

# COLQ Rabbit pAb

COLQ Rabbit pAb Catalog # AP54326

#### **Product Information**

**Application** IHC-P, IHC-F, IF, E

Primary Accession <u>Q9Y215</u>

**Reactivity** Rat, Pig, Mouse, Horse

Host Rabbit
Clonality Polyclonal
Calculated MW 47766
Physical State Liquid

Immunogen KLH conjugated synthetic peptide derived from human COLQ

Epitope Specificity 301-400/455

Isotype IgG

**Purity** affinity purified by Protein A

**Buffer** 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.

**SUBCELLULAR LOCATION** Cell junction; synapse.

SIMILARITY Belongs to the COLQ family. Contains 2 collagen-like domains.

Post-translational The triple-helical tail is stabilized by disulfide bonds at each end.

modifications

DISEASE

Defects in COLQ are the cause of congenital myasthenic syndrome Eng

Defects in COLQ are the cause of congenital myasthenic syndrome Engel type (CMSE) [MIM:603034]; also known as end-plate acetylcholinesterase deficiency or congenital myasthenic syndrome type IC (CMS-IC). CMSE is a rare autosomal recessive congenital myasthenic syndrome characterized by onset during childhood, generalized weakness, abnormal fatigability on exertion, refrectoriness to acetylcholinesterase drugs, decremental electromyographic response and morphological abnormalities of the

neuromuscular junctions.

**Important Note** This product as supplied is intended for research use only, not for use in

human, therapeutic or diagnostic applications.

**Background Descriptions** This gene encodes the subunit of a collagen-like molecule associated with

acetylcholinesterase in skeletal muscle. Each molecule is composed of three identical subunits. Each subunit contains a proline-rich attachment domain (PRAD) that binds an acetylcholinesterase tetramer to anchor the catalytic subunit of the enzyme to the basal lamina. Mutations in this gene are associated with endplate acetylcholinesterase deficiency. Multiple transcript variants encoding different isoforms have been found for this gene. [provided

by RefSeq, Jul 2008]

## **Additional Information**

**Gene ID** 8292

Other Names Acetylcholinesterase collagenic tail peptide, AChE Q subunit,

Acetylcholinesterase-associated collagen, COLQ

**Target/Specificity** Found at the end plate of skeletal muscle.

**Dilution** IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:500

0-10000

**Storage** Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When

reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody

is stable for at least two weeks at 2-4 °C.

### **Protein Information**

Name COLQ

**Function** Anchors the catalytic subunits of asymmetric AChE to the synaptic basal

lamina.

**Cellular Location** Synapse.

**Tissue Location** Found at the end plate of skeletal muscle.

## **Background**

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