

ACADSB (h): 293T Lysate: sc-113801

BACKGROUND

The Acyl-CoA dehydrogenase (ACAD) family of enzymes are involved in the catabolism of fatty acids and amino acids. They provide a major source of energy for the heart and skeletal muscle. The short/branched chain specific acyl-CoA dehydrogenase (ACADSB), also designated 2-methylbutyryl-coenzyme A dehydrogenase, is a 432 amino acid protein that is ubiquitously expressed. Specifically, ACADSB forms a homotetramer within the mitochondrial matrix. ACADSB catalyzes the degradation of L-isoleucine and has the highest affinity for (s)-2-methylbutyryl-CoA, isobutyryl-CoA and 2-methylhexanoyl-CoA as substrates. Mutations in the gene encoding ACADSB result in Defects in ACADSB are the cause of short/branched-chain acyl-CoA dehydrogenase deficiency (SBCADD), an autosomal recessive disorder characterized by an increase of 2-methylbutyrylglycine and 2-methylbutyrylcarnitine in blood and urine. Patients with SBCADD have seizures and psychomotor delay as the main clinical features.

REFERENCES

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3. Korman, S.H., et al. 2005. 2-ethylhydracrylic aciduria in short/branched-chain acyl-CoA dehydrogenase deficiency: application to diagnosis and implications for the R-pathway of isoleucine oxidation. *Clin. Chem.* 51: 610-617.
4. Korman, S.H. 2006. Inborn errors of isoleucine degradation: a review. *Mol. Genet. Metab.* 89: 289-299.
5. Kanavin, O.J., et al. 2007. 2-methylbutyryl-CoA dehydrogenase deficiency associated with autism and mental retardation: a case report. *J. Med. Case Rep.* 1: 98.
6. Kamide, K., et al. 2007. Association of genetic polymorphisms of ACADSB and COMT with human hypertension. *J. Hypertens.* 25: 103-110.
7. Sass, J.O., et al. 2008. 2-Methylbutyryl-coenzyme A dehydrogenase deficiency: functional and molecular studies on a defect in isoleucine catabolism. *Mol. Genet. Metab.* 93: 30-35.

CHROMOSOMAL LOCATION

Genetic locus: ACADSB (human) mapping to 10q26.13.

PRODUCT

ACADSB (h): 293T Lysate represents a lysate of human ACADSB 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.

APPLICATIONS

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

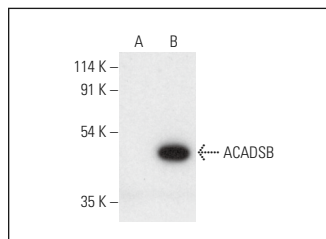
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

ACADSB (C-9): sc-398773 is recommended as a positive control antibody for Western Blot analysis of enhanced human ACADSB expression in ACADSB 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



ACADSB (C-9): sc-398773. Western blot analysis of ACADSB expression in non-transfected: sc-117752 (A) and human ACADSB transfected: sc-113801 (B) 293T whole cell lysates.

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.