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SANTA CRUZ BIOTECHNOLOGY, INC.

MMACHC siRNA (m): sc-149475

BACKGROUND

MMACHC (methylmalonic aciduria and homocystinuria type C protein), also known as cblC, is a 282 amino acid widely expressed protein that may be involved in the binding and intracellular trafficking of cobalamin (vitamin B12). Defects in the gene encoding MMACHC are the cause of methylmalonic aciduria and homocystinuria type cblC, a disorder of cobalamin metabolism characterized by decreased levels of the coenzymes adenosylcobalamin (AdoCbl) and methylcobalamin (MeCbl). AdoCbl is an essential cofactor utilized by MUT, the mitochondrial methylmalonyl-CoA mutase that plays an important role in the catabolism of cholesterol, branched chain amino acids, odd-numbered fatty acids and other metabolites. MeCbl is an active coenzyme form of vitamin B12 and is essential for cell growth and replication. Individuals affected by methylmalonic aciduria and homocystinuria type cblC experience negative developmental, hematologic, neurologic, metabolic, ophthalmologic and dermatologic manifestations.

REFERENCES

1. Morel, C.F., et al. 2006. Combined methylmalonic aciduria and homocystinuria (cblC): phenotype-genotype correlations and ethnic-specific observations. Mol. Genet. Metab. 88: 315-321.
2. Tsai, A.C., et al. 2007. Late-onset combined homocystinuria and methylmalonic aciduria (cblC) and neuropsychiatric disturbance. Am. J. Med. Genet. A 143A: 2430-2434.
3. Nogueira, C., et al. 2008. Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. Mol. Genet. Metab. 93: 475-480.
4. Kim, J., et al. 2008. Decyanation of vitamin B12 by a trafficking chaperone. Proc. Natl. Acad. Sci. USA 105: 14551-14554.
5. Lerner-Ellis, J.P., et al. 2009. Spectrum of mutations in MMACHC, allelic expression, and evidence for genotype-phenotype correlations. Hum. Mutat. 30: 1072-1081.

CHROMOSOMAL LOCATION

Genetic locus: Mmachc (mouse) mapping to 4 D1.

PRODUCT

MMACHC siRNA (m) 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 µM solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see MMACHC shRNA Plasmid (m): sc-149475-SH and MMACHC shRNA (m) Lentiviral Particles: sc-149475-V as alternate gene silencing products.

For independent verification of MMACHC (m) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-149475A, sc-149475B and sc-149475C.

PROTOCOLS

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

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 µl of the RNAse-free water provided. Resuspension of the siRNA duplex in 330 µl of RNAse-free water makes a 10 µM solution in a 10 µM Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

MMACHC siRNA (m) is recommended for the inhibition of MMACHC expression in mouse 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 µM in 66 µl. 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 MMACHC gene expression knockdown using RT-PCR Primer: MMACHC (m)-PR: sc-149475-PR (20 µl). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

RESEARCH USE

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