CP gene

ceruloplasmin

Normal Function

The *CP* gene provides instructions for making a protein called ceruloplasmin. Ceruloplasmin helps move iron from the organs and tissues of the body into the blood. This protein prepares iron for incorporation into a molecule called transferrin, which transports the iron to red blood cells.

There are two forms of ceruloplasmin. One form, serum ceruloplasmin, is made primarily in the liver. It is involved in transporting iron from most of the body, but is unable to enter the brain. The other form of ceruloplasmin, called the glycosylphosphatidylinositol (GPI)-anchored form, is important for processing iron in the brain and releasing it from brain tissue. This form of ceruloplasmin is made in nervous system cells called glia, which protect and maintain nerve cells (neurons).

Health Conditions Related to Genetic Changes

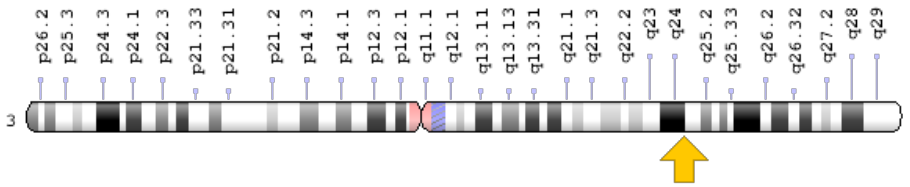
Aceruloplasminemia

Approximately 40 mutations in the *CP* gene that cause aceruloplasminemia have been identified. Some of these mutations substitute one protein building block (amino acid) for another amino acid in the ceruloplasmin protein, resulting in an unstable protein that quickly breaks down (degrades). Other mutations result in the production of an abnormally short, nonfunctional version of the protein or prevent the protein from being secreted by the cells in which it is made. Absence of functional ceruloplasmin results in iron transport problems that lead to the iron accumulation, neurological dysfunction, and other health problems seen in aceruloplasminemia.

Chromosomal Location

Cytogenetic Location: 3q24-q25.1, which is the long (q) arm of chromosome 3 between positions 24 and 25.1

Molecular Location: base pairs 149,162,410 to 149,222,008 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CERU_HUMAN
- ceruloplasmin (ferroxidase)
- CP-2
- ferroxidase

Additional Information & Resources

Clinical Information from GeneReviews

- Aceruloplasminemia
  https://www.ncbi.nlm.nih.gov/books/NBK1493

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CP%5BTIAB%5D%29+OR+%28ceruloplasmin%5BTIAB%5D%29%29+AND+%28%28alpha%282%29-ceruloplasmin%5BMAJR%5D%29+OR+%28ferroxidase%5BMAJR%5D%29+OR+%28ceruloplasmin%5BMAJR%5D%29+OR+%28ceruloplasmin+oxidase%5BMAJR%5D%29+OR+%28ceruloplasmin+ferroxidase%5BMAJR%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%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

- CERULOPLASMIN
  http://omim.org/entry/117700

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CP.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CP%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2295
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1356
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1356
- UniProt
  https://www.uniprot.org/uniprot/P00450

Sources for This Summary

- OMIM: CERULOPLASMIN
  http://omim.org/entry/117700
- Hellman NE, Gitlin JD. Ceruloplasmin metabolism and function. Annu Rev Nutr. 2002;22:439-58. Epub 2002 Apr 4. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12055353
- Kono S, Miyajima H. Molecular and pathological basis of aceruloplasminemia. Biol Res. 2006;39(1): 15-23.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16629161
- Kono S, Suzuki H, Oda T, Miyajima H, Takahashi Y, Shirakawa K, Ishikawa K, Kitagawa M. Biochemical features of ceruloplasmin gene mutations linked to aceruloplasminemia. Neuromolecular Med. 2006;8(3):361-74.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16775387
- Kono S, Suzuki H, Oda T, Shirakawa K, Takahashi Y, Kitagawa M, Miyajima H. Cys-881 is essential for the trafficking and secretion of truncated mutant ceruloplasmin in aceruloplasminemia. J Hepatol. 2007 Dec;47(6):844-50. Epub 2007 Jun 18.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17637479
- Nittis T, Gitlin JD. The copper-iron connection: hereditary aceruloplasminemia. Semin Hematol. 2002 Oct;39(4):282-9. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12382203
- Vassiliev V, Harris ZL, Zatta P. Ceruloplasmin in neurodegenerative diseases. Brain Res Brain Res Rev. 2005 Nov;49(3):633-40. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16269323

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