PNKD gene

PNKD metallo-beta-lactamase domain containing

Normal Function

Researchers have not determined the role of the *PNKD* gene (frequently called the *MR1* gene) in the human body. This gene is highly active (expressed) in the brain, which suggests that it plays an important role in normal brain function. The protein produced from the *PNKD* gene may help control the release of chemicals called neurotransmitters, which allow nerve cells (neurons) to communicate with one another.

The PNKD protein is similar to another protein that helps break down a chemical called methylglyoxal. Methylglyoxal is found in alcoholic beverages, coffee, tea, and cola. Research has demonstrated that this chemical is toxic to nerve cells (neurons). The PNKD protein may perform a function similar to this protein.

Health Conditions Related to Genetic Changes

Familial paroxysmal nonkinesigenic dyskinesia

At least three mutations in the *PNKD* gene have been shown to cause familial paroxysmal nonkinesigenic dyskinesia, which is characterized by episodes of involuntary movement. The two most common mutations, each found in several affected families, replace the protein building block (amino acid) alanine with the amino acid valine in the PNKD protein. One of the mutations occurs at position 7 (written as Ala7Val or A7V), and the other mutation is at position 9 (written as Ala9Val or A9V). Research suggests that the *PNKD* gene mutations alter the structure of the PNKD protein and interfere with its ability to function. It is not known how mutations in the *PNKD* gene lead to the signs and symptoms of familial paroxysmal nonkinesigenic dyskinesia.

Other Names for This Gene

- brain protein 17
- BRP17
- DKFZp564N1362
- DYT8
- FKSG19

- FPD1
- KIAA1184
- KIPP1184
- MGC31943
- MR-1
- MR-1S
- MR1
- myofibrillogenesis regulator 1
- paroxysmal nonkinesigenic dyskinesia
- PDC
- PKND1
- PNKD_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PNKD (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=25953[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PNKD%5BTIAB%5D%29+OR+%28paroxysmal+nonkinesigenic+dyskinesia%5BTIAB%5D%29%29+OR+%28%28MR-1%5BTIAB%5D%29+OR+%28MR1%5BTIAB%5D%29+OR+%28myofibrillogenesis+regulator+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28histocompatibility%5BTIAB%5D%29+OR+%28Doxepin%5BTIAB%5D%29+OR+%28MHC%5BTIAB%5D%29+OR+%28HLA%5BTIAB%5D%29+OR+%28immunotoxin%5BTIAB%5D%29+OR+%28mecA%5BTIAB%5D%29+OR+%28autoimmune%5BTIAB%5D%29+OR+%28HeLa%29+OR+%28Immunomodulation%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PNKD METALLO-BETA-LACTAMASE DOMAIN-CONTAINING PROTEIN; PNKD (https://omim.org/entry/609023)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/25953)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PNKD[gene])

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Genomic Location

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

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