



L2HGDH siRNA (h): sc-92295

BACKGROUND

L2HGDH (L-2-hydroxyglutarate dehydrogenase, mitochondrial), also known as C14orf160, is a 463 amino acid protein that belongs to the L2HGDH family. Localizing to the mitochondrion, L2HGDH is widely expressed, with high levels found in brain, testis and muscle. Lower levels of L2HGDH expression can be found in lymphocytes, fibroblasts, keratinocytes, placenta, bladder, small intestine, liver and bone marrow. Utilizing FAD as a cofactor, L2HGDH catalyses L-2-hydroxyglutarate to α -ketoglutarate. The gene encoding L2HGDH maps to human chromosome 14q21.3 and mouse chromosome 12 C2; defects to this gene result in L-2-hydroxyglutaric aciduria (L2HGA). L2HGA is a rare autosomal recessive neurometabolic disorder in which L-2-hydroxyglutaric acid accumulates in urine, blood and cerebrospinal fluid. Progressive cerebellar ataxia, dysarthria and moderate to severe mental retardation are characteristic of L2HGA.

REFERENCES

1. Duran, M., et al. 1980. L-2-Hydroxyglutaric aciduria: an inborn error of metabolism? *J. Inher. Metab. Dis.* 3: 109-112.
2. Topçu, M., et al. 2004. L-2-Hydroxyglutaric aciduria: identification of a mutant gene C14orf160, localized on chromosome 14q22.1. *Hum. Mol. Genet.* 13: 2803-2811.
3. Rzem, R., et al. 2004. A gene encoding a putative FAD-dependent L-2-hydroxyglutarate dehydrogenase is mutated in L-2-hydroxyglutaric aciduria. *Proc. Natl. Acad. Sci. USA* 101: 16849-16854.
4. Vilarinho, L., et al. 2005. Novel L2HGDH mutations in 21 patients with L-2-hydroxyglutaric aciduria of Portuguese origin. *Hum. Mutat.* 26: 395-396.
5. Rzem, R., et al. 2006. The gene mutated in L-2-hydroxyglutaric aciduria encodes L-2-hydroxyglutarate dehydrogenase. *Biochimie* 88: 113-116.
6. Steenweg, M.E., et al. 2010. An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. *Hum. Mutat.* 31: 380-390.
7. Short, A.D., et al. 2010. Exonic mutations in the L2HGDH gene in Staffordshire bull terriers. *Vet. Rec.* 167: 455-457.
8. Brehmer, S., et al. 2011. Mutational analysis of D2HGDH and L2HGDH in brain tumours without IDH1 or IDH2 mutations. *Neuropathol. Appl. Neurobiol.* 37: 330-332.
9. Krell, D., et al. 2011. Screen for IDH1, IDH2, IDH3, D2HGDH and L2HGDH mutations in glioblastoma. *PLoS ONE* 6: e19868.

CHROMOSOMAL LOCATION

Genetic locus: L2HGDH (human) mapping to 14q21.3.

RESEARCH USE

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

PROTOCOLS

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

PRODUCT

L2HGDH 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 L2HGDH shRNA Plasmid (h): sc-92295-SH and L2HGDH shRNA (h) Lentiviral Particles: sc-92295-V as alternate gene silencing products.

For independent verification of L2HGDH (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-92295A, sc-92295B and sc-92295C.

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

L2HGDH siRNA (h) is recommended for the inhibition of L2HGDH 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 L2HGDH gene expression knockdown using RT-PCR Primer: L2HGDH (h)-PR: sc-92295-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.