CHRNE gene

cholinergic receptor nicotinic epsilon subunit

Normal Function

The *CHRNE* gene provides instructions for making the epsilon (ε) component (subunit) of the acetylcholine receptor (AChR) protein. The AChR protein is found in the membrane of skeletal muscle cells and plays a critical role in the neuromuscular junction, which is the area where signaling between nerve and muscle cells occurs. Signaling between nerve and muscle 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. There are two major forms of the AChR protein, a fetal type that is present before birth and an adult type. The ε subunit is found only in the adult AChR protein. At about the 33rd week of pregnancy, the ε subunit replaces the gamma (γ) subunit (found only in fetal AChR) to form adult AChR protein.

Health Conditions Related to Genetic Changes

Congenital myasthenic syndrome

More than 90 mutations in the *CHRNE* gene have been found to cause congenital myasthenic syndrome. Most of these mutations replace one DNA building block (nucleotide) in the gene, but other mutations add or delete small sections of DNA. These mutations can lead to the production of an altered ε subunit. A change in the ε subunit leads to an increase in AChR protein signaling or a decrease in AChR protein signaling, which impairs cell-to-cell communication at the neuromuscular junction. Decreased signaling can lead to a decrease in muscle movement and cause weakness, while an increase in signaling can damage muscle cells and cause weakness. Problems with communication between nerve and muscle cells can lead to the signs and symptoms of congenital myasthenic syndrome, including muscle weakness and delayed development of motor skills such as crawling and walking. Some people with *CHRNE* mutations seem to have a milder course of the disease compared with other affected individuals, likely because the fetal γ subunit (active at low levels after birth) can partially compensate for the lack of ε subunit in the adult AChR protein.

Other Names for This Gene

- acetylcholine receptor subunit epsilon
- ACHE_HUMAN
- AchR epsilon subunit
- ACHRE
- cholinergic receptor, nicotinic epsilon
- cholinergic receptor, nicotinic, epsilon
- cholinergic receptor, nicotinic, epsilon (muscle)
- cholinergic receptor, nicotinic, epsilon polypeptide

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CHRNE (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1145[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28CHRNE%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+2520+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CHOLINERGIC RECEPTOR, NICOTINIC, EPSILON POLYPEPTIDE; CHRNE (https://omim.org/entry/100725)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1145)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CHRNE[gene])

References

- Engel AG. Current status of the congenital myasthenic syndromes. NeuromusculDisord. 2012 Feb;22(2):99-111. doi: 10.1016/j.nmd.2011.10.009. Epub 2011 Nov 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22104196) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3269564/)

- 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 managementchallenges. 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)
- Palace J, Lashley D, Bailey S, Jayawant S, Carr A, McConville J, Robb S,Beeson D. Clinical features in a series of fast channel congenital myastheniasyndrome. Neuromuscul Disord. 2012 Feb;22(2):112-7. doi:10.1016/j.nmd.2011.08.002. Epub 2011 Sep 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21940170)

Genomic Location

The *CHRNE* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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