gene that encodes COH1 are the cause of Cohen syndrome, which is characterized by intellectual deficit, abnormalities of the hands and feet, hypotonia and obesity. Craniofacial dysmorphisms commonly seen with Cohen syndrome include a low hairline, thick hair, high-arched or wave-shaped eyelids and a short philtrum. COH1 is expressed as five isoforms produced by alternative splicing. Isoform 1 is expressed in brain and retina while isoform 2 is expressed ubiquitously.

REFERENCES

1. Kolehmainen, J., et al. 2003. Cohen syndrome is caused by mutations in a novel gene, COH1, encoding a transmembrane protein with a presumed role in vesicle-mediated sorting and intracellular protein transport. *Am. J. Hum. Genet.* 72: 1359-1369.
2. Hennies, H.C., et al. 2004. Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome. *Am. J. Hum. Genet.* 75: 138-145.
3. Velayos-Baeza, A., et al. 2004. Analysis of the human VPS13 gene family. *Genomics* 84: 536-549.
4. Farooqi, I.S. 2005. Genetic and hereditary aspects of childhood obesity. *Best Pract. Res. Clin. Endocrinol. Metab.* 19: 359-374.
5. Kondo, I., et al. 2005. COH1 analysis and linkage study in two Japanese families with Cohen syndrome. *Clin. Genet.* 67: 270-272.
6. Zarzour, W., et al. 2005. Two novel CHS1 (LYST) mutations: clinical correlations in an infant with Chediak-Higashi syndrome. *Mol. Genet. Metab.* 85: 125-132.
7. Khan, A., et al. 2006. Corneal ectasia associated with Cohen syndrome: a role for COH1 in corneal development and maintenance? *Br. J. Ophthalmol.* 90: 390-391.

CHROMOSOMAL LOCATION

Genetic locus: VPS13B (human) mapping to 8q22.2.

PRODUCT

COH1 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 COH1 shRNA Plasmid (h): sc-105225-SH and COH1 shRNA (h) Lentiviral Particles: sc-105225-V as alternate gene silencing products.

For independent verification of COH1 (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-105225A, sc-105225B and sc-105225C.

RESEARCH USE

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

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

COH1 siRNA (h) is recommended for the inhibition of COH1 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 COH1 gene expression knockdown using RT-PCR Primer: COH1 (h)-PR: sc-105225-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

PROTOCOLS

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