PRICKLE1 gene

prickle planar cell polarity protein 1

Normal Function

The *PRICKLE1* gene provides instructions for making a protein called prickle homolog 1. The function of this protein is unclear, although it appears to play an important role in the development of the nervous system. Prickle homolog 1 is likely part of a chemical signaling pathway known as noncanonical Wnt signaling. During development before birth, noncanonical Wnt signaling helps to determine the position of various components within cells (cell polarity). This pathway also regulates the movement of nerve cells (neurons) in the developing nervous system.

Studies suggest that prickle homolog 1 interacts with other proteins, including RE1-silencing transcription factor (REST). The REST protein regulates several critical genes in neurons by turning off (suppressing) their activity. To regulate these genes, REST must enter the nucleus and attach (bind) to particular regions of DNA. Researchers believe that prickle homolog 1 controls REST by transporting it out of the nucleus, which prevents it from binding to DNA and suppressing gene activity. It remains unclear how the interaction between prickle homolog 1 and REST contributes to the normal development of the nervous system.

Health Conditions Related to Genetic Changes

PRICKLE1-related progressive myoclonus epilepsy with ataxia

At least three mutations in the *PRICKLE1* gene have been identified in people with *PRICKLE1*-related progressive myoclonus epilepsy with ataxia. Each mutation changes a single protein building block (amino acid) in the prickle homolog 1 protein. One of the known mutations appears to disrupt the interaction between prickle homolog 1 and REST, blocking the transport of REST out of the nucleus. As a result, REST may inappropriately suppress certain genes in the developing nervous system. It is unclear how mutations in the *PRICKLE1* gene lead to movement problems, seizures, and the other features of *PRICKLE1*-related progressive myoclonus epilepsy with ataxia.

Other Names for This Gene

- EPM1B
- FLJ31627

- FLJ31937
- MGC138902
- MGC138903
- PRIC1_HUMAN
- prickle homolog 1
- prickle-like 1
- REST (RE-1 silencing transcription factor)/NRSF (neuron-restrictive silencer factor)-interacting LIM domain protein
- REST/NRSF-interacting LIM domain protein
- RILP

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PRICKLE1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=144165[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PRICKLE1%5BTIAB%5D%29+OR+%28RILP%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

- PRICKLE PLANAR CELL POLARITY PROTEIN 1; PRICKLE1 (https://omim.org/entry/608500)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/144165)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PRICKLE1[gene])

References

- Bassuk AG, Wallace RH, Buhr A, Buller AR, Afawi Z, Shimojo M, Miyata S, ChenS, Gonzalez-Alegre P, Griesbach HL, Wu S, Nashelsky M, Vladar EK, Antic D, Ferguson PJ, Cirak S, Voit T, Scott MP, Axelrod JD, Gurnett C, Daoud AS, KivityS, Neufeld MY, Mazarib A, Straussberg R, Walid S, Korczyn AD, Slusarski DC,

Berkovic SF, El-Shanti HI. A homozygous mutation in human PRICKLE1 causes anautosomal-recessive progressive myoclonus epilepsy-ataxia syndrome. Am J HumGenet. 2008 Nov;83(5):572-81. doi: 10.1016/j.ajhg.2008.10.003. Epub 2008 Oct 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18976727) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668041/)

- Katoh M, Katoh M. Identification and characterization of human PRICKLE1 andPRICKLE2 genes as well as mouse Prickle1 and Prickle2 genes homologous toDrosophila tissue polarity gene prickle. Int J Mol Med. 2003 Feb;11(2):249-56. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12525887)
- Shimojo M, Hersh LB. Characterization of the REST/NRSF-interacting LIM domainprotein (RILP): localization and interaction with REST/NRSF. J Neurochem. 2006Feb;96(4):1130-8. doi: 10.1111/j.1471-4159.2005.03608.x. Epub 2006 Jan 17. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16417580)
- Shimojo M, Hersh LB. REST/NRSF-interacting LIM domain protein, a putativenuclear translocation receptor. Mol Cell Biol. 2003 Dec;23(24):9025-31. doi: 10.1128/MCB.23.24.9025-9031.2003. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14645515) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC309669/)
- Tao H, Manak JR, Sowers L, Mei X, Kiyonari H, Abe T, Dahdaleh NS, Yang T, WuS, Chen S, Fox MH, Gurnett C, Montine T, Bird T, Shaffer LG, Rosenfeld JA, McConnell J, Madan-Khetarpal S, Berry-Kravis E, Griesbach H, Saneto RP, Scott MP,Antic D, Reed J, Boland R, Ehaideb SN, El-Shanti H, Mahajan VB, Ferguson PJ, Axelrod JD, Lehesjoki AE, Fritzsch B, Slusarski DC, Wemmie J, Ueno N, Bassuk AG. Mutations in prickle orthologs cause seizures in flies, mice, and humans. Am JHum Genet. 2011 Feb 11;88(2):138-49. doi: 10.1016/j.ajhg.2010.12.012. Epub 2011Feb 3. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21276947) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3035715/)

Genomic Location

The *PRICKLE1* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

Last updated December 1, 2011