

ALDH5A1 (h2): 293T Lysate: sc-175083

BACKGROUND

Aldehyde dehydrogenases (ALDHs) mediate the NADP⁺-dependent oxidation of aldehydes into acids and play an important role in the detoxification of alcohol-derived acetaldehyde, as well as in lipid peroxidation and in the metabolism of corticosteroids, biogenic amines and neurotransmitters. ALDH5A1 (aldehyde dehydrogenase 5 family, member A1), also known as SSDH or SSADH, is a 535 amino acid protein that localizes to the mitochondria and belongs to the aldehyde dehydrogenase family. Expressed in a variety of tissues, including liver, heart, lung, brain, kidney and placenta, ALDH5A1 is required for gamma-aminobutyric acid (GABA) recycling from the synaptic cleft. Mutations of ALDH5A1 leads to succinate semialdehyde dehydrogenase deficiency (SSADH deficiency) that is characterized by severe ataxia and by mildly retarded psychomotor development.

REFERENCES

1. Kang, J.H., et al. 2005. High-level expression and characterization of the recombinant enzyme, and tissue distribution of human succinic semialdehyde dehydrogenase. *Protein Expr. Purif.* 44: 16-22.
2. Leone, O., et al. 2006. A human derived SSADH coding variant is replacing the ancestral allele shared with primates. *Ann. Hum. Biol.* 33: 593-603.
3. Jansen, E.E., et al. 2006. Increased guanidino species in murine and human succinate semialdehyde dehydrogenase (SSADH) deficiency. *Biochim. Biophys. Acta* 1762: 494-498.
4. Blasi, P., et al. 2006. SSADH variation in primates: intra- and interspecific data on a gene with a potential role in human cognitive functions. *J. Mol. Evol.* 63: 54-68.
5. Mehta, A.K., et al. 2006. Succinate semialdehyde dehydrogenase deficiency does not downregulate γ -hydroxybutyric acid binding sites in the mouse brain. *Mol. Genet. Metab.* 88: 86-89.
6. Barcelo-Coblijn, G., et al. 2007. Lipid abnormalities in succinate semialdehyde dehydrogenase (ALDH5A1^{-/-}) deficient mouse brain provide additional evidence for myelin alterations. *Biochim. Biophys. Acta* 1772: 556-562.
7. Knerr, I., et al. 2007. Therapeutic concepts in succinate semialdehyde dehydrogenase (SSADH; ALDH5A1) deficiency (γ -hydroxybutyric aciduria). Hypotheses evolved from 25 years of patient evaluation, studies in ALDH5A1^{-/-} mice and characterization of γ -hydroxybutyric acid pharmacology. *J. Inher. Metab. Dis.* 30: 279-294.
8. Sauer, S.W., et al. 2007. Enzymatic and metabolic evidence for a region specific mitochondrial dysfunction in brains of murine succinic semialdehyde dehydrogenase deficiency (ALDH5A1^{-/-} mice). *Neurochem. Int.* 50: 653-659.
9. Malaspina, P., et al. 2009. Comparative genomics of aldehyde dehydrogenase 5A1 (succinate semialdehyde dehydrogenase) and accumulation of γ -hydroxybutyrate associated with its deficiency. *Hum. Genomics* 3: 106-120.

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.

CHROMOSOMAL LOCATION

Genetic locus: ALDH5A1 (human) mapping to 6p22.3.

PRODUCT

ALDH5A1 (h2): 293T Lysate represents a lysate of human ALDH5A1 transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

ALDH5A1 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive ALDH5A1 antibodies. Recommended use: 10-20 μ l per lane.

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

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.