SANTA CRUZ BIOTECHNOLOGY, INC.

BCKDHB (h): 293T Lysate: sc-115538

BACKGROUND

BCKDHB (branched chain keto acid dehydrogenase E1, β polypeptide), also known as 2-oxoisovalerate dehydrogenase subunit β mitochondrial or E1B, is a 392 amino acid mitochondrial matrix protein and component of branched-chain keto acid dehydrogenase, a multienzyme complex involved in the catabolism of branched-chain amino acids. Existing as a heterodimer, BCKDHB is encoded by a gene mapping to human chromosome 6q14.1, whose defects are the cause of an autosomal recessive disorder known as maple syrup urine disease type IB (MSUD1B). Characterized by urine with maple syrup odor, patients with maple syrup urine disease may suffer severe neurological damage, mental retardation and feeding problems.

REFERENCES

1. Chuang, J.L., et al. 1990. Molecular cloning of the mature E1b-β subunit of human branched-chain α-keto acid dehydrogenase complex. FEBS Lett. 262: 305-309.
2. Zneimer, S.M., et al. 1991. Regional assignment of two genes of the human branched-chain α-keto acid dehydrogenase complex: the E1 β gene (BCKDHB) to chromosome 6p21-22 and the E2 gene (DBT) to chromosome 1p31. Genomics 10: 740-747.
3. Patel, M.S. and Harris, R.A. 1995. Mammalian α-keto acid dehydrogenase complexes: gene regulation and genetic defects. FASEB J. 9: 1164-1172.
4. Chuang, J.L., et al. 1996. Maple syrup urine disease: the E1β gene of human branched-chain α-ketoacid dehydrogenase complex has 11 rather than 10 exons, and the 3' UTR in one of the two E1b mRNAs arises from intronic sequences. Am. J. Hum. Genet. 58: 1373-1377.
5. Chuang, D.T. 1998. Maple syrup urine disease: it has come a long way. J. Pediatr. 132: S17-S23.
6. Chuang, J.L., et al. 2004. Structural and biochemical basis for novel mutations in homozygous Israeli maple syrup urine disease patients: a proposed mechanism for the thiamin-responsive phenotype. J. Biol. Chem. 279: 17792-17800.
7. Machius, M., et al. 2006. A versatile conformational switch regulates reactivity in human branched-chain α-ketoacid dehydrogenase. Structure 14: 287-298.
8. Kang, H., et al. 2008. Branched chain α-keto acid dehydrogenase, E1-β subunit gene is associated with premature ovarian failure. Fertil. Steril. 89: 728-731.

CHROMOSOMAL LOCATION

Genetic locus: BCKDHB (human) mapping to 6q14.1.

PRODUCT

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

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

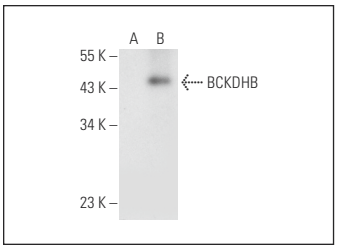
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

BCKDHB (H-6): sc-374630 is recommended as a positive control antibody for Western Blot analysis of enhanced human BCKDHB expression in BCKDHB 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



BCKDHB (H-6): sc-374630. Western blot analysis of BCKDHB expression in non-transfected: sc-117752 (**A**) and human BCKDHB transfected: sc-115538 (**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.