# COQ2 gene

coenzyme Q2, polyprenyltransferase

## Normal Function

The *COQ2* gene provides instructions for making an enzyme that carries out one step in the production of a molecule called coenzyme Q10, which has several critical functions in cells throughout the body. In cell structures called mitochondria, coenzyme Q10 plays an essential role in a process called oxidative phosphorylation, which converts the energy from food into a form cells can use. Coenzyme Q10 is also involved in producing pyrimidines, which are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell. In cell membranes, coenzyme Q10 acts as an antioxidant, protecting cells from damage caused by unstable oxygen-containing molecules (free radicals), which are byproducts of energy production.

## Health Conditions Related to Genetic Changes

### Primary coenzyme Q10 deficiency

At least nine mutations in the *COQ2* gene have been found to cause a disorder known as primary coenzyme Q10 deficiency. This rare disease usually becomes apparent in infancy or early childhood, but it can occur at any age. It can affect many parts of the body, most often the brain, muscles, and kidneys. The *COQ2* gene mutations associated with this disorder greatly reduce or eliminate the production of the COQ2 enzyme, which prevents the normal production of coenzyme Q10. Studies suggest that a shortage (deficiency) of coenzyme Q10 impairs oxidative phosphorylation and increases the vulnerability of cells to damage from free radicals. A deficiency of coenzyme Q10 may also disrupt the production of pyrimidines. These changes can cause cells throughout the body to malfunction, which may help explain the variety of organs and tissues that can be affected by primary coenzyme Q10 deficiency.

### Multiple system atrophy

Several variations in the *COQ2* gene have been suggested to increase the risk of multiple system atrophy, a progressive brain disorder that affects movement and balance and disrupts the function of the autonomic nervous system. The autonomic nervous system controls body functions that are mostly involuntary, such as regulation of blood pressure.

The identified variations alter single protein building blocks (amino acids) in the COQ2 enzyme. Most of the variations are very rare, but a genetic change that replaces the amino acid valine with the amino acid alanine at position 393 (written

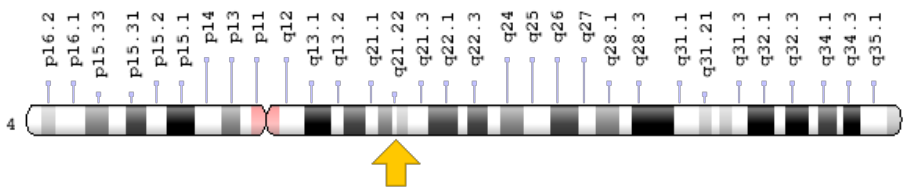
as Val393Ala or V393A) is relatively common. Studies suggest that these variations, including V393A, are associated with an increased risk of developing multiple system atrophy in the Japanese population. However, studies have not found a correlation between *COQ2* gene variations and multiple system atrophy in other populations, including Koreans, Europeans, and North Americans. It remains unclear whether *COQ2* gene variations represent a significant risk factor for this disease.

Researchers speculate that changes in the *COQ2* gene could impair the activity of the COQ2 enzyme, which would affect the production of coenzyme Q10. Levels of coenzyme Q10 are reduced in the brains of people with multiple system atrophy. However, the *COQ2* gene variations associated with an increased risk of this disorder are thought to affect coenzyme Q10 levels less severely than the *COQ2* gene mutations that cause primary coenzyme Q10 deficiency (described above). A reduction in the amount of coenzyme Q10 may impair cellular energy production from oxidative phosphorylation and increase the vulnerability of cells to damage from free radicals. However, it is unknown how these changes are related to the specific features of multiple system atrophy.

## Chromosomal Location

Cytogenetic Location: 4q21.22-q21.23, which is the long (q) arm of chromosome 4 between positions 21.22 and 21.23

Molecular Location: base pairs 83,263,824 to 83,285,134 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 4-HB polyprenyltransferase
- 4-hydroxybenzoate decaprenyltransferase
- 4-hydroxybenzoate polyprenyltransferase, mitochondrial
- CL640
- coenzyme Q2 4-hydroxybenzoate polyprenyltransferase
- coenzyme Q2 homolog, prenyltransferase

- COQ10D1
- FLJ26072
- MSA1
- para-hydroxybenzoate-polyprenyltransferase, mitochondrial
- PHB:polyprenyltransferase
- PHB:PPT

## Additional Information & Resources

Educational Resources

- Linus Pauling Institute, Oregon State University: Coenzyme Q10
  https://lpi.oregonstate.edu/mic/dietary-factors/coenzyme-Q10
- Molecular Biology of the Cell (fourth edition, 2002): How Cells Obtain Energy from Food
  https://www.ncbi.nlm.nih.gov/books/NBK26882/
- The Cell: A Molecular Approach (second edition, 2000): The Mechanism of Oxidative Phosphorylation
  https://www.ncbi.nlm.nih.gov/books/NBK9885/

Clinical Information from GeneReviews

- Primary Coenzyme Q10 Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK410087

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28COQ2%5BTIAB%5D%29+OR+%28coenzyme+Q2%5BTIAB%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

- COENZYME Q2, POLYPRENYLTRANSFERASE
  http://omim.org/entry/609825

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_COQ2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=COQ2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:25223

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:27235
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/27235

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