

Product name:	COLQ Rabbit Polyclonal Antibody
Cat number:	ABN09221
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from human COLQ
Reactivity:	Human, Mouse, Rat
Applications:	WB 1:500-1:2000, IHC 1:50-1:200, ICC/IF 1:50-1:200
Molecular Weight:	50kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

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],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, refractoriness to acetylcholinesterase drugs, decremental electromyographic response and morphological abnormalities of the neuromuscular junctions.,domain:The proline-rich attachment domain (PRAD) binds the AChE catalytic subunits.,function:Anchors the catalytic subunits of asymmetric AChE to the synaptic basal lamina.,PTM:The triple-helical tail is stabilized by disulfide bonds at each end.,similarity:Belongs to the COLQ family.,similarity:Contains 2 collagen-like domains.,subunit:Homotrimer. Component of the asymmetric form of AChE, a disulfide-bonded oligomer composed of the collagenic subunits (Q) and a variable number of asymmetric catalytic subunits (T). The N-terminal of a collagenic subunit (Q) associates with the C-terminal of a catalytic subunit (T).,tissue specificity:Found at the end plate of skeletal muscle.,