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# ETFDH (m2): 293T Lysate: sc-120129

## BACKGROUND

ETFDH (electron-transferring-flavoprotein dehydrogenase), also known as electron transfer flavoprotein-ubiquinone oxidoreductase, MADD or ETFQO, is a 617 amino acid membrane-bound electron transfer protein that exists as a monomer, localizes to the mitochondrial inner membrane and belongs to the ETF-QO/fixC family. ETFDH accepts electrons from electron-transfer flavoprotein (ETF) in the mitochondrial matrix while reducing ubiquinone in the mitochondrial membrane. ETFDH is encoded by a gene mapping to human chromosome 4q32.1, and contains one molecule of FAD and a 4Fe-4S cluster. As a result of alternative splicing events, two ETFDH isoforms exist. Defects in ETFDH are responsible for an autosomal recessive disorder of amino acid, fatty acid and choline metabolism known as glutaric aciduria type 2C (GA2C) or multiple acyl-CoA dehydrogenation deficiency (MADD). GA2C is characterized by severe hypoketotic hypoglycemia and acidosis.

## REFERENCES

1. Lehnert, W., et al. 1982. Multiple acyl-CoA dehydrogenation deficiency (glutaric aciduria type II), congenital polycystic kidneys, and symmetric warty dysplasia of the cerebral cortex in two brothers. I. Clinical, metabolic, and biochemical findings. *Eur. J. Pediatr.* 139: 56-59.
2. Böhm, N., et al. 1982. Multiple acyl-CoA dehydrogenation deficiency (glutaric aciduria type II), congenital polycystic kidneys, and symmetric warty dysplasia of the cerebral cortex in two newborn brothers. II. Morphology and pathogenesis. *Eur. J. Pediatr.* 139: 60-65.
3. White, R.A., et al. 1996. Assignment of Etfhd, Etfb, and EtfA to chromosomes 3, 7, and 13: the mouse homologs of genes responsible for glutaric acidemia type II in human. *Genomics* 33: 131-134.
4. Olsen, R.K., et al. 2003. Clear relationship between ETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. *Hum. Mutat.* 22: 12-23.
5. Online Mendelian Inheritance in Man, OMIM™. 2005. Johns Hopkins University, Baltimore, MD. MIM Number: 231675. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
6. Olsen, R.K., et al. 2007. ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. *Brain* 130: 2045-2054.
7. Fielding, A.J., et al. 2008. Electron spin relaxation enhancement measurements of interspin distances in human, porcine, and Rhodobacter electron transfer flavoprotein-ubiquinone oxidoreductase (ETF-QO). *J. Magn. Reson.* 190: 222-232.
8. Law, L.K., et al. 2009. Novel mutations in ETFDH gene in Chinese patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. *Clin. Chim. Acta* 404: 95-99.
9. Liang, W.C., et al. 2009. ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. *Neuromuscul. Disord.* 19: 212-216.

## CHROMOSOMAL LOCATION

Genetic locus: Etfhd (mouse) mapping to 3 E3.

## PRODUCT

ETFDH (m2): 293T Lysate represents a lysate of mouse ETFDH transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

## APPLICATIONS

ETFDH (m2): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive ETFDH 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](http://www.scbt.com) for detailed protocols and support products.