SANTA CRUZ BIOTECHNOLOGY, INC.

MMACHC (D-12): sc-161861

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

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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.
6. Richard, E., et al. 2009. Genetic and cellular studies of oxidative stress in methylmalonic aciduria (MMA) cobalamin deficiency type C (cblC) with homocystinuria (MMACHC). Hum. Mutat. 30: 1558-1566.
7. Kraus, J.P. 2009. cblC: advances in defining the MMACHC mutation spectrum. Hum. Mutat. 30: v.
8. Loewy, A.D., et al. 2009. Epigenetic modification of the gene for the vitamin B12 chaperone MMACHC can result in increased tumorigenicity and methionine dependence. Mol. Genet. Metab. 96: 261-267.
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CHROMOSOMAL LOCATION

Genetic locus: MMACHC (human) mapping to 1p34.1; Mmachc (mouse) mapping to 4 D1.

SOURCE

MMACHC (D-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of MMACHC of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-161861 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

MMACHC (D-12) is recommended for detection of MMACHC of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

MMACHC (D-12) is also recommended for detection of MMACHC in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for MMACHC siRNA (h): sc-88005, MMACHC siRNA (m): sc-149475, MMACHC shRNA Plasmid (h): sc-88005-SH, MMACHC shRNA Plasmid (m): sc-149475-SH, MMACHC shRNA (h) Lentiviral Particles: sc-88005-V and MMACHC shRNA (m) Lentiviral Particles: sc-149475-V.

Molecular Weight of MMACHC: 32 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

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

PROTOCOLS

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