

COQ4 gene

coenzyme Q4

Normal Function

The *COQ4* gene provides instructions for making a protein that is involved in the production of a molecule called coenzyme Q10, although its specific role in this process is unknown. Research suggests that the COQ4 protein may help organize other proteins involved in coenzyme Q10 production into a stable functional group (a protein complex).

Coenzyme Q10 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 12 mutations in the *COQ4* 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 *COQ4* gene mutations associated with this disorder greatly reduce or eliminate the production of the COQ4 protein, 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

- CGI-92

- coenzyme Q biosynthesis protein 4 homolog
- coenzyme Q4 homolog
- COQ10D7
- ubiquinone biosynthesis protein COQ4 homolog, mitochondrial isoform 1 precursor
- ubiquinone biosynthesis protein COQ4 homolog, mitochondrial isoform 2 precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COQ4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51117\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51117[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28COQ4%5BTIAB%5D%29+OR+%28coenzyme+Q4%5BTIAB%5D%29%29+OR+%28%28coenzyme+Q+biosynthesis+protein+4+homolog%5BTIAB%5D%29+OR+%28coenzyme+Q4+homolog%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+protein+COQ4+homolog,+mitochondrial+isoform+1+precursor%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+protein+COQ4+homolog,+mitochondrial+isoform+2+precursor%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

Catalog of Genes and Diseases from OMIM

- COENZYME Q4; COQ4 (<https://omim.org/entry/612898>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/51117>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COQ4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COQ4[gene]))

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Genomic Location

The COQ4 gene is found on chromosome 9 (<https://medlineplus.gov/genetics/chromosome/9/>).

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