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 syndrome

At least 45 mutations in the *RAPSN* gene have been found to cause congenital myasthenic syndrome. Most of these mutations change single protein building blocks (amino acids) in the rapsyn protein. A common mutation replaces the amino acid asparagine with the amino acid lysine at position 88 in the rapsyn protein (written as Asn88Lys or N88K). Most mutations in the *RAPSN* gene result in a reduction in functional rapsyn protein. The lack of rapsyn protein results in decreased binding between rapsyn and the AChR protein, which leads to disorganization of the receptor protein in the muscle cell membrane and a reduction in the number of receptors. As a result, signaling at the neuromuscular junction is decreased, which leads to decreased muscle movement and the muscle weakness characteristic of congenital myasthenic syndrome.

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])

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])

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Genomic Location

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

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