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

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 below). 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.

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.

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

Tests Listed in the Genetic Testing Registry

- Tests of COQ2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=27235[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; COQ2 (https://omim.org/entry/609825)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/27235)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=COQ2[gene])

References

- Acosta MJ, Vazquez Fonseca L, Desbats MA, Cerqua C, Zordan R, Trevisson E, Salviati L. Coenzyme Q biosynthesis in health and disease. Biochim Biophys Acta. 2016 Aug;1857(8):1079-1085. doi: 10.1016/j.bbabio.2016.03.036. Epub 2016 Apr 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/27060254)
- Desbats MA, Lunardi G, Doimo M, Trevisson E, Salviati L. Genetic bases andclinical manifestations of coenzyme Q10 (CoQ 10) deficiency. J Inherit Metab Dis.2015 Jan;38(1):145-56. doi: 10.1007/s10545-014-9749-9. Epub 2014 Aug 5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25091424)
- Doimo M, Desbats MA, Cerqua C, Cassina M, Trevisson E, Salviati L. Genetics ofcoenzyme q10 deficiency. Mol Syndromol. 2014 Jul;5(3-4):156-62. doi:10.1159/000362826. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25126048) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4112527/)
- Mitsui J, Tsuji S. Mutant COQ2 in multiple-system atrophy. N Engl J Med. 2014Jul 3;371(1):82-3. doi: 10.1056/NEJMc1311763. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24988566)
- Mollet J, Giurgea I, Schlemmer D, Dallner G, Chretien D, Delahodde A, Bacq D,de Lonlay P, Munnich A, Rotig A. Prenyldiphosphate synthase, subunit 1 (PDSS1)and OH-benzoate polyprenyltransferase (COQ2) mutations in ubiquinone deficiencyand oxidative phosphorylation disorders. J Clin Invest. 2007 Mar;117(3):765-72.doi: 10.1172/JCI29089. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17332895) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1804361/)
- Multiple-System Atrophy Research Collaboration. Mutations in COQ2 in familialand sporadic multiple-system atrophy. N Engl J Med. 2013 Jul 18;369(3):233-44.doi: 10.1056/NEJMoa1212115. Epub 2013 Jun 12. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23758206)

- Quinzii C, Naini A, Salviati L, Trevisson E, Navas P, Dimauro S, Hirano M. Amutation in para-hydroxybenzoate-polyprenyl transferase (COQ2) causes primarycoenzyme Q10 deficiency. Am J Hum Genet. 2006 Feb;78(2):345-9. doi:10.1086/500092. Epub 2005 Dec 22. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16400613) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1380241/)
- Salviati L, Trevisson E, Agosto C, Doimo M, Navas P. Primary CoenzymeQ10 Deficiency Overview. 2017 Jan 26 [updated 2023 Jun 8]. In: AdamMP, Bick S, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors.GeneReviews(R) [Internet]. Seattle (WA): University of Washington,Seattle; 1993-2025. Available from http://www.ncbi.nlm.nih.gov/books/NBK410087/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/28125198)
- Schottlaender LV, Bettencourt C, Kiely AP, Chalasani A, Neergheen V, HoltonJL, Hargreaves I, Houlden H. Coenzyme Q10 Levels Are Decreased in the Cerebellumof Multiple-System Atrophy Patients. PLoS One. 2016 Feb 19;11(2):e0149557. doi:10.1371/journal.pone.0149557. eCollection 2016. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/26894433) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4760984/)
- Sun Z, Ohta Y, Yamashita T, Sato K, Takemoto M, Hishikawa N, Abe K. Newsusceptible variant of COQ2 gene in Japanese patients with sporadic multiplesystem atrophy. Neurol Genet. 2016 Mar 3;2(2):e54. doi:10.1212/NXG.0000000000000054. eCollection 2016 Apr. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/27123473) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4830192/)
- Zhao Q, Yang X, Tian S, An R, Zheng J, Xu Y. Association of the COQ2 V393Avariant with risk of multiple system atrophy in East Asians: a case-control studyand meta-analysis of the literature. Neurol Sci. 2016 Mar;37(3):423-30. doi:10.1007/s10072-015-2414-8. Epub 2015 Nov 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/26590992)

Genomic Location

The *COQ2* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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