SANTA CRUZ BIOTECHNOLOGY, INC.

COLQ (D-22): sc-133472



BACKGROUND

COLQ (collagen-like tail subunit (single strand of homotrimer) of asymmetric acetylcholinesterase), also known as EAD, is a 455 amino acid protein that localizes to the end plate of skeletal muscle. COLQ anchors the catalytic subunits of asymmetric AChE (acetylcholinesterase) to the basal lamina at the neuromuscular junctions of vertebrates. Mutations of COLQ lead to congenital myasthenic syndromes, which are rare autosomal recessive diseases characterized by general weakness increased by exertion, ophthalmoplegia and refractoriness to anticholinesterase drugs. Eight isoforms exist due to alternative splicing events.

REFERENCES

- 1. Shapira, Y.A., et al. 2002. Three novel COLO mutations and variation of phenotypic expressivity due to G240X. Neurology 58: 603-609.
- Ishigaki, K., et al. 2003. Two novel mutations in the COLO gene cause endplate acetylcholinesterase deficiency. Neuromuscul. Disord. 13: 236-244.
- Lee, H.H., et al. 2004. Transcriptional regulation of acetylcholinesterase-associated collagen COLO: differential expression in fast and slow twitch muscle fibers is driven by distinct promoters. J. Biol. Chem. 279: 27098-27107.
- Ting, A.K., et al. 2005. Transcriptional regulation of acetylcholinesteraseassociated collagen COLQ in fast- and slow-twitch muscle fibers. Chem. Biol. Interact. 157-158: 63-70.
- Girard, E., et al. 2006. Remodeling of the neuromuscular junction in mice with deleted exons 5 and 6 of acetylcholinesterase. J. Mol. Neurosci. 30: 99-100.
- Tsim, K.W., et al. 2006. Transcriptional control of different acetylcholinesterase subunits in formation and maintenance of vertebrate neuromuscular junctions. J. Mol. Neurosci. 30: 189-192.
- Schreiner, F., et al. 2007. Novel COLQ mutation 950delC in synaptic congenital myasthenic syndrome and symptomatic heterozygous relatives. Neuromuscul. Disord. 17: 262-265.
- Mihaylova, V., et al. 2008. Clinical and molecular genetic findings in COLQmutant congenital myasthenic syndromes. Brain 131: 747-759.
- Lau, F.T., et al. 2008. Myocyte enhancer factor 2 mediates acetylcholineinduced expression of acetylcholinesterase-associated collagen COLQ in cultured myotubes. Mol. Cell. Neurosci. 39: 429-438.

CHROMOSOMAL LOCATION

Genetic locus: COLQ (human) mapping to 3p25.1.

SOURCE

COLQ (D-22) is an affinity purified rabbit polyclonal antibody raised against synthetic COLQ peptide of human origin.

PRODUCT

Each vial contains 50 μg IgG in 500 μl PBS with < 0.1% sodium azide, 0.1% gelatin and < 0.02% sucrose.

APPLICATIONS

COLQ (D-22) is recommended for detection of COLQ of 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)] and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for COLO siRNA (h): sc-72964, COLO shRNA Plasmid (h): sc-72964-SH and COLO shRNA (h) Lentiviral Particles: sc-72964-V.

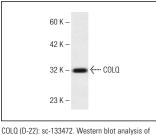
Molecular Weight of COLQ isoforms I-VII: 28-48 kDa.

Positive Controls: Jurkat whole cell lysate: sc-2204.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml).

DATA



COLQ (D-22). SC-133472. Western blot analysis c COLQ expression in Jurkat whole cell lysate.

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.

PROTOCOLS

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