PANK2 gene
pantothenate kinase 2

Normal Function

The *PANK2* gene provides instructions for making an enzyme called pantothenate kinase 2. This enzyme is active in specialized cellular structures called mitochondria, which are the cell’s energy-producing centers. Within mitochondria, pantothenate kinase 2 regulates the formation of a molecule called coenzyme A. Coenzyme A is found in all living cells, where it is essential for the body’s production of energy from carbohydrates, fats, and some protein building blocks (amino acids).

*PANK2* is one of four human genes that provide instructions for making versions of pantothenate kinase. The functions of these different versions probably vary among tissue types and parts of the cell. The version produced by the *PANK2* gene is active in cells throughout the body, including nerve cells in the brain.

Health Conditions Related to Genetic Changes

**Pantothenate kinase-associated neurodegeneration**

About 100 mutations in the *PANK2* gene have been identified in people with pantothenate kinase-associated neurodegeneration. Typically, people with the more severe, early-onset form of the disorder have *PANK2* mutations that prevent cells from producing any functional pantothenate kinase 2. People affected by the atypical, later-onset form usually have mutations that change single amino acids in the enzyme, which makes the enzyme unstable or disrupts its activity. In some cases, single amino acid changes allow the enzyme to retain some function. The most common *PANK2* mutation replaces the amino acid glycine with the amino acid arginine at position 411 of the enzyme (written as Gly411Arg or G411R).

When pantothenate kinase 2 is altered or missing, the normal production of coenzyme A is disrupted and potentially harmful compounds can build up in the brain. This buildup leads to swelling, tissue damage, and an abnormal accumulation of iron in certain areas of the brain. Researchers are uncertain how a lack of functional pantothenate kinase 2 causes the specific features of pantothenate kinase-associated neurodegeneration. Because the enzyme functions in mitochondria, the signs and symptoms of this condition may be related to impaired energy production.

Mutations in the *PANK2* gene are also found in people with a condition called HARP (hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration). HARP was historically described as a separate syndrome but is now considered part of pantothenate kinase-associated neurodegeneration. Although
HARP is much rarer than classic pantothenate kinase-associated neurodegeneration, both conditions involve problems with movement, dementia, and vision abnormalities.

Chromosomal Location

Cytogenetic Location: 20p13, which is the short (p) arm of chromosome 20 at position 13

Molecular Location: base pairs 3,888,823 to 3,933,087 on chromosome 20 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

[Diagram of chromosome 20 with p13 highlighted]

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- NBIA1
- PANK2_HUMAN
- pantothenate kinase 2 (Hallervorden-Spatz syndrome)
- pantothenic acid kinase

Additional Information & Resources

Clinical Information from GeneReviews
- Pantothenate Kinase-Associated Neurodegeneration
  https://www.ncbi.nlm.nih.gov/books/NBK1490

Scientific Articles on PubMed
- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PANK2%5BTIAB%5D%29+OR+%28pantothenate+kinase+2%5BTIAB%5D%29+OR+%28NBIA1%5BTIAB%5D+OR+%28pantothenic+acid+kinase%5BTIAB%5D%29+AND+%28Genes%5BMH%5D+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+OR+%22last+3600+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- **PANTOTHENATE KINASE 2**
  http://omim.org/entry/606157

Research Resources

- **ClinVar**

- **HGNC Gene Symbol Report**

- **Monarch Initiative**
  https://monarchinitiative.org/gene/NCBIGene:80025

- **NCBI Gene**

- **UniProt**
  https://www.uniprot.org/uniprot/Q9BZ23

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12374576

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16416393  
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2117327/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16437574

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12510040

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12614941

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14631201
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15105273

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15659606

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15105272

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16272150


Reviewed: October 2006
Published: September 11, 2018

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services