REEP1 gene

receptor accessory protein 1

Normal Function

The REEP1 gene provides instructions for making a protein called receptor expression-enhancing protein 1 (REEP1), which is found in nerve cells (neurons) in the brain and spinal cord. The REEP1 protein is located within cell compartments called mitochondria, which are the energy-producing centers in cells, and the endoplasmic reticulum, which helps with protein processing and transport.

The REEP1 protein plays a role in forming the network of tubules that make up the structure of the endoplasmic reticulum, regulating its size and determining how many proteins it can process. As part of its role in the endoplasmic reticulum, the REEP1 protein enhances the activity of certain other proteins called G protein-coupled receptors. These receptor proteins are eventually embedded within the outer membrane of cells, where they relay chemical signals from outside the cell to the interior of the cell.

The function of the REEP1 protein in the mitochondria is unknown.

Health Conditions Related to Genetic Changes

Distal hereditary motor neuropathy, type V

MedlinePlus Genetics provides information about Distal hereditary motor neuropathy, type V

Spastic paraplegia type 31

At least 44 mutations in the *REEP1* gene have been found to cause spastic paraplegia type 31. This condition is characterized by muscle stiffness (spasticity) and paralysis of the lower limbs (paraplegia) caused by degeneration of nerve cells (neurons) that trigger muscle movement. Most of the *REEP1* gene mutations that cause this condition insert or remove small pieces of DNA from the gene or alter the way the gene's instructions are used to make the protein. These mutations often result in a short, nonfunctional protein that is quickly broken down. As a result, there is a reduction in functional REEP1 protein.

It is unclear how *REEP1* gene mutations lead to the signs and symptoms of spastic paraplegia type 31. Researchers have shown that mitochondria in cells of affected

individuals are less able to produce energy, which may contribute to the death of neurons and lead to the progressive movement problems of spastic paraplegia type 31; however, the exact mechanism that causes this condition is unknown.

Other Names for This Gene

- C2orf23
- FLJ13110
- receptor expression-enhancing protein 1
- REEP1_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of REEP1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=65055[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28REEP1%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- RECEPTOR EXPRESSION-ENHANCING PROTEIN 1; REEP1 (https://omim.org/entry/609139)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/65055)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=REEP1[gene])

References

- Beetz C, Pieber TR, Hertel N, Schabhuttl M, Fischer C, Trajanoski S, Graf E,Keiner S, Kurth I, Wieland T, Varga RE, Timmerman V, Reilly MM, Strom TM,Auer-Grumbach M. Exome sequencing identifies a REEP1 mutation involved in distalhereditary motor neuropathy type V. Am J Hum Genet. 2012 Jul 13;91(1):139-45.doi: 10.1016/j.ajhg.2012.05.007. Epub 2012 Jun 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22703882) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3397265/)

- Bjork S, Hurt CM, Ho VK, Angelotti T. REEPs are membrane shaping adapterproteins that modulate specific g protein-coupled receptor trafficking byaffecting ER cargo capacity. PLoS One. 2013 Oct 2;8(10):e76366. doi:10.1371/journal.pone.0076366. eCollection 2013. Erratum In: PLoS One. 2013;8(12).doi:10.1371/annotation/6f86410c-63c3-4fcd-b1cb-9fd8d2ea95d0. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24098485) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3788743/)
- Goizet C, Depienne C, Benard G, Boukhris A, Mundwiller E, Sole G, Coupry I,Pilliod J, Martin-Negrier ML, Fedirko E, Forlani S, Cazeneuve C, Hannequin D,Charles P, Feki I, Pinel JF, Ouvrard-Hernandez AM, Lyonnet S, Ollagnon-Roman E,Yaouanq J, Toutain A, Dussert C, Fontaine B, Leguern E, Lacombe D, Durr A,Rossignol R, Brice A, Stevanin G. REEP1 mutations in SPG31: frequency, mutationalspectrum, and potential association with mitochondrial morpho-functionaldysfunction. Hum Mutat. 2011 Oct;32(10):1118-27. doi: 10.1002/humu.21542. Epub2011 Sep 9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21618648)
- Hurt CM, Bjork S, Ho VK, Gilsbach R, Hein L, Angelotti T. REEP1 and REEP2proteins are preferentially expressed in neuronal and neuronal-like exocytotictissues. Brain Res. 2014 Jan 30;1545:12-22. doi: 10.1016/j.brainres.2013.12.008.Epub 2013 Dec 16. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24355597) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3919455/)
- Schlang KJ, Arning L, Epplen JT, Stemmler S. Autosomal dominant hereditaryspastic paraplegia: novel mutations in the REEP1 gene (SPG31). BMC Med Genet.2008 Jul 21;9:71. doi: 10.1186/1471-2350-9-71. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18644145) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2492855/)
- Zuchner S, Wang G, Tran-Viet KN, Nance MA, Gaskell PC, Vance JM, Ashley-KochAE, Pericak-Vance MA. Mutations in the novel mitochondrial protein REEP1 causehereditary spastic paraplegia type 31. Am J Hum Genet. 2006 Aug;79(2):365-9. doi:10.1086/505361. Epub 2006 May 26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16826527) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1559498/)

Genomic Location

The *REEP1* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

Last updated April 1, 2015