

MTHFD1 (C-3): sc-271413

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 three 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* 132A: 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.

SOURCE

MTHFD1 (C-3) is a mouse monoclonal antibody raised against amino acids 1-120 mapping at the N-terminus of MTHFD1 of human origin.

PRODUCT

Each vial contains 200 µg IgG₁ kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

STORAGE

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

APPLICATIONS

MTHFD1 (C-3) is recommended for detection of MTHFD1 of human origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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).

Suitable for use as control antibody for MTHFD1 siRNA (h): sc-61082, MTHFD1 shRNA Plasmid (h): sc-61082-SH and MTHFD1 shRNA (h) Lentiviral Particles: sc-61082-V.

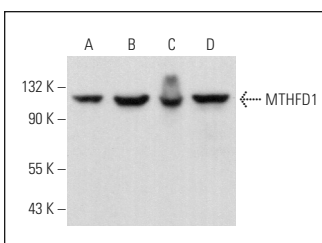
Molecular Weight of MTHFD1: 100 kDa.

Positive Controls: Jurkat whole cell lysate: sc-2204, CCRF-CEM cell lysate: sc-2225 or Hep G2 cell lysate: sc-2227.

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. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



MTHFD1 (C-3): sc-271413. Western blot analysis of MTHFD1 expression in Jurkat (A), CCRF-CEM (B), HEL 92.1.7 (C) and Hep G2 (D) whole cell lysates.

SELECT PRODUCT CITATIONS

1. Field, M.S., et al. 2015. Human mutations in methylenetetrahydrofolate dehydrogenase 1 impair nuclear *de novo* thymidylate biosynthesis. *Proc. Natl. Acad. Sci. USA* 112: 400-405.
2. Sdelci, S., et al. 2019. MTHFD1 interaction with BRD4 links folate metabolism to transcriptional regulation. *Nat. Genet.* 51: 990-998.
3. Tordonato, C., et al. 2021. miR-146 connects stem cell identity with metabolism and pharmacological resistance in breast cancer. *J. Cell Biol.* 220: e202009053.

RESEARCH USE

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