PGK1 gene

phosphoglycerate kinase 1

Normal Function

The *PGK1* gene provides instructions for making an enzyme called phosphoglycerate kinase. This enzyme is found in cells and tissues throughout the body, where it is involved in a critical energy-producing process known as glycolysis. During glycolysis, the simple sugar glucose is broken down to produce energy.

Phosphoglycerate kinase helps carry out a chemical reaction that converts a molecule called 1,3-diphosphoglycerate, which is produced during the breakdown of glucose, to another molecule called 3-phosphoglycerate. This reaction generates one molecule of adenosine triphosphate (ATP), which is the main energy source in cells.

Researchers suspect that phosphoglycerate kinase may have additional functions, although little is known about the other roles this enzyme may play in cells.

Health Conditions Related to Genetic Changes

Phosphoglycerate kinase deficiency

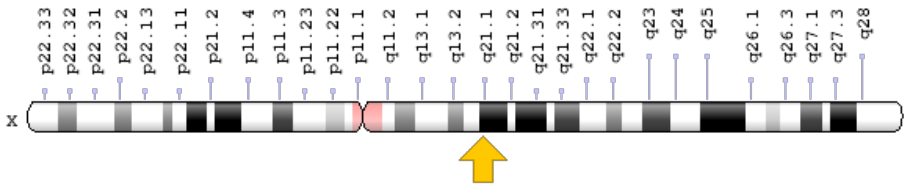
At least 18 mutations in the *PGK1* gene have been found to cause phosphoglycerate kinase deficiency. In some affected individuals, this condition causes a shortage of red blood cells (chronic hemolytic anemia) with or without neurological symptoms. In others, the condition is characterized by muscle weakness and cramping.

Most *PGK1* gene mutations change single protein building blocks (amino acids) in phosphoglycerate kinase. A few other types of mutations, including insertions and deletions of a small amount of DNA in the *PGK1* gene, have also been reported. Studies suggest that *PGK1* gene mutations reduce the activity of phosphoglycerate kinase, which disrupts normal energy production and leads to cell damage or cell death. It is unclear why this abnormality preferentially affects red blood cells and brain cells in some people and muscle cells in others. Researchers speculate that different *PGK1* gene mutations may have varying effects on the activity of phosphoglycerate kinase in different types of cells.

Chromosomal Location

Cytogenetic Location: Xq21.1, which is the long (q) arm of the X chromosome at position 21.1

Molecular Location: base pairs 78,104,248 to 78,129,295 on the X chromosome (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cell migration-inducing gene 10 protein
- MGC8947
- MGC117307
- MGC142128
- MIG10
- PGK1_HUMAN
- PGKA
- primer recognition protein 2
- PRP 2

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Glycolysis is an energy-conversion pathway in many organisms
  https://www.ncbi.nlm.nih.gov/books/NBK22593/
- Neuroproteomics (second edition, 2010): Phosphoglycerate kinase enzymatic reaction (figure)
  https://www.ncbi.nlm.nih.gov/books/NBK56020/figure/ch10_f11/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PGK1%5BTI%5D%29+OR+%28phosphoglycerate+kinase%5BTI%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- PHOSPHOGLYCERATE KINASE 1
  http://omim.org/entry/311800

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PGK1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PGK1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:8896
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:5230
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5230
- UniProt
  https://www.uniprot.org/uniprot/P00558

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