

Rabbit Anti-COLQ antibody

SL10932R

Product Name:	COLQ
Chinese Name:	乙酰胆碱酯酶相关Collagen protein多肽抗体
Alias:	Acetylcholinesterase-associated collagen; AChE Q subunit; asymmetric acetylcholinesterase; Collagen-like tail subunit (single strand of homotrimer) of asymmetric acetylcholinesterase; Colq; COLQ_HUMAN; EAD;
	OTTHUMP00000209566; OTTHUMP00000209567; single strand of homotrimeric collagen-like tail subunit of asymmetric acetylcholinesterase.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Pig, Horse, Rabbit,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications.
Molecular weight:	optimal dilutions/concentrations should be determined by the end user. 45kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	lmg/ml
immunogen:	KLH conjugated synthetic peptide derived from human COLQ:301-400/455
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20 °C. 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.
PubMed:	PubMed
Product Detail:	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]

Function:

Anchors the catalytic subunits of asymmetric AChE to the synaptic basal lamina.

Subcellular Location:

Cell junction; synapse.

Tissue Specificity:

Found at the end plate of skeletal muscle.

Post-translational modifications:

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

DISEASE:

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.

Similarity:

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

SWISS:

O9Y215

Gene ID:

8292

Database links:

Entrez Gene: 8292 Human

Omim: 603033 Human

SwissProt: Q9Y215 Human

Unigene: 146735 Human

Important Note: This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

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