

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.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DGUOK ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1716\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1716[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DGUOK%5BTIAB%5D%29+OR+%28deoxyguanosine+kinase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+engli sh%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- DEOXYGUANOSINE KINASE; DGUOK (<https://omim.org/entry/601465>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1716>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DGUOK\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DGUOK[gene]))

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Genomic Location

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

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