

Antiquitin (C-4): sc-365642

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

1. 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.
2. Mills, P.B., et al. 2006. Mutations in antiquitin in individuals with pyridoxine-dependent seizures. *Nat. Med.* 12: 307-309.
3. Salomons, G.S., et al. 2007. An intriguing "silent" mutation and a founder effect in antiquitin (ALDH7A1). *Ann. Neurol.* 62: 414-418.
4. 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.
5. 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.
6. 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.
7. Been, J.V., et al. 2008. Mutations in the ALDH7A1 gene cause pyridoxine-dependent seizures. *Arq. Neuropsiquiatr.* 66: 288.

CHROMOSOMAL LOCATION

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

SOURCE

Antiquitin (C-4) is a mouse monoclonal antibody specific for an epitope mapping between amino acids 489-506 at the C-terminus of Antiquitin of human origin.

PRODUCT

Each vial contains 200 μ g IgG_{2b} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-365642 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% stabilizer protein).

APPLICATIONS

Antiquitin (C-4) is recommended for detection of Antiquitin of human origin by Western Blotting (starting dilution 1:100, 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), immunohistochemistry (including paraffin-embedded sections) (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 (C-4) 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 shRNA Plasmid (h): sc-72507-SH and Antiquitin shRNA (h) Lentiviral Particles: sc-72507-V.

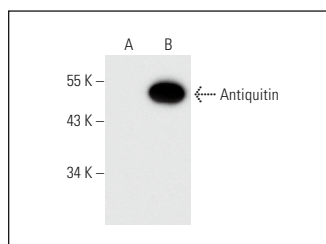
Molecular Weight of Antiquitin: 55 kDa.

Positive Controls: HeLa whole cell lysate: sc-2200 or Antiquitin (h): 293T lysate: sc-111586.

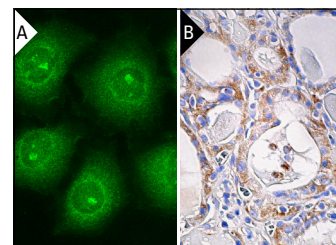
RECOMMENDED SUPPORT REAGENTS

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 Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgG κ BP-FITC: sc-516140 or m-IgG κ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850. 4) Immunohistochemistry: use m-IgG κ BP-HRP: sc-516102 with DAB, 50X: sc-24982 and Immunohistomount: sc-45086, or Organo/Limonene Mount: sc-45087.

DATA



Antiquitin (C-4): sc-365642. Western blot analysis of Antiquitin expression in non-transfected: sc-117752 (A) and human Antiquitin transfected: sc-111586 (B) 293T whole cell lysates.



Antiquitin (C-4): sc-365642. Immunofluorescence staining of methanol-fixed HeLa cells showing cytoplasmic and nuclear localization (A). Immunoperoxidase staining of formalin fixed, paraffin-embedded human thyroid tissue showing cytoplasmic staining of glandular cells (B).

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.