

## PINK1 gene

PTEN induced kinase 1

### Normal Function

The *PINK1* gene provides instructions for making a protein called PTEN induced putative kinase 1. This protein is found in cells throughout the body, with highest levels in the heart, muscles, and testes. Within cells, the protein is located in the mitochondria, the energy-producing centers that provide power for cellular activities. The function of PTEN induced putative kinase 1 is not fully understood. It appears to help protect mitochondria from malfunctioning during periods of cellular stress, such as unusually high energy demands.

Researchers believe that two specialized regions of PTEN induced putative kinase 1 are essential for the protein to function properly. One region, called the mitochondrial-targeting motif, serves as a delivery address: after the protein is made, this motif helps ensure that it is delivered to the mitochondria. Another region, called the kinase domain, probably carries out the protein's protective function.

### Health Conditions Related to Genetic Changes

#### Parkinson's disease

Researchers have identified more than 70 mutations in the *PINK1* gene that can cause Parkinson's disease, a condition characterized by progressive problems with movement and balance. *PINK1* gene mutations are associated with the early-onset form of the disorder, which typically begins before age 50.

Many *PINK1* gene mutations alter or eliminate the kinase domain, leading to a loss of protein function. At least one mutation affects the mitochondrial-targeting motif and may disrupt delivery of the protein to mitochondria. With reduced or absent PTEN induced putative kinase 1 activity, mitochondria may malfunction, particularly when cells are stressed. Cells can die if energy is not provided for essential activities. It is unclear how *PINK1* gene mutations cause the selective death of nerve cells that characterizes Parkinson's disease. The loss of these cells weakens communication between the brain and muscles, and ultimately the brain becomes unable to control muscle movement.

## Other Names for This Gene

- BRPK
- PARK6
- PINK1\_HUMAN
- PTEN induced putative kinase 1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of PINK1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=65018\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=65018[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PINK1%5BTIAB%5D%29+OR+%28PTEN+induced+putative+kinase+1%5BTIAB%5D%29%29+OR+%28%28Park6%5BTIAB%5D%29+OR+%28BRPK%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+720+days%22%5Dp%5D>)

### Catalog of Genes and Diseases from OMIM

- PTEN-INDUCED KINASE 1; PINK1 (<https://omim.org/entry/608309>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/65018>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PINK1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PINK1[gene]))

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## Genomic Location

The *PINK1* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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