HCCS gene

holocytochrome c synthase

Normal Function

The *HCCS* gene carries instructions for producing an enzyme called holocytochrome c-type synthase. This enzyme is active in many tissues of the body and is found in the mitochondria, the energy-producing centers within cells.

Within the mitochondria, the holocytochrome c-type synthase enzyme helps produce a molecule called cytochrome c. Specifically, holocytochrome c-type synthase is involved in a reaction that adds an iron-containing molecule called heme to make mature cytochrome c, also called holocytochrome c, from a precursor form called apocytochrome c.

Cytochrome c is involved in a process called oxidative phosphorylation, by which mitochondria generate adenosine triphosphate (ATP), the cell's main energy source. It also plays a role in the self-destruction of cells (apoptosis).

Health Conditions Related to Genetic Changes

Microphthalmia with linear skin defects syndrome

At least three *HCCS* gene mutations have been identified in individuals with microphthalmia with linear skin defects syndrome. Deletions of genetic material that include the *HCCS* gene have also been identified in affected individuals. *HCCS* gene mutations result in a holocytochrome c-type synthase enzyme that cannot perform its function. A deletion of genetic material that includes the *HCCS* gene prevents the production of the enzyme from that copy of the gene. This loss of functional holocytochrome c-type synthase enzyme can damage cells by impairing their ability to generate energy. In addition, without sufficient holocytochrome c-type synthase enzyme, the damaged cells may not be able to undergo apoptosis. These cells may instead die in a process called necrosis that causes inflammation and damages neighboring cells. During early development this spreading cell damage may lead to the eye and skin abnormalities characteristic of microphthalmia with linear skin defects syndrome.

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- CCHL
- CCHL_HUMAN
- DKFZp779I1858
- holocytochrome c synthase (cytochrome c heme-lyase)
- MCOPS7

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of HCCS (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3052[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28HCCS%5BTI%5D%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

- HOLOCYTOCHROME C SYNTHASE; HCCS (https://omim.org/entry/300056)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/3052)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=HCCS[gene])

References

- Prakash SK, Cormier TA, McCall AE, Garcia JJ, Sierra R, Haupt B, Zoghbi HY,Van Den Veyver IB. Loss of holocytochrome c-type synthetase causes the malelethality of X-linked dominant microphthalmia with linear skin defects (MLS)syndrome. Hum Mol Genet. 2002 Dec 1;11(25):3237-48. doi: 10.1093/hmg/11.25.3237. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12444108)
- Wimplinger I, Morleo M, Rosenberger G, Iaconis D, Orth U, Meinecke P, Lerer I, Ballabio A, Gal A, Franco B, Kutsche K. Mutations of the mitochondrialholocytochrome c-type synthase in X-linked dominant microphthalmia with linearskin defects syndrome. Am J Hum Genet. 2006 Nov;79(5):878-89. doi:10.1086/508474. Epub 2006 Sep 6. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17033964) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/

articles/PMC1698567/)

- Wimplinger I, Shaw GM, Kutsche K. HCCS loss-of-function missense mutation in afemale with bilateral microphthalmia and sclerocornea: a novel gene for severeocular malformations? Mol Vis. 2007 Aug 27;13:1475-82. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17893649)

Genomic Location

The *HCCS* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

Last updated October 1, 2009