COQ6 gene

coenzyme Q6, monooxygenase

Normal Function

The *COQ6* gene provides instructions for making an enzyme that carries out one step in the production of a molecule called coenzyme Q10. 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 seven mutations in the *COQ6* 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 *COQ6* gene mutations associated with this disorder result in a COQ6 enzyme with an abnormal structure that likely impairs its function, leading to reduced 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-10
- CGI10
- coenzyme Q10 monooxygenase 6

- coenzyme Q6 homolog, monooxygenase
- COQ10D6
- ubiquinone biosynthesis monooxygenase COQ6, mitochondrial isoform a
- ubiquinone biosynthesis monooxygenase COQ6, mitochondrial isoform b

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of COQ6 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51004[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28COQ6%5BTIAB%5D%29+OR+%28coenzyme+Q6,+monooxygenase%5BTIAB%5D%29%29+OR+%28%28coenzyme+Q10+monooxygenase+6%5BTIAB%5D%29+OR+%28coenzyme+Q6+homolog,+monooxygenase%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+monooxygenase+COQ6,+mitochondrial+isoform+a%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+monooxygenase+COQ6,+mitochondrial+isoform+b%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 Q6, MONOOXYGENASE; COQ6 (https://omim.org/entry/614647)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/51004)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=COQ6[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/)

- Heeringa SF, Chernin G, Chaki M, Zhou W, Sloan AJ, Ji Z, Xie LX, Salviati L,Hurd TW, Vega-Warner V, Killen PD, Raphael Y, Ashraf S, Ovunc B, Schoeb DS, McLaughlin HM, Airik R, Vlangos CN, Gbadegesin R, Hinkes B, Saisawat P, TrevissonE, Doimo M, Casarin A, Pertegato V, Giorgi G, Prokisch H, Rotig A, Nurnberg G,Becker C, Wang S, Ozaltin F, Topaloglu R, Bakkaloglu A, Bakkaloglu SA, Muller D,Beissert A, Mir S, Berdeli A, Varpizen S, Zenker M, Matejas V, Santos-Ocana C,Navas P, Kusakabe T, Kispert A, Akman S, Soliman NA, Krick S, Mundel P, Reiser J,Nurnberg P, Clarke CF, Wiggins RC, Faul C, Hildebrandt F. COQ6 mutations in humanpatients produce nephrotic syndrome with sensorineural deafness. J Clin Invest.2011 May;121(5):2013-24. doi: 10.1172/JCI45693. Epub 2011 Apr 11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21540551) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3083770/)
- Salviati L, Trevisson E, Agosto C, Doimo M, Navas P. Primary CoenzymeQ10 Deficiency Overview. 2017 Jan 26 [updated 2023 Jun 8]. In: AdamMP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A,editors. GeneReviews(R) [Internet]. Seattle (WA): University ofWashington, Seattle; 1993-2024. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK410087/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/28125198)

Genomic Location

The *COQ6* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

Last updated April 1, 2017