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MMACHC (h): 293T Lysate: sc-374860

BACKGROUND

MMACHC (methylmalonic aciduria and homocystinuria type C protein), also known as cbIC, 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 cbIC, 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 B-12 and is essential for cell growth and replication. Individuals affected by methylmalonic aciduria and homocystinuria type cbIC experience negative developmental, hematologic, neurologic, metabolic, ophthalmologic and dermatologic manifestations.

REFERENCES

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7. Kraus, J.P. 2009. cbIC: advances in defining the MMACHC mutation spectrum. *Hum. Mutat.* 30: v.
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CHROMOSOMAL LOCATION

Genetic locus: MMACHC (human) mapping to 1p34.1.

PRODUCT

MMACHC (h): 293T Lysate represents a lysate of human MMACHC transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

MMACHC (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive MMACHC antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. 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 for detailed protocols and support products.