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MTHFD1 (h): 293T Lysate: sc-171409

BACKGROUND

Methylenetetrahydrofolate dehydrogenase 1 (MTHFD1) is a 935 amino acid, folate-dependent protein that is responsible for the consecutive inter-conversion of tetrahydrofolate derivatives which drive the synthesis of purine, methionine, and thymidylate. The cytosolic MRHFD1 contains 3 subunits, 5,10-methylenetetrahydrofolate dehydrogenase, 5,10-methenyltetrahydrofolate cyclohydrolase, and 10-formyltetrahydrofolate synthetase, each with distinct activities. MTHFD1 functions as a homodimer consisting of two major domains, an N-terminal containing the dehydrogenase and cyclohydrolase activities and a larger synthetase domain in the C-terminus. Mutations in the MTHFD1 gene in pregnant women are associated with an increased risk of giving birth to a child with a neural tube defect, along with a possible risk of decreased embryo survival. MTHFD1 also plays a role in migraine development, since folate metabolism is involved in migraine pathophysiology, mainly in migraine with aura.

REFERENCES

1. Arakawa, T. 1970. Congenital defects in folate utilization. *Am. J. Med.* 48: 594-598.
2. Online Mendelian Inheritance in Man, OMIM[™]. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 172460. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
3. Krajcinovic, M., et al. 2004. Role of polymorphisms in MTHFR and MTHFD1 genes in the outcome of childhood acute lymphoblastic leukemia. *Pharmacogenomics J.* 4: 66-72.
4. Christensen, K.E., et al. 2005. Disruption of the MTHFD1 gene reveals a monofunctional 10-formyltetrahydrofolate synthetase in mammalian mitochondria. *J. Biol. Chem.* 280: 7597-7602.
5. Parle-McDermott, A., et al. 2005. MTHFD1 R653Q polymorphism is a maternal genetic risk factor for severe abruptio placentae. *Am. J. Med. Genet. A* 132: 365-368.
6. Oterino, A., et al. 2005. Thymidylate synthase promoter tandem repeat and MTHFD1 R653Q polymorphisms modulate the risk for migraine conferred by the MTHFR T677 allele. *Brain Res. Mol. Brain Res.* 139: 163-168.

CHROMOSOMAL LOCATION

Genetic locus: MTHFD1 (human) mapping to 14q23.3.

PRODUCT

MTHFD1 (h): 293T Lysate represents a lysate of human MTHFD1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

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.

PROTOCOLS

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

APPLICATIONS

MTHFD1 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive MTHFD1 antibodies. Recommended use: 10-20 µl per lane.

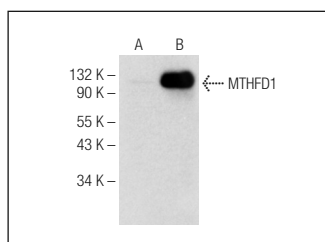
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

MTHFD1 (A-1): sc-271444 is recommended as a positive control antibody for Western Blot analysis of enhanced human MTHFD1 expression in MTHFD1 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

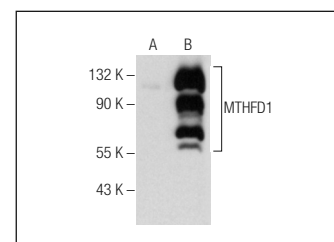
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:
1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker[™] Molecular Weight Standards: sc-2035, UltraCruz[®] Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



MTHFD1 (A-1): sc-271444. Western blot analysis of MTHFD1 expression in non-transfected: sc-117752 (A) and human MTHFD1 transfected: sc-171409 (B) 293T whole cell lysates.



MTHFD1/1L (D-9): sc-376722. Western blot analysis of MTHFD1 expression in non-transfected: sc-117752 (A) and human MTHFD1 transfected: sc-171409 (B) 293T whole cell lysates.

RESEARCH USE

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