

## COQ8B gene

coenzyme Q8B

### Normal Function

The *COQ8B* gene provides instructions for making a protein that is involved 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 11 mutations in the *COQ8B* 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 *COQ8B* gene mutations associated with this disorder greatly reduce or eliminate the function of the COQ8B 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

- aarF domain containing kinase 4
- aarF domain-containing protein kinase 4
- ADCK4
- atypical kinase COQ8B, mitochondrial isoform a
- atypical kinase COQ8B, mitochondrial isoform b

- coenzyme Q protein 8B
- COQ8
- FLJ12229
- NPHS9

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of COQ8B ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79934\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79934[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28coenzyme+Q8B%5BTIAB%5D%29+OR+%28COQ8B%5BTIAB%5D%29%29+OR+%28%28ADCK4%5BTIAB%5D%29+OR+%28COQ8%5BTIAB%5D%29+OR+%28aarF+domain+containing+kinase+4%5BTIAB%5D%29+OR+%28aarF+domain-containing+protein+kinase+4%5BTIAB%5D%29+OR+%28atypical+kinase+COQ8B,+mitochondrial+isoform+a%5BTIAB%5D%29+OR+%28atypical+kinase+COQ8B,+mitochondrial+isoform+b%5BTIAB%5D%29+OR+%28coenzyme+Q+protein+8B%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>)

### Catalog of Genes and Diseases from OMIM

- COENZYME Q8B; COQ8B (<https://omim.org/entry/615567>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/79934>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=COQ8B\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=COQ8B[gene]))

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## Genomic Location

The *COQ8B* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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