DGUOK gene

deoxyguanosine kinase

Normal Function

The DGUOK gene provides instructions for making the enzyme deoxyguanosine kinase. This enzyme plays a critical role in mitochondria, which are structures within cells that convert the energy from food into a form that cells can use. Mitochondria each contain a small amount of DNA, known as mitochondrial DNA or mtDNA, which is essential for the normal function of these structures. Deoxyguanosine kinase is involved in producing and maintaining the building blocks of mitochondrial DNA.

Health Conditions Related to Genetic Changes

Deoxyguanosine kinase deficiency

Approximately 40 mutations in the DGUOK gene have been identified in people with deoxyguanosine kinase deficiency. Some of these mutations change single protein building blocks (amino acids) in the deoxyguanosine kinase enzyme. Other mutations result in an abnormally shortened, nonfunctional enzyme or cause the enzyme to be pieced together incorrectly.

Mutations in the DGUOK gene reduce or eliminate the activity of the deoxyguanosine kinase enzyme. Reduced enzyme activity leads to problems with the production and maintenance of mitochondrial DNA. A reduction in the amount of mitochondrial DNA (known as mitochondrial DNA depletion) impairs mitochondrial function in many of the body’s cells and tissues. These problems lead to the neurological and liver dysfunction associated with deoxyguanosine kinase deficiency.
**Chromosomal Location**

Cytogenetic Location: 2p13.1, which is the short (p) arm of chromosome 2 at position 13.1

Molecular Location: base pairs 73,926,827 to 73,958,961 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

**Other Names for This Gene**

- deoxyguanosine kinase isoform a precursor
- deoxyguanosine kinase isoform b precursor
- deoxyguanosine kinase, mitochondrial
- dGK
- DGUOK_HUMAN

**Additional Information & Resources**

**Educational Resources**

- Washington University (St. Louis)
  https://neuromuscular.wustl.edu/mitosyn.html#hepcerdep

**Clinical Information from GeneReviews**

- Deoxyguanosine Kinase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK7040

- Mitochondrial DNA Maintenance Defects Overview
  https://www.ncbi.nlm.nih.gov/books/NBK487393

**Scientific Articles on PubMed**

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DGUOK%5BTIAB%5D%29+OR+%28deoxyguanosine+kinase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM
• DEOXYGUANOSINE KINASE
  http://omim.org/entry/601465

Research Resources
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DGUOK%5Bgene%5D
• HGNC Gene Symbol Report
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1716
• NCBI Gene
• UniProt
  https://www.uniprot.org/uniprot/Q16854

Sources for This Summary
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19394258
• OMIM: DEOXYGUANOSINE KINASE
  http://omim.org/entry/601465
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18205204
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301766
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14623087

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