

COLQ gene

collagen like tail subunit of asymmetric acetylcholinesterase

Normal Function

The *COLQ* gene provides instructions for making a protein that plays an important role in the neuromuscular junction. The neuromuscular junction is the area between the ends of nerve cells and muscle cells where signals are relayed to trigger muscle movement.

The ColQ protein anchors another protein called acetylcholinesterase to the muscle cell membrane at the neuromuscular junction. The ColQ protein is made up of three identical parts (subunits). Each subunit attaches (binds) to a bundle of four acetylcholinesterase proteins. Acetylcholinesterase plays a role in regulating the length of signaling between nerve cells and muscle cells by breaking down the signaling protein acetylcholine.

Health Conditions Related to Genetic Changes

Congenital myasthenic syndrome

More than 35 mutations in the *COLQ* gene have been found to cause congenital myasthenic syndrome. Most of these mutations change single protein building blocks (amino acids) in the ColQ protein or lead to the production of a shortened, nonfunctional protein. A lack of functional ColQ protein leads to a reduction in the amount of acetylcholinesterase that is available in the neuromuscular junction. As a result, acetylcholine is not broken down so signaling between nerve and muscle cells is prolonged. This signaling overload can damage muscle cells, leading to the muscle weakness characteristic of congenital myasthenic syndrome.

Other Names for This Gene

- acetylcholinesterase collagenic tail peptide
- acetylcholinesterase-associated collagen
- AChE Q subunit
- collagen-like tail subunit (single strand of homotrimer) of asymmetric acetylcholinesterase
- collagenic tail of endplate acetylcholinesterase

- COLQ_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COLQ ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8292\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8292[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28COLQ%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- COLLAGENIC TAIL OF ENDPLATE ACETYLCHOLINESTERASE; COLQ (<https://omim.org/entry/603033>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8292>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COLQ\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COLQ[gene]))

References

- Barisic N, Chaouch A, Muller JS, Lochmuller H. Genetic heterogeneity and pathophysiological mechanisms in congenital myasthenic syndromes. *Eur J Paediatr Neurol*. 2011 May;15(3):189-96. doi: 10.1016/j.ejpn.2011.03.006. Epub 2011 Apr 17. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21498094>)
- Engel AG, Shen XM, Selcen D, Sine SM. What have we learned from the congenital myasthenic syndromes. *J Mol Neurosci*. 2010 Jan;40(1-2):143-53. doi:10.1007/s12031-009-9229-0. Epub 2009 Aug 18. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19688192>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3050586/>)
- Engel AG. Congenital myasthenic syndromes in 2012. *Curr Neurol Neurosci Rep*. 2012 Feb;12(1):92-101. doi: 10.1007/s11910-011-0234-7. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21997714>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4209912/>)
- Kinali M, Beeson D, Pitt MC, Jungbluth H, Simonds AK, Aloysius A, Cockerill H, Davis T, Palace J, Manzur AY, Jimenez-Mallebrera C, Sewry C, Muntoni F, Robb

SA. Congenital myasthenic syndromes in childhood: diagnostic and management challenges. J Neuroimmunol. 2008 Sep 15;201-202:6-12. doi:10.1016/j.jneuroim.2008.06.026. Epub 2008 Aug 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18707767>)

Genomic Location

The *COLQ* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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