CPOX gene

coproporphyrinogen oxidase

Normal Function

The *CPOX* gene provides instructions for making an enzyme known as coproporphyrinogen oxidase. This enzyme is involved in the production of a molecule called heme. Heme is vital for all of the body's organs, although it is most abundant in the blood, bone marrow, and liver. Heme is an essential component of iron-containing proteins called hemoproteins, including hemoglobin (the protein that carries oxygen in the blood).

The production of heme is a multi-step process that requires eight different enzymes. Coproporphyrinogen oxidase is responsible for the sixth step in this process, the removal of carbon and oxygen atoms from coproporphyrinogen III (the product of the fifth step) to form protoporphyrinogen IX. In subsequent steps, two other enzymes modify protoporphyrinogen IX and incorporate an iron atom to produce heme.

Health Conditions Related to Genetic Changes

Porphyria

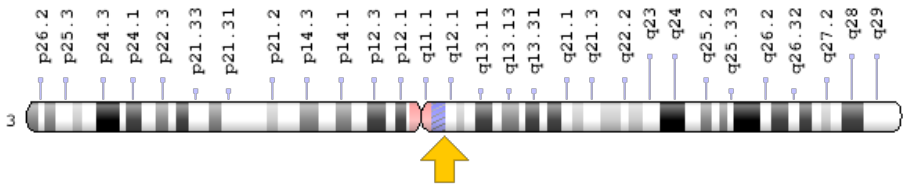
At least 45 mutations in the *CPOX* gene have been found to cause porphyria. Mutations in this gene can cause two types of porphyria: hereditary coproporphyria and a variant known as harderoporphyria.

Most *CPOX* gene mutations change single protein building blocks (amino acids) in coproporphyrinogen oxidase. A single mutation appears to be responsible for harderoporphyria; this genetic change replaces the amino acid glycine with the amino acid glutamic acid at position 404 (written as Lys404Glu or K404E). Mutations in the *CPOX* gene reduce the activity of coproporphyrinogen oxidase, allowing compounds called porphyrins to build up in the body. These compounds are formed during the normal process of heme production, but reduced activity of coproporphyrinogen oxidase allows them to accumulate to toxic levels. This buildup, in combination with nongenetic factors such as certain drugs, alcohol, and dieting, leads to the signs and symptoms of hereditary coproporphyria and harderoporphyria.

Chromosomal Location

Cytogenetic Location: 3q11.2, which is the long (q) arm of chromosome 3 at position 11.2

Molecular Location: base pairs 98,569,837 to 98,593,684 on chromosome 3 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Coprogen oxidase
- Coproporphyrinogen III oxidase, mitochondrial
- Coproporphyrinogen III Oxidases
- coproporphyrinogen oxidase (coproporphyria, harderoporphyria)
- Coproporphyrinogen:oxygen oxidoreductase (decarboxylating)
- Coproporphyrinogenase
- COX
- CPO
- CPX
- HCP
- HEM6_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Mammalian Porphyrins Are Synthesized from Glycine and Succinyl Coenzyme A
  https://www.ncbi.nlm.nih.gov/books/NBK22446/#A3395

Clinical Information from GeneReviews

- Hereditary Coproporphyria
  https://www.ncbi.nlm.nih.gov/books/NBK114807

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CPOX+gene%5BTIAB%5D%29+OR+%28coproporphyrinogen+oxidase%5BTIAB%5D%29+OR+%28Coproporphyrinogen+III+Oxidase%5BTIAB%5D%29+OR+%28Coproporphyrinogen%5BTIAB%5D%29+OR+%28CPO+gene%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- COPROPORPHYRINOGEN OXIDASE
  http://omim.org/entry/612732

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CPOX.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CPOX%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2321
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1371
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1371
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https://ghr.nlm.nih.gov/gene/CPOX

Reviewed: July 2009
Published: June 11, 2019

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