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COLQ Rabbit pAb

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Catalog # AP54326

Product Information

Application	IHC-P, IHC-F, IF, E
Primary Accession	Q9Y215
Predicted	Human, Mouse, Rat, Pig, Horse, Rabbit
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 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.
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=1:5000-10000
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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