

Antiquitin (h): 293T Lysate: sc-111586

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. Antiquitin, also known as ALDH7A1 (aldehyde dehydrogenase 7 family, member A1), ATQ1, EPD or PDE, is a 539 amino acid protein that is highly expressed in ovary, heart, eye, kidney and ear tissue and plays an important role in the detoxification of lipid peroxidation- and alcohol metabolism-related aldehydes. Mutations in the gene encoding Antiquitin are the cause of pyridoxine-dependent epilepsy (PDE), a rare disorder that is characterized by seizures that begin at infancy and involve muscle rigidity, convulsions and loss of consciousness. Additionally, PDE is associated with poor muscle tone, hypothermia and irritability.

REFERENCES

- Skvorak, A.B., et al. 1997. An ancient conserved gene expressed in the human inner ear: identification, expression analysis, and chromosomal mapping of human and mouse antiquitin (ATQ1). *Genomics* 46: 191-199.
- Mills, P.B., et al. 2006. Mutations in antiquitin in individuals with pyridoxine-dependent seizures. *Nat. Med.* 12: 307-309.
- Salomons, G.S., et al. 2007. An intriguing "silent" mutation and a founder effect in antiquitin (ALDH7A1). *Ann. Neurol.* 62: 414-418.
- Bok, L.A., et al. 2007. Pyridoxine-dependent seizures in Dutch patients: diagnosis by elevated urinary α -aminoadipic semialdehyde levels. *Arch. Dis. Child.* 92: 687-689.
- Plecko, B., et al. 2007. Biochemical and molecular characterization of 18 patients with pyridoxine-dependent epilepsy and mutations of the antiquitin (ALDH7A1) gene. *Hum. Mutat.* 28: 19-26.
- Kanno, J., et al. 2007. Allelic and non-allelic heterogeneities in pyridoxine dependent seizures revealed by ALDH7A1 mutational analysis. *Mol. Genet. Metab.* 91: 384-389.
- Been, J.V., et al. 2008. Mutations in the ALDH7A1 gene cause pyridoxine-dependent seizures. *Arq. Neuropsiquiatr.* 66: 288.
- Bennett, C.L., et al. 2008. Prevalence of ALDH7A1 mutations in 18 North American pyridoxine-dependent seizure (PDS) patients. *Epilepsia* 50: 1167-1175.
- Online Mendelian Inheritance in Man, OMIMTM. 2008. Johns Hopkins University, Baltimore, MD. MIM Number: 107323. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

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.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.

CHROMOSOMAL LOCATION

Genetic locus: ALDH7A1 (human) mapping to 5q23.2.

PRODUCT

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

APPLICATIONS

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

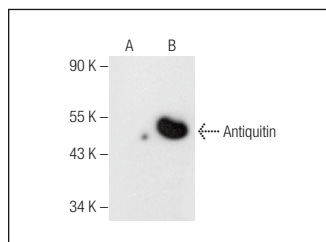
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

Antiquitin (D-7): sc-376906 is recommended as a positive control antibody for Western Blot analysis of enhanced human Antiquitin expression in Antiquitin 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-IgG κ BP-HRP: sc-516102 or m-IgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz MarkerTM
Molecular Weight Standards: sc-2035, UltraCruz[®] Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



Antiquitin (D-7): sc-376906. Western blot analysis of Antiquitin expression in non-transfected: sc-117752 (A) and human Antiquitin transfected: sc-111586 (B) 293T whole cell lysates.

RESEARCH USE

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