

MMAA siRNA (m): sc-149474

BACKGROUND

MMAA (methylmalonic aciduria type A), also known as methylmalonic aciduria (cobalamin deficiency) cblA type, is a 418 amino acid mitochondrial protein that belongs to the ArgK family and is suggested to function as a GTPase. Implicated in the transport of cobalamin into mitochondria during late adenosylcobalamin synthesis, MMAA exists as a widely expressed homodimer that is found at highest levels in skeletal muscle and liver. The gene encoding MMAA maps to human chromosome 4q31.21, and defects in the gene are the cause of an autosomal recessive disease known as methylmalonic aciduria type cblA (MMAA). Chromosome 4 represents approximately 6% of the human genome, contains nearly 900 genes and is associated with Huntington's disease, Ellis-van Creveld syndrome, methylmalonic acidemia and polycystic kidney disease.

REFERENCES

1. Kalchman, M.A., Graham, R.K., Xia, G., Koide, H.B., Hodgson, J.G., Graham, K.C., Goldberg, Y.P., Gietz, R.D., Pickart, C.M. and Hayden, M.R. 1996. Huntingtin is ubiquitinated and interacts with a specific ubiquitin-conjugating enzyme. *J. Biol. Chem.* 271: 19385-19394.
2. Howard, T.D., Guttmacher, A.E., McKinnon, W., Sharma, M., McKusick, V.A. and Jabs, E.W. 1997. Autosomal dominant postaxial polydactyly, nail dys trophy, and dental abnormalities map to chromosome 4p16, in the region containing the Ellis-van Creveld syndrome locus. *Am. J. Hum. Genet.* 61: 1405-1412.
3. Sommardahl, C., Cottrell, M., Wilkinson, J.E., Woychik, R.P. and Johnson, D.K. 2001. Phenotypic variations of orpk mutation and chromosomal local-ization of modifiers influencing kidney phenotype. *Physiol. Genomics* 7: 127-134.
4. Dobson, C.M., Wai, T., Leclerc, D., Wilson, A., Wu, X., Dore, C., Hudson, T., Rosenblatt, D.S. and Gravel, R.A. 2002. Identification of the gene responsi-ble for the cblA complementation group of vitamin B12-responsive methyl-malonic acidemia based on analysis of prokaryotic gene arrangements. *Proc. Natl. Acad. Sci. USA* 99: 15554-15559.
5. Lerner-Ellis, J.P., Dobson, C.M., Wai, T., Watkins, D., Tirone, J.C., Leclerc, D., Dore, C., Lepage, P., Gravel, R.A. and Rosenblatt, D.S. 2004. Mutations in the MMAA gene in patients with the cblA disorder of vitamin B12 metab-olism. *Hum. Mutat.* 24: 509-516.
6. Honjo, R.S., Casella, E.B., Vieira, M.A., Bertola, D.R., Albano, L.M., Oliveira, L.A., Nomachi, S., Hanai, J., Benoist, J.F., Ellard, S., Young, E. and Kim, C.A. 2009. Spondylocostal dysostosis associated with methylmalonic aciduria. *Genet. Test. Mol. Biomarkers* 13: 181-183.
7. Froese, D.S., Kochan, G., Muniz, J.R., Wu, X., Gileadi, C., Ugochukwu, E., Krysztofinska, E., Gravel, R.A., Oppermann, U. and Yue, W.W. 2010. Structures of the human GTPase MMAA and vitamin B12-dependent methylmalonyl-CoA mutase and insight into their complex formation. *J. Biol. Chem.* 285: 38204-38213.

PROTOCOLS

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

CHROMOSOMAL LOCATION

Genetic locus: Mmaa (mouse) mapping to 8 C2.

PRODUCT

MMAA 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 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see MMAA shRNA Plasmid (m): sc-149474-SH and MMAA shRNA (m) Lentiviral Particles: sc-149474-V as alternate gene silencing products.

For independent verification of MMAA (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-149474A, sc-149474B and sc-149474C.

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

MMAA siRNA (m) is recommended for the inhibition of MMAA 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 recom-mended. 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 MMAA gene expres-sion knockdown using RT-PCR Primer: MMAA (m)-PR: sc-149474-PR (20 μ l). 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.