# CHRNG gene

cholinergic receptor nicotinic gamma subunit

## Normal Function

The *CHRNG* gene provides instructions for making the gamma (γ) protein component ( subunit) of the acetylcholine receptor (AChR) protein. The AChR protein is found in the membrane of skeletal muscle cells and is critical for signaling between nerve and muscle cells. Signaling between these cells is necessary for movement. The AChR protein consists of five subunits, each of which is produced from a different gene. The subunits are assembled into the AChR protein in the endoplasmic reticulum, a cell structure involved in protein processing and transport, before being transported to the cell membrane. The γ subunit is found only in the fetal AChR protein. At about the thirty-third week of pregnancy, the γ subunit is replaced by the epsilon (ε) subunit, which is produced by the *CHRNE* gene, to form the adult AChR protein.

## Health Conditions Related to Genetic Changes

Multiple pterygium syndrome

At least 14 mutations in the *CHRNG* gene have been found to cause multiple pterygium syndrome, a condition characterized by webbing of the skin (pterygium) and a lack of muscle movement (akinesia) before birth. These mutations include replacing, adding, or deleting DNA building blocks (nucleotides). *CHRNG* gene mutations result in an impaired or missing γ subunit. The severity of the *CHRNG* gene mutation influences the severity of the condition. Typically, mutations that prevent the production of any γ subunit will result in lethal multiple pterygium syndrome, which is fatal before birth, while mutations that allow the production of some γ subunit will lead to the milder form of this condition called multiple pterygium syndrome, Escobar type. A shortage of a functional γ subunit prevents the fetal AChR protein from being assembled or properly placed in the muscle cell membrane. As a result, the fetal AChR protein cannot function and the communication between nerve and muscle cells in the developing fetus is impaired. A lack of signaling between nerve and muscle cells leads to akinesia and pterygium before birth, and many of the other signs and symptoms of multiple pterygium syndrome.

## Other Names for This Gene

- acetylcholine gamma muscle receptor subunit
- acetylcholine receptor subunit gamma
- acetylcholine receptor, muscle, gamma subunit
- ACHG_HUMAN
- ACHRG
- cholinergic gamma nicotinic receptor
- cholinergic receptor, nicotinic gamma
- cholinergic receptor, nicotinic, gamma
- cholinergic receptor, nicotinic, gamma (muscle)
- cholinergic receptor, nicotinic, gamma polypeptide

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CHRNG (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1146[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CHRNG%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CHOLINERGIC RECEPTOR, NICOTINIC, GAMMA POLYPEPTIDE; CHRNG (https://omim.org/entry/100730)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1146)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CHRNG[gene])

## References


- Hoffmann K, Muller JS, Stricker S, Megarbane A, Rajab A, Lindner TH, Cohen M, Chouery E, Adaimy L, Ghanem I, Delague V, Boltshauser E, Talim B, Horvath R, Robinson PN, Lochmuller H, Hubner C, Mundlos S. Escobar syndrome is a

prenatalmyasthenia caused by disruption of the acetylcholine receptor fetal gammasubunit. Am J Hum Genet. 2006 Aug;79(2):303-12. doi: 10.1086/506257. Epub 2006Jun 20. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16826520) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1559482/)

- Michalk A, Stricker S, Becker J, Rupps R, Pantzar T, Miertus J, Botta G,Naretto VG, Janetzki C, Yaqoob N, Ott CE, Seelow D, Wieczorek D, Fiebig B, WirthB, Hoopmann M, Walther M, Korber F, Blankenburg M, Mundlos S, Heller R, HoffmannK. Acetylcholine receptor pathway mutations explain various fetal akinesiadeformation sequence disorders. Am J Hum Genet. 2008 Feb;82(2):464-76. doi:10.1016/j.ajhg.2007.11.006. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18252226) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2427255/)
- Morgan NV, Brueton LA, Cox P, Greally MT, Tolmie J, Pasha S, Aligianis IA, vanBokhoven H, Marton T, Al-Gazali L, Morton JE, Oley C, Johnson CA, Trembath RC,Brunner HG, Maher ER. Mutations in the embryonal subunit of the acetylcholinereceptor (CHRNG) cause lethal and Escobar variants of multiple pterygiumsyndrome. Am J Hum Genet. 2006 Aug;79(2):390-5. doi: 10.1086/506256. Epub 2006Jun 20. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16826531) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1559492/)

## Genomic Location

The *CHRNG* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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