Antiquitin (Y-20): sc-79401



The Power to Question

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., Robertson, N.G., Yin, Y., Weremowicz, S., Her, H., Bieber, F.R., Beisel, K.W., Lynch, E.D., Beier, D.R. and Morton, C.C. 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.
- 2. Mills, P.B., Struys, E., Jakobs, C., Plecko, B., Baxter, P., Baumgartner, M., Willemsen, M.A., Omran, H., Tacke, U., Uhlenberg, B., Weschke, B. and Clayton, P.T. 2006. Mutations in Antiquitin in individuals with pyridoxine-dependent seizures. Nat. Med. 12: 307-309.
- Salomons, G.S., Bok, L.A., Struys, E.A., Pope, L.L., Darmin, P.S., Mills, P.B., Clayton, P.T., Willemsen, M.A. and Jakobs, C. 2007. An intriguing "silent" mutation and a founder effect in Antiquitin (ALDH7A1). Ann. Neurol. 62: 414-418.
- 4. Bok, L.A., Struys, E., Willemsen, M.A., Been, J.V. and Jakobs, C. 2007. Pyridoxine-dependent seizures in Dutch patients: diagnosis by elevated urinary α -aminoadipic semialdehyde levels. Arch. Dis. Child. 92: 687-689.
- Plecko, B., Paul, K., Paschke, E., Stoeckler-Ipsiroglu, S., Struys, E., Jakobs, C., Hartmann, H., Luecke, T., di Capua, M., Korenke, C., Hikel, C., Reutershahn, E., Freilinger, M., Baumeister, F., Bosch, F. and Erwa, W. 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., Kure, S., Narisawa, A., Kamada, F., Takayanagi, M., Yamamoto, K., Hoshino, H., Goto, T., Takahashi, T., Haginoya, K., Tsuchiya, S., Baumeister, F.A., Hasegawa, Y., Aoki, Y., Yamaguchi, S. and Matsubara, Y. 2007. Allelic and non-allelic heterogeneities in pyridoxine dependent seizures revealed by ALDH7A1 mutational analysis. Mol. Genet. Metab. 91: 384-389.

CHROMOSOMAL LOCATION

Genetic locus: ALDH7A1 (human) mapping to 5q23.2; Aldh7a1 (mouse) mapping to 18 D3.

SOURCE

Antiquitin (Y-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of Antiquitin of human origin.

PRODUCT

Each vial contains 200 μg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-79401 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Antiquitin (Y-20) is recommended for detection of Antiquitin of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 μ g per 100-500 μ g of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

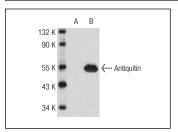
Antiquitin (Y-20) is also recommended for detection of Antiquitin in additional species, including equine, canine, bovine and avian.

Suitable for use as control antibody for Antiquitin siRNA (h): sc-72507, Antiquitin siRNA (m): sc-72508, Antiquitin shRNA Plasmid (h): sc-72507-SH, Antiquitin shRNA Plasmid (m): sc-72508-SH, Antiquitin shRNA (h) Lentiviral Particles: sc-72507-V and Antiquitin shRNA (m) Lentiviral Particles: sc-72508-V.

Molecular Weight of Antiquitin: 55 kDa.

Positive Controls: Antiquitin (h): 293T Lysate: sc-111586.

DATA



Antiquitin (Y-20): sc-79401. Western blot analysis of Antiquitin expression in non-transfected: sc-117752 (A) and human Antiquitin transfected: sc-111586 (B) 293T whole cell lysates.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

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