

## RAPSN gene

receptor associated protein of the synapse

### Normal Function

The *RAPSN* gene provides instructions for making a protein called rapsyn that attaches (binds) to the different parts (subunits) of a protein found in the muscle cell membrane called acetylcholine receptor (AChR). This binding helps keep the receptor subunits together and anchors the AChR protein in the muscle cell membrane. The AChR protein plays a critical role in the normal function of 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.

### Health Conditions Related to Genetic Changes

#### Congenital myasthenic syndromes

MedlinePlus Genetics provides information about Congenital myasthenic syndromes

#### Multiple pterygium syndrome

MedlinePlus Genetics provides information about Multiple pterygium syndrome

### Other Names for This Gene

- RAPSN\_HUMAN
- RAPSYN
- receptor-associated protein of the synapse
- RING finger protein 205
- RNF205

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of RAPSN ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5913\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5913[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RAPSN%5BTIAB%5D%29+OR+%28RAPSYN%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>)

## Catalog of Genes and Diseases from OMIM

- RECEPTOR-ASSOCIATED PROTEIN OF THE SYNAPSE, 43-KD; RAPSN (<https://omim.org/entry/601592>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5913>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RAPSN\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RAPSN[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>)
- Beeson D, Webster R, Cossins J, Lashley D, Spearman H, Maxwell S, Slater CR, Newsom-Davis J, Palace J, Vincent A. Congenital myasthenic syndromes and the formation of the neuromuscular junction. *Ann N Y Acad Sci.* 2008;1132:99-103. doi:10.1196/annals.1405.049. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18567858>)
- 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/>)
- Engel AG. Current status of the congenital myasthenic syndromes. *Neuromuscul Disord.* 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 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>)
- Milone M, Shen XM, Selcen D, Ohno K, Brengman J, Iannaccone ST, Harper CM,

Engel AG. Myasthenic syndrome due to defects in rapsyn: Clinical and molecular findings in 39 patients. *Neurology*. 2009 Jul 21;73(3):228-35. doi:10.1212/WNL.0b013e3181ae7cbc. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19620612>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2715575/>)

## **Genomic Location**

The *RAPSN* gene is found on chromosome 11 (<https://medlineplus.gov/genetics/chromosome/11/>).

**Last updated November 1, 2011**